تحت رعاية

صاحبة السمو الشيخة سبيكة بنت إبراهيم آل خليفة

قرينة صاحب الجلالة ملك مملكة البحرين المفدى

رئيسة المجلس الأعلى للمرأة

المؤتمر الخليجي الأول للوراثة

Under the Patronage of Her Highness
Shaikha Sabeeka bint Ibrahim Al-Khalifa
Wife of His Majesty the King
Chairwoman of the Supreme Council for Women

The First GCC Genetic Conference

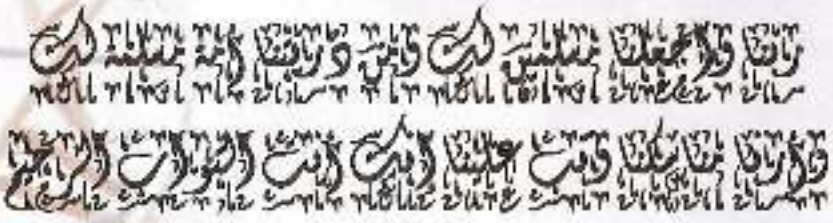
Bahrain 5-7 October 2003

Abstracts and Programme

http://www.gccgenetic.com

Genetic Medicine Benefits for All

The First GCC Genetic Conference - Bahrain 5-7 October 2003 - Abstracts and P



(البقرة: ١٢٨)

Our Lord! make of us Muslims, bowing to they (will), and of our progeny a people Muslim, bowing to thy (will; and show us our places for the celebration of (due) rites; and turn unto us (in mercy); for thou art the oft returning, most merciful. {AlBaqara 128}

صاحب السمو
الشيخ سلمان بن حمد آل خليفة
ولي العهد الأمين
القائد العام لقوة دفاع البحرين

حضرة صاحب الجلالة
الملك حمد بن عيسى آل خليفة
ملك مملكة البحرين المفدى

صاحب السمو
الشيخ خليفة بن سلمان آل خليفة
رئيس الوزراء الموقر

بسم الله الرحمن الرحيم

"وليخش الذين لو تركوا من خلفهم ذرية ضعافا خافوا عليهم فليتقوا الله وليقولوا قولا سديدا" صدق الله العظيم

يطيب لنا رعاية "المؤتمر الخليجي الأول للوراثة" الذي يعقد على أرض مملكة البحرين ليكون باكورة مؤتمرات ولقاءات خليجية متواصلة تهدف إلى تعميق التعاون والتدارس في القضايا والأمور الطبية العامة بمجالاتها المختلفة.

إن احتضان مملكة البحرين لهذه النخبة المتميزة من المتخصصين في علم الوراثة، يؤكد حرص هذا البلد السباق في تسخير كل إمكاناته وطاقاته من أجل مواكبة المستجدات والتطورات العلمية الكبيرة في مجال علم الوراثة التي أصبحت مجتمعاتنا العربية الآن بحاجة ماسة إليها للتعمق في استيعاب قوانين الوراثة ودراستها باعتبارها الوسيلة المثلى لمكافحة الأمراض الوراثية والتخلص منها قدر الإمكان والوقاية منها، وبما يتفق مع التزامات الدول العربية الموقعة على الاتفاقيات الدولية الخاصة بحقوق الطفل لضمان بيئة صحية أفضل لأجيال المستقبل.

فالعوامل الوراثية هي أساس تكوين الإنسان، وهي التي تتحكم في توارث صفاته من جيل إلى جيل سواء كانت هذه الصفات طبيعية أو مرضية، كما بين لنا ديننا الإسلامي الحنيف الأسس العامة للمحافظة على حياتنا وحياة أطفالنا بقوله سبحانه وتعالى (ولا تلقوا بأيديكم إلى التهلكة) فالتحسس الصحيح أثر كبير في حياة الإنسان وسلامة تفكيره، والعافية هي من أفضل ما أنعم الله به علينا.

نأمل أن يكون هذا المؤتمر الأول فرصة طيبة لتحقيق الأهداف المرجوة منه بتعميق وتعزيز التواصل الخليجي المنشود وزيادة الحصيلة المعلوماتية للأطباء المتخصصين في هذا المجال الحيوي وزيادة الوعي الجماهيري بأهمية التدقيق في كل ما يؤثر ويساهم في انتشار الأمراض الوراثية والطرق المناسبة للوقاية من هذه الأمراض.

فأهلا بكل ضيوف البحرين المشاركين في هذا المؤتمر والتي تستقبلهم بكل حفاوة وترحاب، آملين أن يحقق المؤتمر أهدافه المرجوة، متوجين في الوقت ذاته بكل الجهود التي ساهمت في الإعداد والتنظيم وتوفير سبل النجاح لهذا الملتقى الخير.

سبيكة بنت إبراهيم آل خليفة
رئيسة المجلس الأعلى للمرأة

بسم الله الرحمن الرحيم

يسعدني الترحيب بجميع المشاركين في المؤتمر الخليجي الأول للوراثة الذي تستضيفه مملكة البحرين برعاية كريمة من صاحبة السمو الشيخة سبيكة بنت إبراهيم آل خليفة قرينة حضرة صاحب الجلالة الملك حمد بن عيسى آل خليفة ملك البلاد المفدى رئيسة المجلس الأعلى للمرأة متمنيا لكم طيب الإقامة في بلدكم الثاني البحرين، والخروج بنتائج وتوصيات تسهم في محاصرة الأمراض الوراثية وتخفيض أعداد المصابين بها في منطقتنا الخليجية والعربية.

إن مملكة البحرين تولي مشكلة الأمراض الوراثية اهتماما بالغا وتبذل جهودا مكثفة للحد من هذه المشكلة وتخليص أعداد المصابين بها وذلك بالمبادرات الوقائية وأهمها التوعية والتثقيف الصحي، وتطبيق مشروع الفحص قبل الزواج الذي يسهم إلى حد كبير في حالة الالتزام به في تفادي هذه المشكلة وتخليص المجتمع منها.

وهذا فان وزارة الصحة والأجهزة الأخرى في المملكة تعول كثيرا على مناقشات هذا المؤتمر الذي يعقد بتوصية من لجنة خبراء الوراثة بمجلس وزراء الصحة لدول مجلس التعاون الخليجي، بهدف التعرف على المستجدات في مجال الأمراض الوراثية وزيادة الوعي الجماهيري بالطرق المناسبة للوقاية من هذه الأمراض.

ولكون مملكة البحرين تعاني من مشكلة الأمراض الوراثية وخاصة مرض فقر الدم المنجلي فقد قامت وزارة الصحة بالتعاون مع وزارة التربية والتعليم والجمعية البحرينية لأمراض الدم الوراثية بتنفيذ مشروع لفحص الدم لطلاب المدارس، وتحديدا طلاب الصف الثاني الثانوي في مختلف مدارس مملكة البحرين، حيث استمر هذا المشروع منذ عام ١٩٩٩ وحتى الآن، وتم خلاله فحص ٢٤ ألف طالب وطالبة، لرصد معدلات الإصابة بالأمراض الوراثية بين شريحة الطلاب التي تقترب من سن الزواج وتستعد أعداد منها للانخراط في سوق العمل.

إن مملكة البحرين تثق في أهمية التكاتف والتعاون الجماعي للقضاء على أمراض الدم الوراثية من خلال تبادل الخبرات وتنفيذ خطط مشتركة لمكافحة هذه الأمراض التي تكلف دولنا نفقات باهظة، وتتسبب في معاناة الكثير من مواطنيها، فضلا عن تأثير الإصابة بهذه الأمراض على معدلات الإنتاج، وتسببها في حرمان أعداد كبيرة من أبناء مجتمعاتنا من المساهمة في عملية التنمية.

وهذا فان وزارة الصحة تتطلع إلى أن تخرج مؤتمركم بحلول ومقترحات عملية لمواجهة هذه المشكلة والمساعدة في التخلص منها وحماية مجتمعاتنا من آثارها وتداعياتها.. وهو ما سنحرص مملكة البحرين على الاستمرار فيه من خلال تطوير الخدمات الصحية وابتكار أساليب جديدة للتعامل مع هذه الأمراض، خاصة في ظل ما يحظى به القطاع الصحي من دعم حضرة صاحب الجلالة الملك حمد بن عيسى آل خليفة ملك مملكة البحرين المفدى وصاحب السمو الشيخ خليفة بن سلمان آل خليفة رئيس الوزراء الموقر وصاحب السمو الشيخ سلمان بن حمد آل خليفة ولي العهد القائد العام لقوة دفاع البحرين.

وفقكم الله وكلل أعمال مؤتمركم بالنجاح.

والسلام عليكم ورحمة الله وبركاته،،،

الدكتور خليل بن إبراهيم حسن

وزيـــر الصحـــة

رسالة اللجنة المنظمة

باسم اللجنة المنظمة للمؤتمر أرحب بكم أجمل ترحيب في لؤلؤة الخليج البحرين في افتتاح المؤتمر الخليجي الأول للوراثة.

هذا المؤتمر سوف يلقي الضوء على التطورات في مجالات وراثة الأمراض الشائعة. وذلك لزيادة الوعي عن هذه الأمراض وطرق مكافحتها. هذه التطورات الجديدة ساهمت في وضع الطرق للوقاية من الأمراض. تطبيقا للمثل القائل الوقاية خير من العلاج.

كما نعلم فإن الوراثة تدخل في مسببات العديد من الأمراض الشائعة. والمشوهات الخلقية وقد قام مشروع الجين البشري بفتح أبواب جديدة لتشخيص الأمراض وعلاجها والوقاية منها. فوسائل مثل المسوح الوراثية والإرشاد الوراثي أصبحت مهمة للوقاية من الأمراض الوراثية ، وخاصة الأجيال القادمة منها. في هذه المرحلة يجب أن يطلع الأطباء وجميع العاملين في المجال الصحي على هذه الأساليب الجديدة إلى جانب فهم علم الوراثة وتفاعل الجينات مع البيئة ليتمكن الطبيب من إرشاد المرضى بالأمراض مثل السرطان وأمراض القلب والسكري والروماتيزم وغيرها. إن تبني هذا الأسلوب الوقائي قد يمنع ٥٠% من التشوهات الخلقية و ١٠% من الأمراض الوراثية و ٢% من أمراض الكروموسومات وغيرها.

لذا يجب إدخال تدريس مادة الوراثة في جميع مراحل الدراسة خاصة للأطباء وجميع العاملين الصحيين. حيث إن العناية بطفل مصاب بهذه الأمراض تستدعي فريق من العاملين من الكثير من التخصصات.

وقد استضافت البحرين في هذا المؤتمر علماء وأطباء وراثة مشهود لهم بالعلم والخبرة ليثروا المؤتمر بعلمهم وخبرتهم. لذا فان البرنامج العلمي يشمل على ١٢٠ بحث متميز. ٧٥ بحثا سوف يلقى شفهيا أثناء جلسات المؤتمر إلى جانب ٥٠ بحثا سوف يعرض كملصق علمي.

فترة المؤتمر سوف تكون مناسبة ممتازة لنتعرف على بعضنا ولبدأ في تكوين شبكة اتصالات وإقامة مواصلة للأعمال المشتركة. لذا ندعو الراغبين إلى الانضمام لتأسيس جمعية الوراثة الخليجية الأولى.

في اليوم الأخير سوف يكون هناك لقاء مع المرضى في حفل جمعية أمراض الدم الوراثية وجمعية تنمية الطفولة حيث يلتقي المرضى بأطباء الوراثة ليتعرفوا على آخر المستجدات في شئون علاج الأمراض الوراثية والإجابة على استفساراتهم.

أود أن أوجه الشكر الجزيل إلى صاحبة السمو الشيخة سبكة بنت إبراهيم آل خليفة قرينة عاهل البلاد المفدى على رعايتها الكريمة لهذا المؤتمر. فان دل هذا على شيء فانما يدل على عنايتها بالعلم وبحماية أجيال المستقبل من الأطفال من الإمراض الوراثية المستعصية.

كما أوجه الشكر وزير الصحة الدكتور خليل بن إبراهيم حسن على اهتمامه الكبير بمكافحة الأمراض الوراثية وخاصة أمراض الدم الوراثية.

وأشكر كذلك المكتب التنفيذي لمجلس وزراء الصحة لدول مجلس التعاون على ترتيبه انعقاد هذا المؤتمر كما نشكر منظمة الصحة العالمية ممثلة في المدير الإقليمي لمنطقة الشرق الأوسط سعادة الدكتور حسين الجزائري ورئيس الأمراض غير المعدية ونشكر منظمة الصحة العالمية على المساندة والدعم.

كما نشكر جميع من ساند ورعى المؤتمر أخص بالذكر شركة البحرين للاتصالات بملكو الراعي الذهبي للمؤتمر وكذلك شركة الطيران الوطنية شركة طيران الخليج الناقل الرسمي للمؤتمر وجميع الشركات المساهمة.

يسعدني الترحيب بكم مرة أخرى وأتمنى لكم طيب الإقامة الاستفادة الفائقة من المؤتمر.

والسلام عليكم ورحمة الله وبركاته،،،

الدكتورة شيخة سالم العريض
رئيس اللجنة المنظمة للمؤتمر

Message form the Organisation Committee

Dr. Shaikha S. Al Arrayed

On Behalf of the organizing committee, it is my pleasure to welcome you in the kingdom of Bahrain, the pearl of the Gulf, at the beginning of the 1st GCC genetic conference to be held in the Gulf.

What use to be scientific fantasy has became reality, and what use to be fiction has become fact in genetic progress in the last 20 years. Recently the Human Genome Project has revealed new dimensions for diagnosis, treatment and prevention of genetic disorders. These new developments have contributed significantly in controlling genetic diseases. Measures such as screening and counseling are becoming essential tools of both public health and individual medical care.

Genetic factors contribute to almost every common disease, which afflict the human being. Genetic diseases include Single Gene, Chromosomal, Multi-factorial and Mitochondrial disorders. Because of these, considerable number of births is affected with an associated disability.

Physicians need to understand genes and their interactions, in order to manage and counsel patients with common disorders such as Cancer, Cardiovascular disease, Hypertension, Asthma, Diabetes, Rheumatoid Arthritis, Deafness, Glaucoma, and many other diseases.

Therefore, integration of the new genetic knowledge into routine practice is mandatory. These new ways of diagnosing and managing illness need to be incorporated into medical education for future physicians, and health care personnel. A multidisciplinary team approach including physicians,' nurses, social workers, and families of the affected persons is required to manage patients with genetic disorder.

If we implement the new genetic approach, we would expect to reduce the disability and mortality caused by genetic diseases. We would be able to eliminate 50% of congenital abnormalities, ten percent of inherited diseases and 2% of chromosomal disorders, which can be treated or prevented.

This conference will highlight some of the advances made in the field of genetics of common genetic diseases. Our aim is to enrich the knowledge of physicians and other health professionals in this field, to enhance awareness of genetic diseases and methods of prevention, and highlight some of the recent discoveries made in the field.

In this conference, we are proud to gather distinguished National and International expertise and scientists who have been invited to participate and share with us their vast experience

One hundred and twenty scientific papers will be presented; seventy papers were assigned to Oral presentation, while 50 papers were assigned for poster presentation.

This conference will be great opportunity to be introduced to each other, and learn from each other experience. This will help us to start from where the others ended. No doubt it is a continuous learning process. We endeavor to establish a network of collaborative institutions which can leads us in the 21 century. For this reason, we invite those interested to join as founding members in establishing the GCC genetic society to fill the application form, which will be available at the registration disk.

On the last day, there will be special session for the patients, families and community members to meet with the experts and discuss with them the latest knowledge concerning the care and prevention of genetic disorders.

On behalf of the organizing committee, I would like to thank her highness Shaikha Sabika bint Ibrahim Al Khalifa for her kind gesture in accepting the patronage of this conference. This indicate her continuous support and encouragement of science and scientists in our beloved country

I would also like to thank the Minister of Health, Dr Khalil Bin Ibrahim Hassan for his support of the activity aiming at preventing and reducing the prevalence of the genetic diseases in the kingdom, as he has clearly indicated that controlling Sickle Cell Disease is priority in his agenda.
Finally, my greatest debt goes to the speakers and all the participants.

On Behalf of the organizing committee I wish you a pleasant stay in Bahrain and a productive meeting.

Organising Committee

• Dr. Shaikha Al-Arrayed	President
• Prof. Mohsen El-Hazmi	Vice President
• Dr. Farouq Al-Zurba	General Secretary
• Dr. Fawzi Amin	Member
• Dr. Tawfeeq Naseeb	Member
• Dr. Abdul Jabbar Al-Abbasi	Member
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• Dr. Amal Al-Jawdar	Member
• Dr. Abdulla Essa	Member
• Ms. Batool Al-Mohandis	Member
• Ms. Amina Abdulla	Member
• Mr. Hamed Al-Hamed	Member

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- Dr. Hassan Abdulla Fakhroo
- Dr. Abdul Aziz Yousuf Hamza
- Dr. Victor Bouljyenkov
- Dr. Mariam Al-Jalahmeh

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• Dr. Layla Bastaki	State of Kuwait
• Dr. Ana Bint Rajab	Sultanate of Oman
• Dr. Amina Al-Zaman	State of Qatar
• Dr. Abdul Aziz Nabaz	Kingdom of Saudi Arabia
• Dr. Abdulla E. Al-Sharoef	Kingdom of Saudi Arabia
• Dr. Jamal Abbas Ma'tooq	Kingdom of Saudi Arabia
• Dr. Nabeel Al-Qahtani	Kingdom of Saudi Arabia
• Dr. Hajar Al-Hoosani	United Arab Emirates

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• Mr. Nabeel Jassim	Member

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• Ms. Layla S. Hashim	Members
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• **Ms. Betty Popovich**	Chairperson
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• Ms. Ebtisam Al-Arayyed	Member
• Mr. Majeed Marhoon	Member
• Ms. Naseema Al-Arrayed	Member

Places of Interest

The location of the conference by itself is enjoyable and relaxing. Different social programmes such as tours, sight seeing etc are available.

PLACES OF INTEREST

There will be a free visit to Bahrain National Museum. Other tours can be arranged by the organizing committee. To make the necessary arrangement please inform the registration office if you wish to visit any of the following places:

Bahrain National Museum, Bait Al Quran, Jebel Al Dukhan, Tree of Life, King Fahad Causeway, Al Jasra House, Heritage Centre, Al Areen Wildlife Park , Bahrain Fort, Arad Fort, Barbar Temple, and more.

Bahrain National Museum

Since 1988 a rich collection of Bahrain's ancient archaeological artifacts have been on display at the purpose-built Bahrain National Museum. The complex is situated in Manama Bay includes three halls devoted to archaeology and two halls depicting the culture and lifestyle of Bahrain's recent pre-industrial past. On National Day, 1995, another hall was opened, the Natural History Hall, focusing on the environment of Bahrain. Foremost among the exhibits in the ancient history section is an actual burial mound which was transported from its site in the desert and reassembled in the museum. Another feature is a tableau which depicts a scene from the Epic of Gilgamesh (in which reference to Bahrain is made as the paradise of Dilmun). Old Quranic manuscripts, notes on astronomy and historical documents and letters are exhibited in a Documents and Manuscripts Hall.

King Fahad Causeway

Linking Bahrain to Saudi Arabia, this is one of the most expensive bridges in the world. The causeway traverses Umm Na'san Island, a wildlife sanctuary, and halfway across the 25 km span is a facility area and border crossing customs zone, complete with a restaurant which provides refreshment for weary travelers. For BD 2 anyone can drive out to the facility area.

Bait Al-Quran

Not far from the National Museum is Bait Al Quran, one of the island's most distinctive pieces of architecture and home to a rare collection of Islamic manuscripts, prints and books. The building combines the roles of mosque, library, museum and study center. All are welcome, but dress codes should be observed by anyone planning to visit the center.

Jebel Al-Dukhan

Situated almost at the center of the island, the Mountain of Smoke is the highest point of Bahrain at 137 meters. On a hot and humid day it is surrounded by a misty haze, from which it gets its name. With nothing more than a good pair of shoes, the climb can easily be made, and the view is worth the effort. The sea on both sides can be seen, and a good perspective of the entire central basin, with its oil wells, occasional acacia tress and sand dunes, can also be seen.

Tree of Life

This well-matured acacia tree stands alone in the otherwise barren desert about two kilometers from Jebel Dukhan. Its green foliage provides welcome shade and is a popular picnic spot, but its source of water remains a mystery.

Al-Jasra House

A fine example of traditional Bahraini architecture, Al Jasra House, which was built shortly after the turn of the century, is constructed of typical local materials such as coral, gypsum, lime and various parts of the palm tree.

Social Programme

Day	Time	Free Tours
Sunday Oct. 5, 2003	16:00 – 20:00	▪ Bait Al-Quraan ▪ Bahrain National Museum or ▪ Seef Mall, ▪ Marina Mall ▪ 'Ali Mall
Monday Oct. 6, 2003	16:00 – 20:00	▪ Tree of Life ▪ Moulds' Factories or ▪ Bab-ul-Bahrain ▪ The Traditional Market

Other tours could be arranged in coordination with the Public Relations and Social Committee.

Supports

- Supreme Council for Woman Bahrain
- Ministry of Health
- Bahrain National Hereditary Anaemia Society
- The Executive Board of the Health Ministers Council for GCC States
- World Health Organisation
- Arabian Gulf University
- Bahrain Medical Society
- Child Development Society

Sponsors

- Bahrain Telecommunications Company (Batelco)
  - Golden Sponsor
- Gulf Air
  - Official Carrier
- Nanogen, Middle East and Northern Africa

Themes

- Clinical Genetics
- Cyto Genetics
- Molecular Genetics
- Genetic Blood Disorders
- Metabolic Disorders
- Community Genetics

Opening Ceremony

Sunday, 5 Octobers 2003

08.00

REGISTRATION

09.00

OPENING CEREMONY

Recitation From the Holy Quran

Opening Remarks	H.H. Shaikha Sabeeka bint Ibrahim Al-Khalifa Chairwoman of the Supreme Council for Women
Opening Remarks	Dr. Shaikha Al Arrayed Chairperson Organising Committee
Speech by	H.E. Dr. Khalil Ebrahim Hassan Minister of Health
Speech by	H.E. Mr. Abdulla Hassan Saif Minister of Finance and National Economy President – Bahrain National Hereditary Anaemia Society
Speech by	H.E. Dr. Hussein A. Gezairy Regional Director, WHO Eastern Mediterranean Region
Speech by	Dr. Vector Boulyjencove Director, Non-Communicable Disease Programme, WHO

10:30 – 11:30

Reception and Opening Exhibition

تمنياتنا بكل التوفيق والنجاح

للمؤتمر الخليجي الأول للوراثة

شركة غاز البحرين الوطنية (ش.م.ب)

Global Reach through Local Commitment

Batelco (Bahrain Telecommunications Company) is a well-recognised provider of world-class communications in the Middle East.

Through our holding company, Batelco Middle East S.C. (BMEC) in Bahrain, we have set up a number of communications partner companies in several key areas of the region including Saudi Arabia, Kuwait, Jordan and Egypt.

Our close association with Cable & Wireless helps us harness international expertise for the benefit of our corporate customers.

By working as a team and combining our resources, we help businesses succeed in the region. In fact, our state-of-the-art communications have been one of the key factors in attracting international banks, financial institutions, trading companies and foreign investors to the Middle East.

Find out more on how Batelco can help uncover a world of opportunities when you set up your business in the Middle East.

Visit our website www.batelco.com
or contact The Manager, Business Customer Development, Batelco, P.O. Box 14, Manama, Kingdom of Bahrain.
Tel: (+973) 985959, Fax: (+973) 905959, e-mail: batelco@batelco.com.bh

بتلكو
Batelco

GULF AIR'S NEW DINING INNOVATIONS

In demonstration of Gulf Air's commitment to exceeding customer expectations and in taking the tradition of Arabian hospitality – and Gulf Air's value offering - to the rest of the world in a unique way, the airline introduced its programme of on board menu enhancements on its flights between the Gulf and London, Paris and Frankfurt on 01 February.

James Hogan
President Chief Executive Gulf Air

"Today, our guests flying with Gulf Air will find the finest dining in the skies between the Gulf and Europe as a result of these enhancements," said Michael Kent, Head of In flight Services, Gulf Air.

The process of enhancement began when Gulf Air introduced its In flight Chef service in its First Class cabins on its flights between the Gulf and London last September. Only three months after the initial launch of the In flight chefs, Gulf Air introduced modern new menus which have raised the standards of its in-flight cuisine to new heights.

"To achieve such high standards, Gulf Air used its qualified chefs to design and structure the service throughout our catering suppliers in a world airline first in order to ensure that the products served onboard meet the exacting standards of 'five star service'," said Mr Kent.

The new First Class menu, which was introduced in December 2002, provides the In flight chefs with materials and produce to facilitate the delivery of a modern contemporary design in support of the innovative service concept.

Michael Kent
Head of In flight Services Gulf Air

"The new menu in our First Class cabin provides guests with a food and beverage experience tailored specifically to the Chef delivery concept," said Mr Kent. "We introduced Gulf Air's new 'Amuse Bouche', a pre-appetiser, which replaces the standard canapé service offered by most other airlines in first class. This is a very modern concept for the airline industry and is normally only offered in top end restaurants."

The selected cuisine for the new menus combines flavours from produce taken from a large number of European and Middle East regions, and has been constructed in a contemporary plating style.

"We expanded the In flight chef service and this new contemporary menu to our Paris and Frankfurt routes this February. From April the service will be introduced on Singapore and Bangkok routes, and it will also be launched on more Far Eastern routes in response to

popular demand from our guests flying with us. New menus are also being designed to suit this particular segment of the market," added Mr Kent.

Business and Economy Class guests are also enjoying a completely new premium offering as a result of the enhancement programme.

Gulf Air's big changes to its dining service in its Business Class includes an eight-tier service offering the individual selection and plating of a wide choice of dishes, in a premium service which is set to become the envy of many airlines' first class offerings, and, which is in fact, a First Class service in a Business Class cabin.

On offer are canapés and the most extensive main course selection ever presented by Gulf Air, comprising a selection of hot or cold entrees as well as a 'light' choice, as well as a large selection of fresh fruit, a choice of premium cheeses combined with date paste, lavosh bread and crudities as well as plated individual desserts, garnished and sauced to style.

Traditional Arabian sweets, coffee and tea, are served at the end of the meal and the airline 's first class signature Arabian coffee and date service is now also served in Business Class.

"All service offerings are presented personally and individually on 'First Class' standard crockery, reinforcing our claim of a First Class dining experience in Business Class," added Mr Kent.

A number of exciting new changes to Gulf Air's Economy Class service include the main meal being offered in two servings. The appetiser and entree are presented to the customer as the first dining course and the dessert and tea/coffee are presented separately - just like in a restaurant.

Throughout the duration of the flight the cabin service conducts regular 'hydration runs' while the introduction of a unique movie treat service provides additional enjoyment to the in-flight entertainment experience. Add to this ice cream being served mid-flight and a small hot or cold snack prior arrival and you have a dining experience to rival many other airlines' business offering. Gulf Air's modern contemporary café style services provide continuous interaction between its guests and flight attendants ensuring service is at hand to meet every need.

أطباء إستشاريون ذوو خبرات تصل إلى ٣٠ عاماً

أخصائيون واستشاريون في:

- أمراض وجراحة العيون وتصحيح البصر بالليزر
- الأمراض الوراثية والفحص قبل الزواج
- أمراض وجراحة الفم والأسنان
- أمراض وجراحة القلب والصدر والأوعية الدموية
- الكيمياء الحيوية و إستشاريي أطفال الأنابيب
- أمراض وجراحة النساء وأطفال الأنابيب وجراحة المناظير
- أمراض وجراحة العظام والكسور والروماتيزم
- أمراض وجراحة المخ والأعصاب والتخطيط الدماغي
- الأمراض النفسية
- الجراحة العامة والباطنية والغدد وجراحة المناظير
- أمراض المسالك البولية والتناسلية والعقم
- الأمراض الجلدية والتناسلية والتجميل الجلدي
- أمراض الأطفال والمواليد
- الأمراض الباطنية والصدرية والجهاز الهضمي والكبد ضغط الدم والكلى
- العقم والتلقيح وقتيي التلقيح الصناعي

عيادة ٢٤ ساعة

خدماتنا تشمل أيضاً

■ عيادة مفتوحة٢٤ ساعة للحالات الطارئة ■ قسم للولادة والنساء ■ عيادة لجراحات التجميل مركز خاص للأجنة وأطفال الأنابيب ■ غرف للعمليات ■ مختبر متكامل وخدمات للأشعة وسيدلية حديثة ■ قسم للعلاج الطبيعي وقسم الفحص بالمناظير ■ خدمة الزيارة المنزلية للمعاينة ■ أطباء زائرون بتخصصات نادرة

مستشفى إبـن النفـيس

Ibn Al-Nafees Hospital

مستشفى متكامل لجميع التخصصات

مملكة البحرين، هاتف: ٨٢٨٢٨٢ - ٨٢٨٢٣٠ ، فاكس: ٨٢٨٢٣٢

في أي زمـــان.. أو مكان رؤية أوضح ... شعور أفضل

خدمات طبية ممتازة بإشراف الدكتور حسن العريض

فريق طبي لخدمات الطوارىء

- فحص المجال البصري وأعصاب العين بأجهزة إلكترونية حديثة.
- التشخيص المبكر للمياه السوداء.
- فحص ضغط العين بالكمبيوتر و القلم الإلكتروني.
- إجراء عمليات المياه البيضاء بإستخدام جهاز الأشعة فوق الصوتية PHACO وزرع عدسات بلاستيكية طرية.
- عمليات الحول و المياه السوداء و زرع القرنيات.
- جميع عمليات التخلص من النظارات.
- فحص عينات من العين (البكتيريا والفيروس والتراخوما).
- تصوير قاع العين، و دراسة أبعاد العين بالأشعة المقطعية.
- علاج أمراض الشبكية بأحدث أجهزة ليزر الشبكية
- دراسة شرايين وطبقات الشبكية بالأشعة الملونة.
- دراسة سمك القرنية وجغرافيا القرنية.
- التخطيط العصبي للعين.
- برنامج لزيارة أطباء و إستشاريين على مدار العام

٣٠ عاما

مركز العيون التخصصي البحرين لليزر العيون

تطلع دائم نحو خدمات طبية متطورة

أحد أقسام
مستشفى إبن النفيس
Ibn Al-Nafees Hospital
مستشفى متكامل لجميع التخصصات

مملكة البحرين - هاتف: ٨٢٨٢٨٣ / ٨٢٨٢٥٧ / ٨٢٨٢٨٢ فاكس: ٨٢٨٢٨٠

List of Speakers

Abdallah S. Daar	Canada
Abdulkarim Hamedi	Iran
Ahmad Aleyasin	Iran
Ahmad Teebi	Saudi Arabia
Aida Al Aqeel	Saudi Arabia
Al Hosani H.	UAE
Alexander Voevodin	Kuwait
Ali Al-Sanousi	Saudi Arabia
Amin El Aqib Mohamed	Saudi Arabia
Andrew E.Czeizel	Hungary
Anna Rajab	Oman
Arjumand S. Warsy	Saudi Arabia
Azimifar Babak	Iran
Behnaz Bayat	Iran
Behnaz Zarbakhsh	Iran
Brain Meyer	Saudi Arabia
D.D. Farhud	Iran
Das. S Nagalla	Bahrain
David Dennison	Saudi Arabia
Elena Samilchuk	Kuwait
Eman Farid	Bahrain
Erol Baysal	UAE
Farideh Akhlaghi	Iran
Fatima Neama	Bahrain
Gholam Ali Mamouri	Iran
Hamza Eskandarni	Saudi Arabia
Hussain Al Mukaroq	Bahrain
J. Andoni URTIZBEREA	France
Laila Ali Akbari	Kuwait
M. Pourjavad	Iran
Magdy Gawish	Egypt
Maha Abu-Henedi	Kuwait
Mahmoud Taleb	UAE
Mahnaz Zeinali	Iran
Makia Marafie	Kuwait
Mariam A. Ghuloom	Bahrain
Mariam Al Muila Hamas	Bahrain
Marios Kambouris	Saudi Arabia
Marwan Abu-Halaweh	Australia
Maryam Masrouri	Iran
Mehdi Shafa Sh. Panahi	Iran
Michel Angistiniotis	Cyprus
Moeen Al-Sayed	Saudi Arabia
Mohamed S. Rashed	Saudi Arabia
Mohammed El Sawy	Egypt
Mohammed Naveed	UAE
Mohsen A F El-Hazmi	Saudi Arabia
Mona O. El Ruby	Egypt
Mostafa K. El Awady	Egypt
Moussa Alkhalaf	Kuwait
Nabeel Al Momen	Bahrain
Nadia Al-Torki	Kuwait
Naeema Aziz	Bahrain
Nagwa A. Meguid	Egypt
Najat Mahdi	Bahrain
Navid Almadani	Iran
Nevein Abou El-Soud	Egypt
O M N Khatib	EMRO
Olusola Sokefun	Nigeria
P. Derakhshandeh-Peykar	Iran
Pamela Carroll	Iran
Panti Fouladi	Iran
Paul Giangrande	UK
Rafia Ghubbash	Bahrain
Rajagopal Krishnamoorthy	France
Rezk Al-Naggar	Kuwait
S. Abedian	Iran
Sadika Ali Al-Awadi	Kuwait
Samaher Al Ahmed	Saudi Arabia
Sarita Agarwal	India
Sawsan Abulhsasan	Kuwait
Shaikha Al Arrayed	Bahrain
Simous Zeinali	Iran
T. Sharma	Bahrain
V. Kumar	Bahrain
Wafaa Eyaid	Saudi Arabia
Wolfgang Trautwein	Nonegen
Ysbrand Poortman	Sweden
Zewar M.Mohga	Egypt

Scientific Programme

Scientific Programme
Scientific Programme
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Scientific Programme

Programme at a Glance
The 1st GCC Genetic Conference

TIME Hours	SUNDAY 5 / 10 / 2003	MONDAY 6 / 10 / 2003	TUESDAY 7 / 10 / 2003
8:00	Registration		
8:30		Scientific Session 4	Scientific Session 5	Scientific Session 12	Scientific Session 13
9:00	Opening Ceremony		
9:30			
10:00	Reception	Coffee Break	
10:30	Exhibition	Exhibition & Scientific Session 6	Exhibition & Scientific Session 7	Exhibition & Scientific Session 14	Exhibition & Scientific Session 15
11:00			
11:30	Session 1 Plenary Session		
12:00		Coffee Break	
12:30			
13:00		Scientific Session 8	Scientific Session 9	Scientific Session 16	Scientific Session 17
13:30	PRAYER / LUNCH		
14:00		PRAYER / LUNCH	
14:30			
15:00	Scientific Session 2	Scientific Session 3	Workshop: Birth Defects	Scientific Session 10	Scientific Session 11	Workshop: Premarital counsel	Community Seminar B. N.I.A. Society Session 18	Community Seminar Ch. Develop. Society Session 19	CLOSING
15:30			
16:00			
16:30			
17:00			
17:30			
18:00	Free Time		
18:30			
19:00			
19:30	DINNER		

Workshops

Sunday - October 5, 2003

15: 00 - 17:00

Premarital Counseling Workshop

Dr. Shaikha Al-Arrayed

Dr. Sameera Al-Sairafi

Prof. Mohsin El Hazmi

Monday - October 6, 2003

15:30-17:30

Birth Defect Workshop

Dr. Shaikha Al-Arrayed

Prof. Andrew E. Czeizel

Prof. Ysbrand Poortman

- Registration for the attending the workshop is required at the Registration Desk
- Registration Fee for Each workshop - BD. 10:000

Sunday - October 5, 2003

Session 1

Plenary Session

Chairman	Dr. Shaikha Al-Arrayed
Co-Chairman	Mrs. Batool Al Muhandis

11:30	Genetic Programme in GCC
	Dr. Tawfeeq Khooja GCC Executive Office
11:55	Eastern Mediterranean Approach to Genetic Diseases, primary prevention & control.
	Dr. O.M. Khatib – EMRO
12:20	Genetic Disorders among Arabs
	Dr. Ahmad Said Teebi – Saudi Arabia
12:45	Decline trends in the incidence of sickle cell disease among Bahrain newborns
	Dr. Shaikha Al Arrayed – Bahrain
13:10 – 13:30	Panel Discussion

13:30 – 15:30 PRAYER and LUNCH

Sunday - October 5, 2003

Session 2 **Hall 1**

Community Genetics

Chairman	Dr. Zeinali S
Co-Chairman	Dr. A Jabbar

15:30	HH Princes Al-JawharaAl-Ebrahim, Wife of HH the Custodian of the Two Holy Mosques Centre for Molecular Medicine, genetic & Inherited Diseases
	Dr. Rafia Ghubbash
15:50	National Congenital Abnormality Registry (NCAR) in the UAE
	Dr. Al Hosani H.– UAE
16:10	Pharmacogenetics and Pharmacogenomics. Perspectives towards individualized drug therapy?
	R. Krishnamoorthy – France
16:30	Orphan Diseases in Oman, Value of the Data Collection and Prospects for prevention.
	Dr. Anna Rajab – Oman
16:50	National Program for the Prevention of B-Thalassemia in Iran
	Dr. Sirous Zeinali – Iran
16:50 – 17:00	Panel Discussion
19:30	DINNER

Sunday - October 5, 2003

Session 3 Hall 2

Community Genetics

Chairman	Prof. Mohsen El Hazmi
Co-Chairman	Dr. Awatif Sharaf

15:30	A framework for Ethics in relation to community genetics in Islamic countries
	Prof Mohsen El Hazmi – Saudi Arabia
15:50	Primary prevention of some congenital abnormalities.
	Prof Andrew E Czizel – Hungary
16:10	Attitudes of Medical Personnel to Ethical issues in Iran
	Dr. D D Farhud – Iran
16:30	Multifactorial Disorders in Saudi Arabia
	Prof Arjumand S. Warsy - Saudi Arabia
16:50	The European network on Epidemiology of rare disorders
	Dr. Ysbrand Poortman – Sweden
16:50 – 17:00	Panel Discussion
19:30	DINNER

Monday - October 6, 2003

Session 4 **Hall 1**

Metabolic Disorders

Chairman	Dr. Hajar Al-Husani
Co-Chairman	Dr. Moeen Al Sayed

08:30	Spectrum of Inborn Error of metabolism in Saudi Arabia
	Dr. Moeen Al Sayed – Saudi Arabia
08:50	Clinical Application of Tandem Mass Spectrometry: Ten Years of Diagnosis and Screening for Inherited Metabolic Diseases
	Dr. Mohd S Rashid – Saudi Arabia
09:10	Genetic metabolic disorders of Saudi Arabia
	Dr. Aida Al Aqeel – Saudi Arabia
09:30	Inherited metabolic disorders in the united Arab Emirates
	Dr. Mohamed Taleb – UAE
09:50 – 10:00	Panel Discussion

10:00 – 10:30 COFFEE BREAK

Monday - October 6, 2003

Session 5 **Hall 2**

Community Genetics

Chairman	Dr. Ysbrand Poortman
Co-Chairman	Dr. Ghazi Al-Mahrous

08:30	National Genetic Blood disorder project: student screening (1999-2002) Dr. Shaikha Al Arrayed – Bahrain
08:45	Evaluation of the National Neonatal screening program in UAE, analysis of eight years experience Dr. Al Hosani H – UAE
09:00	The GCC genetic disease registry Dr. Ali Al Sanousi – Saudi Arabia
09:15	The international genetic alliances of parents and patients organization Dr. Ysbrand Poortman – Sweden
09:30	Challenges of New Genomics: What is the Human Genome Project, and what challenges is it posing on health systems? Dr. Naeema Aziz
09:45 – 10:00	Panel Discussion

10:00 – 10:30 COFFEE BREAK

Monday - October 6, 2003

Session 6 Hall 1

Cyto & Clinical Genetics

Chairman	Dr. Layla Bastaki
Co-Chairman	Dr. Fatima Neama

10:30	Panhypopitutrism, Cranial Dysmorphia, Hypogonadism and Mental Retardation: A new syndrome
	Dr. Sadika Ali Al Awadi Kuwait
10:50	Three Bahrainis families with Retinitis Pigmentosa, Ataxia, Neuropathy, Cerebellar hypoplasia, Mental Retardation, and hypogonadotrophic hypogonadism
	Dr. Fatima Neama – Bahrain
11:10	Trisomy 5 as the sole cytogenetic abnormality in infantile precursor B cell acute lymphoblastic leukaemia
	Dr. Moussa Al Khalaf – Kuwait
11:30	Cytogenetic and Hematologic Investigation of Severe Chemical Injuries Exposed to Mustard Gas during Iran-Iraq Conflict
	Dr. Mehdi Shafa - Iran
11:50 – 12:00	Panel Discussion

12:00 – 12:30 COFFEE BREAK

Monday - October 6, 2003

Session 7 Hall 2

New Techniques and Services

Chairman	Dr. Ahmad Tebbi
Co-Chairman	Dr. Naeema Aziz

10:30	The Power of Microelectronics meets Molecular Biology and sets a new Standard of Accuracy
	Mr. Wolfgang Trautwein
10:50	The Center for Arab Genomic Studies (CAGS)
	Dr. Mahmoud Taleb Al Ali – UAE
11:10	Hereditary Blood diseases Eradication in Saudi Arabia
	Mrs. Huda Al Mansour
11:30	Rapid Dtection and Identification of C. Jejuni and C. Coliform Chicken Sample by Real Time PCR
	Dr. Abu Halawawh Marwan
11:50 – 12:00	Panel Discussion

12:00 – 12:30 COFFEE BREAK

Monday - October 6, 2003

Session 8 Hall 1

Molecular Genetics

Chairman	Dr. Krishnamoorthy
Co-Chairman	Dr. Baysal E.

12:30	Localization of the gene for a Novel Autosomal Recessive Neuromuscular Disease featuring tremulous and Myoclonic Dystonia with MRI white matter Alterations
	Dr. M. Kambouris – Saudi Arabia
12:45	Mutational analysis of common single Gene Disorders in Kuwait
	Dr. Elena Samilchuk – Kuwait
13:00	Molecular Basis of β Thalassemia in the United Arab Emirates
	Dr. Baysal E. – UAE
13:15	Alpha 2 poly adenylated signal mutation in Bahrain, A genotype/phenotype correlation
	Mr. Naheel Al Momen – Bahrain
13:30	Mutation detection and prenatal diagnosis of patients with Cystic Fibrosis (CF) in Iran
	Dr. P. Derakhshandeh-Peykar – Iran
13:45 – 14:00	Panel Discussion

14:00 – 15:30 PRAYER and LUNCH

Monday - October 6, 2003

Session 9 — Hall 2

Medical Genetics

Chairman	Dr. Faroug Al-Zurba
Co-Chairman	Dr. A. Hadi Khalil

12:30	Medical genetics: A physician approach. Saudi Arabia Dr. Aida Al Aqeel – Saudi Arabia
12:45	Alpha Thalassemia genotype in Iran Dr. Behnaz Zarbakhsh – Iran
13:00	Rare Clinical Presentation of Ethylmalonic Adipicaciduria Dr. Naveed M – Iran
13:15	Molecular Characterization of a novel autosomal dominant hyaline body myopathy (HBM) in a large Saudi family. Dr. Brain Meyer
13:30	Pregnancy Outcomes in Women with Pregestational Diabetes Compared with the Gestational Diabetic Mothers Dr. Farideh akhlaghi – Egypt
13:45 – 14:00	Panel Discussion

14:00 – 15:30 PRAYER and LUNCH

Monday - October 6, 2003

Session 10 **Hall 1**

Molecular Genetics

	Chairman	Dr. Moiz Bakhiet
	Co-Chairman	Dr. M. Kambouris
15:30	Aims and Objectives in creation of National Molecular medicine network in Iran	
	Dr. Sirous Zeinali – Iran	
15:50	Rantes promotes growth and survival of human first trimester forebrain astrocytes	
	Dr. Moiz Bakhiet – Bahrain	
16:10	Study of folic acid pathway genes alteration as maternal risk factor of down syndrome	
	Dr. Ahmed Aleyasin – Iran	
16:30	Localization of genes for a novel disease characterized by intestinal lymphangiectasia to 6p21-22 and 13q14.2-21.1	
	Dr. M. Kambouris – Saudi Arabia	
16:50	Molecular genetics of mild forms of sickle cell disease in Bahrain	
	Mr. Al Momen N Nabeel – Bahrain	
16:50 – 17:00	Panel Discussion	

19:30 **DINNER**

Monday - October 6, 2003

Session 11 Hall 2

Medical Genetics

Chairman	Dr. Amin El Agib
Co-Chairman	Dr. Das N.S.

15:30	Williams Syndrome in Kuwait: A Study of Phenotype-Genotype Correlation Using Molecular Cytogenetics Technique Dr. Sawsan Abu Alhassan - Kuwait
15:50	Anatomical and developmental study of congenital Anomalies of Nervous system in fetus and newborn in Zeinab University Hospital in Mashad from 2001-2002. Dr. M. Pourjavad – Iran
16:10	Fourier transformed infrared spectroscopy (ftir)as a diagnosis tool in Cystinuria Dr. Das N.S. – Bahrain
16:30	An Investigation on chromosomal abnormalities and physiological study of mentally retarded male children in Tehran Dr. Behnaz Bayat – Iran
16:50	A new Arab kindred with two cases of limb/pelvis hypoplasia in Kuwait Dr. Rezk Al Naggar - Kuwait
16:50 – 17:00	Panel Discussion

19:30 DINNER

Tuesday – October 7, 2003

Session 12 Hall 1

Community Genetics

Chairman	Dr. Arjumand Warsi
Co-Chairman	Dr. Tawfeeq Naseeb

08:30	An outline for community genetic services for the gulf – the Saudi experience Prof. Mohsen El Hazmi – Saudi Arabia
08:50	World alliance for the prevention of birth defects Prof. Ysbrand Poortman – Sweden
09:10	Student screening project: The management of school campaign Dr. Mariam Al Mulla – Bahrain
09:30	Bahrain Birth Defect Registry Dr. Shaikha Al-Arrayed – Bahrain
09:50 – 10:00	Panel Discussion

10:00 - 10:30 COFFEE BREAK

Tuesday – October 7, 2003

Session 13 — Hall 2

Community Genetics

Chairman	Dr. Anna Rajab
Co-Chairman	Dr. Reda Ali

08:30	Pscho-social aspect and school performance for children with sickle cell disease in Hafoof and sudan Dr. Amin El Agib – Saudi Arabia
08:45	Hematopoietic Stem Cell Transplantation for Inherited Diseases in the Sultanate of Oman Dr. David Dennison – Oman
09:00	Manifestation of sickle cell disease in Bahrain Dr. Hussain Al Mukaraq – Bahrain
09:15	Endocrine complication of B Thalassemia major in Bahrain Dr. Najat Mahdi – Bahrain
09:30	Spectrum of beta globin gene mutations in west of Iran bordering Turkey, Iraq Dr. P. Fouladi
09:45 – 10:00	Panel Discussion

10:00 - 10:30 COFFEE BREAK

Tuesday – October 7, 2003

Session 14 **Hall 1**

Dysmorphology

Chairman	Dr. O.M Khatib
Co-Chairman	Dr. Fawzi Ameen

10:30	Osteopathia Striata with cranial sclerosis: A Brief clinical report on a Bedouin girl Dr. Rezk Al Naggar – Kuwait
10:45	Primary amenorrhea, infantile uterus, Alopecia. Diabetes mellitus, Intracranial calcification in two sisters: A new syndrome Dr. Laila Bastaki – Kuwait
11:00	5-oxioprolinuria: biochemical observations and case report Dr. Mohammed Naveed – UAE
11:15	Screening for Hypertrophic Cardiomyopathy Impact of Neonatal size on Cardiac dimensions and functions in Infants of diabetic mothers Dr. Wafaa Eyaid – Saudi Arabia
11:30	Recessive Robinow syndrome with additional features Dr. Mohammed Naveed UAE
11:45 – 12:00	**Panel Discussion**

12:00 – 12:30 **COFFEE BREAK**

Tuesday – October 7, 2003

Session 15 **Hall 2**

Molecular

Chairman	Dr. Aida Al Aqeel
Co-Chairman	Dr. Faisal Al Nasser

10:30	Novel Mutations underlying Nephrogenic Diabetes Insipidus (NDI) Dr Carroll P – Saudi Arabia
10:45	Molecular Studies on HbH disease in the United Arab Emirate Dr. Baysal E – UAE
11:00	Correlation of a novel perforin Gene deletion mutation and Flowcytometric detection of Perforin in an Omani patient with Familial Hemophagocytic Lymphohistiocytosis. Implication for rapid diagnosis Dr. David Dennison – Oman
11:15	The allele frequency of ST14(DXS52) in hemophilia A and normal population in Iran Dr. Azimifar Babak – Iran
11:30	Deletion analysis and prenatal diagnosis in Iranian spinal Muscular atrophy patients type 1-11 Dr. P. Derakhshandeh-Peykar – Iran
11:45 – 12:00	Panel Discussion

12:00 – 12:30 COFFEE BREAK

Tuesday – October 7, 2003

Session 16 **Hall 1**

Community Genetics

Chairman	Prof. Arjumond Warsy
Co-Chairman	Dr. Eman Farid

12:30	Distribution of the clinically important HLA Alleles Among Healthy Bahraini Population Dr. Eman Farid – Bahrain
12:45	Report on the out come of 266 prenatal diagnosis of beta thalassemia in a private clinic in Iran since 2000 Dr. M. Masrouri – Iran
13:00	Genetic resistance against HIV infection and Aids Dr. Alexander Voevodin
13:15	Genetics of Obesity Prof. Arjumond Warsy Saudi Arabia
13:30	Two Founder Mutations are Responsible for Papillon Le-Fevre Syndrome in Saudi families Dr. Samaher El-Samadi – Saudi Arabia
13:45 – 14:00	Panel Discussion

14:00 – 15:30 **PRAYER and LUNCH**

Tuesday – October 7, 2003

Session 17 Hall 2

Reproductive Genetics

Chairman	Dr. Abdulla Essa
Co-Chairman	Dr. Mariam A Ghuloom

12:30	Comparison of intra-cytoplasmic sperm injection outcome, using testicular sperm versus seminal sperm Dr. Mariam A Ghuloom – Bahrain
12:50	A study on triple maternal serum screen as a tool of prenatal diagnosis in Bahrain Dr. Das N.S – Bahrain
13:10	Haplotype Analysis of Related ATM Markers Facilitate Prenatal Diagnosis in Iranian Ataxia Telangiectasia Patients Dr. Behnaz Bavar – Iran
13:30	Preimplantation Genetic Diagnosis as Frontier of Early Detection: Are we ready to implement it in the GCC Countries Dr. Hamza Eskandarani – Saudi Arabia
13:50 – 14:00	Panel Discussion

14:00 – 15:30 PRAYER and LUNCH

Tuesday – October 7, 2003

15:30 hours

Closing Ceremony

Community Seminars

Activities of the Societies

برنامج فعالية جمعية أمراض الدم الوراثية

يوم الثلاثاء - ٧ أكتوبر ٢٠٠٣م

من الساعة الثالثة إلى السادسة مساء

- تلاوة من آي الذكر الحكيم
- كلمة منظم الحفل
- كلمة الجمعية الأهلية لأمراض الدم الوراثية – البحرين
- كلمة الدكتور محسن الخازمي .. المملكة العربية السعودية
- كلمة الدكتورة هدى المنصور .. المملكة العربية السعودية
- عرض فيلم تثقيفي حول أمراض الدم

Community Seminars

Activities of the Societies

برنامج فعالية جمعية تنمية الطفولة

يوم الثلاثاء - ٧ أكتوبر ٢٠٠٣م

من الساعة الثالثة إلى السادسة مساء

- تلاوة من آي الذكر الحكيم
- كلمة الدكتور فؤاد شهاب
- عرض بالكمبيوتر حول أهداف وأنشطة الجمعية ومركز النطق والسمع
- كلمة يلقيها أحد أطفال المركز
- نشيد من تقديم أطفال المركز
- تكريم مدرسات المركز المتميزات
- افتتاح المعرض

Abstracts

Abstracts

Abstracts

Abstracts

Abstracts

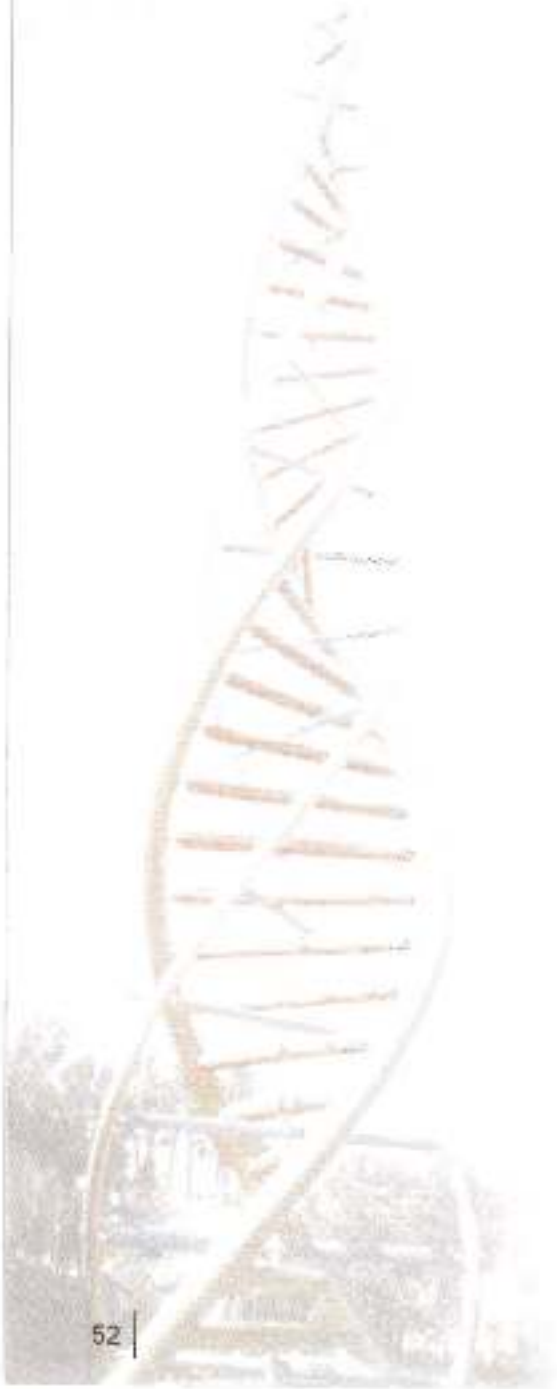

Table of Abstracts

Haemophilia

Paul Giangrande
Oxford Haemophilia Centre (UK)

Haemophilia provides an excellent example of how an understanding of the genetic basis of a disease can offer the prospect of treatment and even a cure for a serious condition. Haemophilia A is an X-linked congenital disorder of coagulation, characterised by deficiency of factor VIII (or IX) in the blood. The molecular basis is now well understood and many cases of haemophilia are the consequence of an inversion in intron 22 of the factor VIII gene. The hallmark of severe haemophilia is recurrent and spontaneous bleeding into joints which can result in disabling arthritis. Bleeding into muscles and soft tissues is also seen frequently. In the absence of effective treatment, survival into adolescence is unusual and it should be borne in mind that the World Federation of Haemophilia estimates that two thirds of people with haemophilia in the world still receive no treatment at all. About 15% of patients with haemophilia A develop inhibitory antibodies. Coagulation factor concentrates were first developed in the early 1970's. These are made from pooled plasma and unfortunately many haemophiliacs were infected with hepatitis and/or HIV. Recombinant factors VIII and IX are now available but are expensive. Transgenic animals have been developed which offer the prospect of production of potentially unlimited quantities of proteins at low cost, and free of human pathogens. Research is also directed to production of modified molecules, with more favourable properties eg. longer half-life or reduced immunogenicity. Clinical trials of gene therapy have begun recently, with encouraging results.

Trisomy 5 as the sole cytogenetic abnormality in infantile precursor-B cell acute lymphoblastic leukemia

Moussa Alkhalaf*; Rajaa Marouf **; Salah Al-Humoud**; Mona H.A. Burhamah***
*Departments of Biochemistry, **Pathology, Faculty of Medicine, Kuwait University; ***Pediatric Hematology Unit, Al-Sabah, ***NBK Pediatric Hospital, Kuwait

Cytogenetic analysis of blast cells in pediatric acute lymphoblastic leukemia (ALL) has led to the recognition of specific nonrandom chromosomal abnormalities of prognostic value. We describe a 22-month-old boy with precursor-B cell (precurser-B-ALL) in whose leukemic cells trisomy 5 (+5) was the only cytogenetic abnormality. The trisomy 5 was identified by G-banding as well as by fluorescent in situ hybridization (FISH) with specific chromosome 5 probe. This patient is the youngest to be reported with trisomy 5 as the sole cytogenetic abnormality in ALL. The cytogenetic aberration in this patient was identified at diagnosis and before the treatment initiation, whereas the trisomy 5 described in the other cases was seen at a delayed stage of the disease and after periods of intensive chemotherapy. The prognostic importance of the trisomy 5 in pediatric-ALL will be discussed.

A Microdeletion of 22q11.2 in a Mosaic Form in a Patient with Unusual Presentation

N.A.Al-Torki, S.J.Abulhasan, A.Al-Adwany, S.A.Al-Awadi
Kuwait Medical Genetics Centre

We report on a 5 years kuwaiti male child with unusual clinical presentation He has minor dysmorphic features in a form of oxycephaly, high broad forehead, downslanting palpebral fissures, squint, cupid bow In addition, he has esophageal atresia, tracheo-esophageal fistula (T.O.F), congenital heart defect (CHD), epilepsy and mild developmental delay. The molecular cytogenetic analysis (FISH)technique revealed presence of a mosaic form of a deletion 22q11.2 region in a ratio of 78% to 22%of normal karyotype

Osteopathia Striata with Cranial Sclerosis: A Brief Clinical Report on a Bedouin Girl

Rezk L. Al-Naggar, Sadika A. Al-Awadi, Maha M. Abuhenedi, Laila A. Bastaki.
Kuwait Medical Genetic Center (KMGC)

Objective: This report describes a case of Osteopathia Striata with Cranial Sclerosis (OS-CS) in a Bedouin girl. This patient, to our knowledge is the first to be reported in KMGC, throwing light on the clinical and radiological findings. Methods: Clinical examination, skeletal survey, echocardiography, ultrasonography and chromosomal study. Results: Clinical examination of the patient showed craniofacial dysmorphic features including overgrowth of the craniofacial bones, developmental delay, hearing impairment, congenital heart(VSD)and repeated attacks of seizures. Radiological findings included marked ossification and sclerosis of the base of the skull and metaphyseal widening of the long bones with linear striations. Conclusion: In the few reported cases of OS-CS all over the world, this case could be a typical form of the syndrome with dominant inheritance and the first case to be described in Kuwait.

Endocrinal Deficits In Down syndrome patients due to Hypozincemia

Rezk L.Al-Naggar, Sadika A.Al-Awadi, Najat A.Al-Awadi
Kuwait Medical Genetic Center

The most common endocrinal deficits in Down syndrome (DS) are overt and subclinical hypothyroidism. Zinc deficiency has been found to impair growth rate and immune response. This study aims to evaluate the role of zinc deficiency in thyroid & pituitary functions in DS patients after zinc supplementation.An inverse correlation has been found between TSH levels and zinc values in hypozincemic patients before therapy ($P=<0.05$). Higher TSH levels (>4.2mIu/L) in 47% of cases. A significant difference in GH &IGF-1 levels between hypozincemic and normozincemic patients ($P=<0.001$ for both) has been found.A significant association between impaired thyroid function and GH and zinc deficiency, ($P=<0.05$).There was an improvement in thyroid function after zinc supplementation, so we recommend zinc therapy in hypozincemic DS patients.

Acrodysostosis in a Kuwaiti boy: brief clinical report.

Rezk L.Al Naggar, Sadika A. Al-Awadi, Samir A. Madi, Laila A. Bastaki.
Kuwait Medical Genetic Center

Objectives: This report describes a Kuwaiti boy with acrodysostosis. This disorder is characterized by short nose, open mouth, prognathism and short hands/feet. Methods: Clinical examination, skeletal survey, cechocardiography, ultrasonography and chromosomal analysis were carried out. Results: On examination he had short stature mainly acromelic and the hands did not show trident sign. He had also a characteristic facies with broad/flat nasal bridge, short nose with upturned nostrils, congenital heart and mental retardation. Radiographic examination showed acromelia, cone-shaped epiphyses of bones of hands/feet and scoliosis of thoracolumber spine. CT-brain scan revealed mild ventriculomegaly and brain atrophy. Conclusion: Our patient could be a typical case of acrodysostosis with autosomal dominant mode of inheritance.

A new Arab Kindred with two cases of Limb/Pelvis hypoplasia (Al-Awadi/Raas-Rothschild) syndrome in Kuwait

Rezk L. Al-Naggar, Sadika A. Al-Awadi, Maha M. Abu-henedi, Amal Al-adwani, Sawsan J Abu-Alhassan, Laila A. Bastaki.
Institution: Kuwait Medical Genetic Center

This study describes two new cases with limb/pelvis hypoplasia.They had intercalary limb reduction defects, hypoplastic pelvic bones and unusual facial features.Clinical examination, skeletal survey, ecchocardiography and chromosomal analysis/FISH techniques were carried out.The two cases had pre/postnatal growth delay.The facial features included cappilary hemangioma, sparse/brown hair, short nose, dysplastic ears and retrognathia Severe and symmetric limb reduction anomaly, phallus/clitoral enlargment have been found. The boy had balanced, t(1; 3)of paternal origin and the other case had 46, XX karyotype. Conclusion: Both cases share in common severe pelvic hypoplasia, symmetric limb RD and recessive inheritance and absence of premature centromere separation which exclude Robert's-SC phocomelia

Wollcot-Rallison Syndrome in a Bedouin Boy: A Case Report

Marafie MJ, Redha MA, Al-Naggar RL
Kuwait Medical Genetic Center

Wolcott-Rallison syndrome (WRS, MIM 226980) is a rare autosomal recessive disorder, presenting mainly with infancy onset insulin-dependant diabetes mellitus, spondylo-epiphyseo-metaphyseal dysplasia, osteoporosis and short stature. Other additional manifestations have been frequently reported such as microcephaly, mental retardation, seizures, hepatic and renal insufficiency, cardio-vascular abnormalities, neutropenia, skin and teeth changes. Chromosomal abnormalities were also found in some patients. The gene EIF2AK3, encoding translation initiation factor 2-alpha kinase 3, located on chromosome 2p12, was postulated to be the candidate gene for WRS. To date, 11 cases from diverse ethnic families have been described in the literature, 4 of which were from 3 consanguineous arab families. Here we report a 7 year-old bedouin boy with the typical clinical and radiological phenotypes of WRS. His karyotype was 46,XY. There were neither aberration involving chromosome 15 or 2, and neither deletion or uniparental disomy of chromosome 6. This syndrome might be underdiagnosed in the population of arab peninsula; possibly due to early death of diabetic infants and late expression of its full phenotype.

Keywords: Wolcott-Rallison syndrome; neonatal diabetes; skeletal dysplasia

Weissenbacher-Zweymuller Syndrome in a Kuwaiti boy: a Brief Clinical Report

Maha M Abu-Henedi, Rezk L. Al-Naggar, Laila A. Bastaki, Sadika A. Al-Awadi.
Kuwait Medical Genetic Center

The weissenbacher-Zweymuller syndrome is a congenital neonatal rhizomelic dwarfism with metaphyseal widening of the long bones and vertebral coronal clefts. The syndrome has been lumped together with Stickler and Marshal syndromes, and has been called the neonatal form of Stickler syndrome. We report on a term female infant born to a nonconsanguineous, healthy parents. She had a rhizomelic shortening of the upper and lower limbs with a body length below 3rd centile. She also had micro-retrognathia, cleft palate, broad forehead, hypertelorism, epicanthic fold, protruding eyes, depressed nasal bridge, congenital heart (VSD & ASD) and enlarged big joints. Radiological findings included short femora and humeri with widening of the metaphyses of the long bones and coronal clefting of the vertebrae. Fundus examination, hearing assessment and chromosomal analysis were normal. Our case could represent a typical phenotype of WZS and considered as the 1st case to be reported in our center.

Molecular Cytogenetic Study of Cases with Short Stature

Nagwa A. Meguid *; Mahmoud A Dardir A. A-M.;Fayek H.; Hammad S.; Atteya H.
Prof. Human Genetics and Head Dept. Research on Children with Special Needs, National Research Centre, Egypt.

Among 150 cases with short stature and delayed puberty referred to the outpatient clinic of National Research Centre, during two years period, we selected 25 Egyptian girls with phenotype far more severely affected than expected in Turner syndrome. Initially, the karyotype in some cases was thought to be 45, X with a chromosome marker. However, re-examination with FISH probes showed 14 subjects with 45, X/46, X r(X) karyotype. Seven subjects with 46, XisoX(q) karyotype; 2 subjects with 45, X karyotype; and 2 subjects with 46, XX karyotype. Tiny ring X was present in 5 cases, and inactivation was proved by molecular cytogenetic techniques. The clinical picture of cases with ring (X) chromosome includes mental retardation in 8 patients (the non-verbal I.Q. tends to be lower than the verbal I.Q.) and learning disability in 3 cases. Dysmorphic features are found in 3 cases and limb anomalies in 5 cases.

Our results showed that the severe phenotype was present in the cases with tiny ring (X) chromosomes suggesting mutation in the X chromosome inactivation pathway and that the inability of these rings to inactivate was responsible for the severe phenotypes. We confirm the advent of in situ hybridization with chromosome specific DNA probes in identifying small structurally abnormal chromosomes.

Isolation of the C-terminal region of a novel tumor suppressor gene

Mostafa K. El Awady*, Wael T. El Garf, Marwa I. Khalil, Khalda S. Amr, Khaled A. Abdel Aziz, Noha G. El-Din
Department of Biomedical Technology, National Research Center Dokki, Egypt

Soluble egg antigen (SEA) of S hematobium has been associated with increased urothelial cell proliferation and higher expression of cell cycle genes. To determine genes involved in SEA induced stimulation of urothelial cells, J82 cells were stimulated with SEA for 24 and 96 hours. Total cellular RNA was isolated and analyzed by differential display technique. A significantly down regulated cDNA after 96 h exposure to SEA, was cloned and sequenced. The obtained sequence was found homologous to E Coli and S flexneri Delta-2 isopentenyl pyrophosphate transferase (IPPT), but without homology to the published human genome draft. Using primers derived from the obtained sequence, a fragment of the same size (196 bp) was obtained upon amplification of genomic DNA from human peripheral leukocytes. Furthermore, RT-PCR analysis was performed on 19 different tumors of diverse ethologies. Our results demonstrated that relative abundance of specific RNA for this gene was reduced in all tumors tested. To further confirm the identity of this transcript, the amplified fragments from three normal human tissues (bladder, liver and maxillary sinus) were sequenced. Alignment on human genome data base as well as pair alignment with 3 key enzymes in mevalonate pathway resulted in no homology with any of the reported sequences. In conclusion, we have isolated and characterized the C-terminal region of a putative tumor suppressor gene that is believed to be a member of the mevalonate pathway, a finding which paves the way for more studies on the role of his pathway in human malignancy.

Williams Syndrome in Kuwait: A Study of Phenotype-Genotype Correlation Using Molecular Cytogenetics Technique

Abulhasan S.; Al-Daboos R.; Al-Turky N.; Al-Ateegi W.; Abdulrasool M.; Al-Adwani A.; Al-Awadi S.
Kuwait Medical Genetics Centre

Williams syndrome (WS) is a developmental disorder affecting vascular and connective tissue and central nervous system.This syndrome is delineated as a microdeletion of 7q11.23 region in hemizygous form.Fluorescence in situ hybridization(FISH)is a useful technique in the diagnosis of all microdeletion syndromes that would otherwise be difficult to diagnos using standard cytogenetics tools.We report 15 sporadic cases with and without typical Williams syndrome features using Elastin gene locus(ELN)on chromosome 7q11.23 as a molecular cytogenetics probe (VYSIS)to clarify the correlation between the phenotype and the elastin locus. Cytogenetics analysis was performed as a complimentary study to exclude any other abberations. The parental origin of the patients were studied. Five out of fifteen patients (33%) were found to have a submicroscopic dejetion of the elastin gene locus(7q11.23).Other results will be discuss. In conclusion, FISH technique with combined with conventional chromosome banding analysis is very

Panhypopututrism, Craniofacial Dysmorphia Hypogonadism and Mental Retardation: A New Syndrome

Sadika Ali Al-Awadi, Kamal Kamel Naguig, Laila Ali akbar bastaki
Kuwait Medical Genetics Centre

We report on a Kuwaiti male with congenital absence of pituitary gland, craniofacial Dysmorphia, PDA, and hypogonadism. Early assessment of hormonal profile revealed panhypopituitrism, low free thyroxin and thyroid stimulating hormones, undetectable prolactin response, and deficiency of ACTH & Cortisol, low serum LH & FSH and growth hormone deficiency. Both testosterone and Dihydrotestosterone (DHT) were low. Magnetic resonance imagining study showed absence of the anterior lobe of the pituitary gland, while the pituitary stalk was normal. There was generalized brain atrophy. Chromosomal study was 46, XY. FISH study showed undetectable deletion Hormonal replacement therapy started early, which modified the phenotype of the proband. Searching in the literature, London dysmorphology database and POSSUM revealed that these combinations of anomalies had not been reported previously and representing a new syndrome.

Keywords: Anterior absence of pituitary, panhypopituitrism, hypogonadism, craniofacial dysmorphia, Mental retardation.

Fetal Diagnosis: In High Risk Couples of b-Thalassemia

Agarwal S, Gupta UR and Gupta A
SGPGIMS Lucknow India

Objectives: Thalassemia is the commonest single gene disorder & is widely distributed in Asian Indians with average prevalence rate of 4% & with a high prevalence among Sindhis, Punjabis, Gujratis & Bengalis. Prevention & control of beta thalassemia disease needs accurate diagnosis of carriers and proper genetic counseling. Method: prenatal diagnosis can be performed in the first or second trimester of pregnancy by DNA analysis using polymerase chain reaction (PCR). Since there are 17 mutations besides rare ones causing beta thalassemia in Asian Indians the point mutation detection by reverse dot blot allele specific oligonucleotide (ASO) hybridization for common mutations along with ARMS technique was developed as one go for prenatal diagnosis. Maternal contamination of fetal DNA was ruled out by variable number of tandem repeat (VNTR) analysis using Apo-B site. Results: Using both techniques (RDB & ARMS) we were able to offer complete diagnosis in 57 pregnancies with 100% accuracy. On molecular analysis 19

Mutation Analysis of Common Single Gene Disorders in Kuwait

Samilchuk E., Al Awadi S.
Kuwait Medical Genetics Center

To identify mutations underlying common single gene disorders in Kuwait, DNA samples from patients with phenylketonuria (PKU), spinal muscular atrophy (SMA), cystic fibrosis (CF) including congenital bilateral absence of vas deferens (CBAVD), as well as from G6PD deficient male Kuwaitis have been studied using PCR, RFLP, heteroduplex analysis, and DNA sequencing. The following pathogenic mutations and polymorphism have been identified:

PKU: 1066nt11g->a in 34.6%, K363fsdelG in 15.4%, IVS4nt5g->t in 15.4%, IVS2nt5g->t in 11.5%, G352fsdelG in 7.6%, and DelE2-IVS2nt1, P281L, R208K, I224T each in 3.8% of PKU chromosomes.

SMA: deletion of the SMN (exons 7 & 8) gene in all SMA families; deletion of the NAIP (exon 5) gene in 85% of SMA type I and in 40% of SMA type II-III families.

CF: deltaF508 in 16.6%, W1282X in 10.4%, I1234V in 8.3%, 1548delG in 8.3%, S549N 8.3%, and the C128R in 4.2% of CF chromosomes. The novel C128R mutation produces a severe clinical phenotype.

CBAVD: deltaF508 in 16.6%, IVS8-5T in 33.3% of CBAVD chromosomes.

G6PD-D: 563C->T in 72.9%, 202 G->A in 14.3%, 1003 G->A in 7.1%, 143 T->C in 1.4% of G6PD-deficient Kuwaiti males.

(TA)n polymorphism in the promoter of the UDPGT1 gene: (TA) 7 allele is present in 60% of G6PD deficient Kuwaiti males (44.3% heterozygous and 15.7% homozygous).

Mutations have also been identified for single families with Tay-Sachs disease, carbonic anhydrase II deficiency, LHON, and CFO.

Orphan Diseases in Oman. Value of the Data Collection and Prospects for Prevention

Anna Rajab
Genetic Blood Disorders Unit, DGHA, MOH, Sultanate of Oman

The revolution in DNA technology has altered completely the understanding of genetic disease and the practice of our profession. The favorable socio-economic development in Oman in the past 25 years has been translated into superior health care services. As infectious diseases and malnutrition disappeared, genetic diseases now account for an increasing proportion of death, handicap and disability. Rare or 'orphan' diseases, defined as those effecting fewer than 1% of people. Over 6, 000 rare orphan diseases have been documented, with the genetic basis for many now well understood. Genetic testing for orphan diseases has not been considered practical because of the rarity for orphan diseases is largely not available, mainly being restricted to research laboratories working on the involved gene or disease. The goal is to develop testing for orphan diseases. The current experience has demonstruted the value of collaburation between a research and clinicians in controlling orphan disease. Genetic disease registration in The Sultanate of Oman had provided the evidence that orphan diseases are the major contributors to mental and physical disabilities. The common types of orphan diseases and the prospects of their prevention discussed. An important challenge for the years to come is the appropriateness of using genetic information in disease prevention, the fundamental mission of public health.

Experience in Cytogenetic Unit in Zagazig University - Egypt

Zewar M.Mohga ; Abdel Maboud Nashwa ; Zidan A. Amal ; El Sebaey Azza; El Said Fatheya
Clinical Pathology Department, Faculty of Medicine.

Six hundred patients referred to cytogenetic unit at Zagazig University for chromosomal analysis from the period of 1997 to 2002 were the material of our study. They were 280 (47 %) clinically suspected cases complaining of Down and Turner syndromes manifestations, 220 (37 %) couples complaining of repeated habitual abortions and 100 (17%) leukemic and breast cancer patients.

The aim of the present work is to detect the percent of chromosomal abnormalities and to differentiate types of abnormalities in these cases. Convential cytogenetic studies in the form of G-banding, sister chromatin study and FISH techniques were performed in this study.

Our results revealed that chromosomal abnormalities was detected in 62% of suspected down and turner cases, 5 % of couple cases and 58 % of leukemic and cancer cases. Sex chromosomes abnormalities was detected in 2.2% of the cases, turner being 1.7%, and Klienfilter 0.3 % with one case of structural abnormality of X chromosomes (t (p p)).The autosomal abnormalities represent 90 % of the cases, Down syndrome was the highest percentage of numerical cases while structural abnormalities represent 5 % of these cases. Numerical abnormalities were detected in 27.3% of all cases, 0.8 % in couples, 25.5 % in suspected down and turner cases and 1% in leukemic and cancer cases. Suspected cases of down and turner syndromes represent the highest percent of numerical abnormalities. Structural abnormalities were detected in 7.3 % of all cases, 3 % in suspected turner and down cases, 1% in couple cases, 3 % in leukemia while it was 80 % in cancer cases.

In conclusion, our study represent meticulous cytogenetic analysis and application of FISH technique for proper counseling.

The Role of Vitamin C in Amelioration of Potassium Bromate Effect on Chromosomes in Albino Rats

Magdy Gawish
Department of Histology, Faculty of Medicine, Zagazig University, Egypt

Potassium bromate is known to be an oxidizing agent that is used not only as a food additive, mainly in the bread-making process, but also as a neutralizer in thioglycate containing hair curling set. It is also used in cosmetics and as a by-product of water disinfection by ozonation. Thirty healthy adult male albino rats were used in this study aiming to investigate the effect of chronic oral administration of potassium bromate on chromosomes and to evaluate the role of vitamin C as an antioxidant. The animals were classified equally into three groups (1, 2 & 3). Those of the first group served as control. The animals of the second group received 30 mg/kg (1/10 of maximum tolerated dose) of potassium bromate orally once daily for 6 months. The animals of the third group received potassium bromate as in the second group concomitant with vitamin C in a dose of 20 mg orally (double therapeutic dose). Cytogenetic study of the bone marrow cells of the animals treated by potassium bromate showed only marked chromosomal aberrations which were statistically highly significant when compared with those of the control group. Administration of vitamin C concomitantly with potassium bromate revealed marked improvement of the chromosomal aberrations which were statistically highly significant when compared with those treated by potassium bromate only. In conclusion, chronic potassium bromate produced chromosomal aberrations. Vitamin C produced improvement in these aberrations. So, it is advisable to take vitamin C as an antioxidant for protection against the cytotoxic effects induced by food additives such as potassium bromate.

Sister Chromatid Exchanges in Lymohocytes of Severe Chemical Injuries Exposed to Sulphur Mustard Gas During Iran-Iraq Conflict

Mehdi Shafa*, Mahmood Khazab**, Yoosef Seyedena*
* Shahid Beheshti University, Department of Biology, Tehran, Iran. ** Genetic Research Center, Razi Institute, Hesarak, Karaj, Iran.

Shahid Beheshti University, Faculty of Basic Sciences, Genetic Department, Tehran, and Iran Razi Vaccine Institute, Hesarak, Karaj, Iran

Sulfur mustard [HD] is a potent alkylating agent with mutagenic properties. It has been widely used in Iran-Iraq conflict.This study assessed the impact of this agent on the frequencies of sister chromatid exchanges (SCE,) in the peripheral lymphocytes of severely injured Iranian combatants.Twenty five patients with severe lung and eye injuries and ten control subjects were included in the study. The subjects of control group were healthy volunteers matched for sex and age. The lymphocytes were cultured with conventional culture methods. At the end of the culture period and 48h prior the harvesting 20 µg/ml 5-bromo-2-deoxyuridine was added into the medium.Harvested cells were stained with Hoechst 33258, illuminated, and restained with Giemsa. Totally, 25 well - spread metaphases were scored for each sample. In this study, we found that the SCE frequencies of peripheral lymphocytes of patients were significantly higher than the control. ($p > 0.01$).Since the elevation of SCEs rate has been proven in the cancer and malignant disorders, study this factor in severe chemical injuries may give us a good prognosis for evaluating malignancies in these patients.

Keywords: Sulfur mustard; Sister Chromatid Exchange

Cytogenetic and Hematologic Investigation of Severe Chemical Injuries Exposed to Mustard Gas during Iran-Iraq Conflict

Shafa Shariat Panahi M*; Khazab M**; Seyedena Y*
*Faculty of Sciences, Department of biology, Shahid Beheshti University, Tehran, Iran
**Genetic Research Center, Razi Institute, Hesarak, Karaj, Iran

Sulfur mustard (HD) is a potent alkylating agent with mutagenic properties.It has been widely used in Iran-Iraq conflict.This study assessed the impact of this agent on the hematologic factors and chromosomal aberrations (CA) in the peripheral blood of severely injured Iranian combatants.

Twenty fives patients with severe lung and eye injuries and ten control subjects were included in the study. The subjects of control group were healthy volunteers matched for sex and age. The lymphocytes were cultured with conventional culture methods. Hematologic factors including CBC, Platelets, Blood index and Smears were studied. Twenty-five well-spread metaphases were also investigated for any CA and breakages for each sample. Two groups were compared with statistic methods including t-test and Chi-square.

In this study, we found that the chromosomal aberrations and breakages of peripheral lymphocytes of patients were significantly higher than the control ($p<0.001$).We also found that the mean Hb, Hct, WBC, Platelets, MCV and lymphocytes in two groups were different($p<0.001$).It seems that the study of CA in accompany with blood factors may be used as a good prognosis for malignancies and early treatment of severe chemical injuries.

Alpha-2 Polyadenylation Signal Mutation in Bahrain: A Genotype/Phenotype Correlations

Al Momen NJ, Al Arrayed SS
Institution: Genetic Department, Salmaniya Medical Complex, Manama, Bahrain

Alpha thalassemia is one of the commonest genetic disorders of the red blood cells in Bahrain. The most severe alpha thalassemia phenotype reported in Bahrain is the so-called hemoglobin H (HbH) disease with the predominant features of anemia (Hb 8-9 g/dL) and jaundice. Another hall mark of this disease is existence of HbH inclusion bodies on the red blood cells in the peripheral blood smear. On the genotype level, the main molecular defect causing HbH disease in Bahrain is homozygosity of the Saudi type polyadenylation signal mutation in the alpha-2 globin gene (AATAAA→AATAAG). During our study of alpha thalassemia in Bahrain several genotype combinations of the polyadenylation signal mutation with other alpha thal determinants were encountered. The various genotype combinations of the polyadenylation signal mutation in correlation with their phenotype would be presented in this report. Moreover, the molecular etiology of the polyadenylation signal mutation in causing such a severe phenotypic presentation, as in HbH disease, would be presented with further emphasis on previously reported studies. Understanding of these genotype and phenotype correlations might be used as a prototype to delinate any other genotype/phenotype complex interactions for other genetic disorders. Finally, the principal rule of the genetic laboratory in addressing such investigations would be highlighted in this report.

Study of folic acid pathway genes alteration as maternal risk factor of Down syndrome

Ahmad Aleysin, Mohammad Ghazanfari, Faezeh Jahanshad
National Research Center for Genetic Engineering and Biotechnology, Tehran, Iran

Chromosome 21 nondisjunction event is responsible for more than 95% of Down syndrome. The etiology of nondisjunction has not been very well described however some factors such as folate metabolism is recently considered as an important factor on chromosome nondisjunction event. This study was to evaluate the impact of abnormal folate metabolism induced by folate pathway genes mutation among mother of children with Down syndrome compared to the normal control mother. Common mutations of C677T and A1298C of MTHFR gene are reported to decrease folic acid level in about 30 to 70 percents in heterozygous and homozygous forms respectively in blood samples. To evaluate the impact of these mutations, the parental origin of Down syndrome of all causes were determined at first place and mothers have been categorized into two groups according to the maternal or paternal origin of chromosome 21 trisomy. This study is unique compared to previous studies, to determined paternally originated Down patients at the first place to increase accuracy of obtaining results. Folic acid gene alterations were studied in 50 Down families and 60 normal controls. Till now our results have showed 4 times higher risk of having Down syndrome child among mothers caring these mutations than normal control mothers or mothers with paternally origin of Down syndrome in Iranian Down syndrome cases.

Opsismodysplasia with Additional Finding of Genetic Hypophosphatemia - Report Confirming Autosomal Recessive Inheritance

Naveed M.; AlHussain, M.A.; Eldin, M.S.
Genetic & Thalassemia Center, Al-Wasl Hospital, Dubai, UAE

Opsismodysplasia is a very rare form of Chondrodysplasia secondary to chondroosseous transformation defect. Only about 20 cases have been described. The diagnosis of Opsismodysplasia can be made at birth based on the association of clinical and radiological manifestations. Rhizomelic micromelia, hypotonia, very retarded bone maturation, thin lamellar vertebral bodies and facial dysmorphism are characteristic findings in these patients. The major complications of this condition are severe dwarfism and susceptibility to respiratory infections due to narrow chest and muscular hypotonia. Based on two observations in which parents were first cousins and a report from two sibs of different sex it is suggested that the most likely mode of inheritance is autosomal recessive. Here we report a female child with Opsismodysplasia, born to consanguineous parents of South Indian origin. The clinical and radiological findings in this case confirm the diagnosis of Opsismodysplasia. The additional finding in our case is hypophosphatemia. This is the second report in literature with an affected female and supports the previous observation of autosomal recessive inheritance of Opsismodysplasia.

Recessive Robinow Syndrome with Additional Features

Naveed M.; AlHussain, M.A.; Eldin, M.S
Genetic & Thalassemia Center, Al-Wasl Hospital, Dubai, UAE

Robinow syndrome is an extremely rare inherited disorder characterized by mesomelic brachymelia, hemivertebrae, dysmorphic facies, genital hypoplasia and moderate short stature. In some cases, Robinow syndrome has autosomal dominant inheritance; in other cases, the disorder may have an autosomal recessive mode of inheritance. The autosomal recessive form of Robinow syndrome (MIM 268310) is a severe skeletal dysplasia with segmental defects of the spine, brachydactyly and a dysmorphic facial appearance. Here we report on the account of its rarity and additional features, a five-year follow-up of a male child with Robinow syndrome born to consanguineous parents of Pakistani origin.

Fourier Transformed Infrared Spectroscopy (FTIR) as a Diagnostic Tool in Cystinuria

Das, N.S.; Hasan Sanad
Clinical Biochemistry Section, Department of Pathology, Salmaniya Medical Complex, Kingom of Bahrain

Cystinuria is an autosomal recessive disease characterized by renal and intestinal transport defects of dibasic amino acids including cystine, ornithine, lysine, and arginine.

Seven patients of incompletely recessive cystinuria with urolithiasis were diagnosed by infrared spectroscopy. The powdered calculus / dried urinary sediment treated as KBR pellets were analyzed by FTIR spectroscopy. The process identified the presence of cystine at low cost and high reliability.

Among the seven patients six patients belong to Kingdom of Bahrain and other belong to Tunisia. M:F ratio among the patients was 3:4. The methodology and FTIR spectra of cystine on KBR pellets of powdered calculus or dried urine sediment are presented.

Molecular Basis of β-Thalassemia in the United Arab Emirates

Bayoal, E., Taleb, M., Yousuf, I., Garcia, G., Ridha, A.
Genetics Department, Pathology Services, Al-Wasl Hospital, Department of Health and Medical Services, Dubai, United Arab Emirates

Hemoglobinopathies are a major public health problem in the UAE. We carried out population screening on 444 newborns and 1147 young women of UAE nationality. The incidence is estimated to be 8.3% in both populations. b-thal and sickle cell disease (SCD) are the most prevalent b-globin gene defects. 750 thalassemia patients are on regular blood transfusions and iron chelation therapy in our center, 46 % of whom are UAE nationals. The molecular characterization of 193 UAE national b-thal patients through various DNA techniques revealed a significant number of homozygotes, reflecting a high degree of consanguinity (estimated at 54% with 30% first cousin marriages). Our molecular studies depict that 66.1% of b-thal patients were homozygous for the IVS-I-5 (G®C), followed equally by Cd 8/9 (+G)(7.9%), HbS gene (7.9%), Cd 39 (C®T)(4.7%) and -25 bp del (3.9%) totalling more than a dozen of homozygous mutations.
The most common b-thal mutation was the IVS-I-5 (G®C)(56.2%) followed by Hb S gene (11.4%), -25 bp del (7.8%) and Cd 8/9(+G)(5.2%). UAE appears to have a diverse spectrum of b-thal mutations, in contrast to Saudi Arabia, Oman, Kuwait and Iran. This is due to genetic influence from India, Baluchistan and Arabian Peninsula. The UAE represents a heterogenous mixture of genes from all regions in the Middle East. To date, 49 b-thal mutations have been identified making the UAE arguably the most heterogeneous population in the world. More b-thal alleles are known in the UAE than all the Mediterranean countries put together.
Data presented here serves as vital prerequisite for establishing prenatal diagnosis in prospective couples to prevent completely the birth of thalassemic babies in the UAE.

Molecular Studies on HbH Disease in the United Arab Emirates

Baysal, E., Taleb, M., Aoudah, H., Yousuf, I., Garcia, G.; Ridha, A.
Genetics Department, Pathology Services, Al-Wasl Hospital, Department of Health and Medical Services, Dubai, United Arab Emirates.

HbH disease is a moderately severe hemolytic anemia with microcytosis, hypochromia, low HbA_2 and HbF levels, and varying quantities of HbH (β_4) (2-30%). Most of the HbH syndromes were thought to be caused by the deletion or inactivation of 3 of the 4 α-globin genes. However, numerous reports published in the last decade demonstrated an increasing number of non-deletional (α^T) α-thalassemia (α-thal) as the molecular basis of many HbH syndromes, particularly in the Gulf region.

A newborn screening survey among the UAE National babies, carried out in our center demonstrated that the frequency of α-thal in the UAE is 49.8 %, one of the highest in the world. Using PCR-based strategies, allele-specific oligonucleotide (ASO) hybridization, manual and automated DNA sequencing, we have defined the genotype of 39 patients with HbH disease in the UAE. Of these, 28 were UAE Nationals, 5 Omanis, 3 Sudanese, 2 Thai and 1 Pakistani. Nine UAE nationals and 4 Omani patients were homozygous for the poly A-1 mutation ($\alpha^{PA-1}\alpha/\alpha^{PA-1}\alpha$) characterized by the AATAAA→G substitution in the α_2 globin gene. Moreover, 11 UAE national patients were diagnosed with the $-\alpha^{3.7}/\alpha^{PA-1}$ α genotype. In a large UAE national family, 2 individuals were homozygous for the Hb Constant Spring ($\alpha^{CS}\alpha/\alpha^{CS}\alpha$) which affects the termination codon (α142 TAA→CAA) of the α_2-globin gene and 3 had the $-\alpha3.7/\alpha^{CS}\alpha$ genotype. Hb CS accounted for 11.5% of the chromosomes. In addition, 9% of the α-thal chromosomes had the -5 nucleotide deletion ($\alpha^{5nt\,del}$), at the splice junction between exon 1 and intron 1. Two siblings of Thai origin had the --SEA deletion and one boy from Oman was characterized with the --MED-I deletion. Both of these deletions were characterized through direct PCR-based approaches. Eight different α-thal genotypes were identified, 4 deletional and 4 non-deletional (α^T).

Almost half (47.4%) of the 78 α-thal chromosomes had the poly A-1 mutation. 13 out of 39 patients (30%) were homozygous for the mutation, $\alpha^{PA-1}\alpha/\alpha^{PA-1}\alpha$, with moderate clinical severity. The data presented here demonstrate a considerable heterogeneity of α-thal in the UAE. The genotype /phenotype correlation suggests that HbH disease in the UAE has, in general, a moderate presentation. The molecular characterization of the patients with HbH disease may be clinically useful for the prediction of the clinical outcome, correct management and for providing genetic counseling to patients and families in the UAE.

Clinical Heterogeneity in One Family with Two Cases of L-2-hyoroxyglutaric Aciduria

Naveed M., Al-Hashmi, H; Taleb M
Genetics Department, Al-Wasl Hospital, Department of Health and Medical Services

L-2-hydroxyglutaric aciduria (MIM 236792) is an extremely rare autosomal recessive disorder. It is characterized by mental retardation, motor delay, cerebellar dysfunction, macrocephaly and characteristic MRI findings (subcortical leukoencephalopathy, cerebellar atrophy and signal changes in the putamina and dentate nuclei). Elevated levels of L-2-hydroxyglutaric acid in urine, plasma, and CSF confirm the diagnosis. No specific biochemical function or catabolic pathway involving L-2-hydroxyglutaric acid is known in mammals, including humans. Here we report two patients in one family born to consanguineous parents of Indian origin with variable onset and severity of neurological disease. The clinical, radiological and GC-MS findings in these two patients were consistent with diagnosis of very rare case of L-2-hydroxyglutaric aciduria.

Inherited Metabolic Disorders in the United Arab Emirates

Taleb, M., Al-Hashimi, H, Al-Hammadi, Z., Al-Muwaffaq, N and Ridha, A.
Genetics Department, Pathology Services, Al-Wasl Hospital, Department of Health and Medical
Services, Dubai, United Arab Emirates

In order to understand the prevalence of inborn errors of metabolism in the United Arab Emirates, we have analyzed all samples from suspected patients with metabolic disorders referred from various hospitals in United Arab Emirates. Over a period of five years, a total of 275 samples were analyzed with Gas Chromatography/Mass Spectrometry (GCMS) technique for the detection of organic acid disorders. We have diagnosed 20 patients with various types of organic aciduirias including methylmalonic academia (4 cases), propionic acidemia (2 cases), 3-OH-3-methyl glutaric aciduirias (3 cases), ethylmalonic aciduria, glutaric aciduria type I, 1,-2-OH glutaric aciduirias (2 cases), ketothiolade deficiency (2 cases), pyroglutamic aciduria, hyperglyceruluria (2 cases), biotinidase deficiency and maple syrup urine disease. With spectrofluorophotometric analysis, we have diagnosed GM1 gangliosidosis (5 cases), fucosidosis, manosidosis, Morteaux-Lamy syndrome, Gaucher's disease (2 cases), Tay-Such's disease (4 cases), Sandhoff disease (4 cases), Pompe's disease (3 cases), Krabbe's disease (2 cases). Our observations suggest that a variety of inherited metabolic disorders are common in the local population. In view of overlapping clinical signs and symptoms in early life, this study highlights a need for continued awareness among the pediatric professionals with regard to prevalence of such conditions in the community.

Rare Clinical Presentation of Ethylmalonic Adipicaciduria

Naveed M.; Al-Hashmi, H.; Taleb, M
Genetic & Thalassemia Center, Al-Wasl Hospital, Department of Health & Medical Services, Dubai, UAE

Ethylmalonic Adipicaciduria, also referred as Glutaricaciduria IIB (Adult Form of Glutaricaciduria II), is a very rare form of organic acidemias, presenting mainly during adulthood. It is characterized by metabolic acidosis and hypoglycemia without ketosis, episodic vomiting, hepatomegaly and proximal myopathy. It is caused by a deficiency of the enzyme multiple acyl- CoA dehydrogenase. Its course and age at presentation is extremely variable. Here we report a rare clinical presentation in a ninth-month-old Egyptian girl with delayed milestones, dysmorphic features, metabolic acidosis, macrocephaly, intractable convulsions, hypotonia, hepatosplenomegaly and ventricular septal defect. The GC-MS analysis confirmed the diagnosis of Ethylmalonic Adipicaciduria.

National Program for the Prevention of Thalassemia in Iran

Zeinali, S.*, Nafissi, Z.**
*Department of Biotechnology, Pasteur Institute of Iran, Pasteur St., Tehran, Iran. And Medical Genetics Lab of Dr. Zeinali. **Department of English Language, Az-Zahra University, Vanak, Tehran, Iran

Thalassemia is the major hereditary blood disorder in Iran. Some 26000 affected individuals receive medical care. The carriership in some parts of the country exceeds 10%. Each year millions of dollars are spent for providing blood and iron chelating drug for these patients. Extra money and resources have to be allocated for them for hospitalization and other cares needed.
Iranian Thalassemia Society with the help of medical doctors and national media have played important role to raise awareness among people, politicians, medical staffs as well as the scientists in favor of these patients and their families. β- thalassemia had plaid a central role for creation of organizations for special diseases such as Special Disease Section within the Ministry of Health and also Charity Foundation for Special Diseases. The later one is a non-profitable NGO which has done a lot for β-thalassemia, hemophilia, etc.
Public awareness plus increasing financial burden for providing medical care for these patients has forced the government to include premarital screening for β-thalassemia as a compulsory test in its Primary Health Care system. With a very effective and wide spread network of Primary Health Care System (PHCS), the program has proven a success even in its initial implementation just in less than 5 years ago. In the program every couple who wants to get married are referred to one of PHC clinic for CBC blood test. If the test shows any sign of anemia then the other couple is tested. When both are confirmed as being carriers, then they are referred to counselor. The couples are usually given a thorough counseling about β-thalassemia and all the possible outcomes if they decide to marry and have children. If the couple still insist in marrying they are given a certificate for the marriage registry office and at same time are refereed to one of several medical genetics clinic for preliminary prenatal diagnosis (PND) tests. If laboratory tests at the PHC clinic are still unclear then the couple are referred to Pasteur Institute of Iran for globin chain synthesis, α-globin mutation analysis and β-globin gene sequencing.
The above program has evolved alongside with several other major developments in the country. In 1992 PND for β-thalassemia become available in Iran by a private genetic clinic by sending blood and amniotic fluid samples to the UK. In early 1994 it became available at Pasteur Institute of Iran. In 1996 our grand Ayatollah Khamenei gave the permission for legal abortion of affected fetuses. This accelerated the β-thalassemia prevention program in the country. The decision was then taken to expand PND centers by tech-transfer strategy from well-trained and experienced centers to staffs from other medical universities in major cities having high incidences of β-thalassemiain their region. This task was helped by extra financial help to those universities and the training centers.
Alongside with this program, the Ministry of Health decided to create a network between governmental and private PND centers. Two national reference centers were selected to aid and oversee the activities of less experienced governmental or private centers.
The Iranian Legal Medicine Organization ordered all of its branches nationwide to permit legal abortion for β-thalassemia upon approval by one of the recognized PND centers.
Until year 2000, the insurance companies were reluctant to pay for PND tests but in late 2000 the major insurance companies accepted to pay the costs. In mid 2001 one of the main Insurance Company decided to pay 100% of all PND costs including CVS.
We are now reorganizing the existing system and are improving the services given. Several new centers are in the process of opening up and their manpower is receiving training at the Pasteur Institute of Iran (Centers in Shiraz and Zahedan now functional).
Another decision was to ask all the members to report each PND tests done by each network member to Genetics and Cancer Prevention Office, Ministry of Health.
Iranian experience in providing medical care, prenatal screening and PND service is something to learn from and implement in other countries particularly in neighboring countries. We are willing to provide assistance and training for other countries in the region in this regard.

Report on the out come of 266 prenatal diagnosis of β-thalassemia in a private clinic in Iran since 2000

M. Masrouri; V. Lotfi; P. Fouladi; M. Zeinali; Mo. Zeinali; R. Alipour; Z. Jamali; S. Azadeh; S. Zeinali.
Medical Genetics Lab of Dr. Zeinali

Thalassemia is the major single gene disorder in Iran. Carrier frequency is high in some parts of the country, therefore, premarital screening has become compulsory by law. When two carrier couple decide to marry and have child then prenatal diagnosis is offered to them at two stages: pre pregnancy molecular analysis and during pregnancy.
In our laboratory (established in 2000) we have admitted more than 530 couples for prenatal diagnosis of β-thalassemia so far.
For more than half of the cases prenatal diagnosis (PND) has been done to the final stage. For PND we use direct mutation analysis (using ARMS/PCR) or indirect method using RFLP. Below is the result of our methods and the percentage of informativeness RFLPs for PND
Total PND completed: 166 PNDs
In 74.7% ARMS alone was informative and in 55.9% RFLP alone was informative. The combination of these two are as follow:
-ARMS 100% & RFLP 100% (43.2%)
-ARMS 100% & RFLP 50% (24%)
-ARMS 100% & RFLP 0% (7.5%)
-ARMS 50% & RFLP 100% (7.1%)
-ARMS 0% & RFLP 100% (5.6%)
-ARMS 50% & RFLP 50% (7.5%)
-ARMS 50% & RFLP 0% (2.6%)
-ARMS 0% & RFLP 50% (1.8%)
Over all PND could be performed in 98% of cases and in 2% nothing could be done for the family during pregnancy particularly if the couple had come for PND during pregnancy. This number would be 96.5% for couples coming for stage one of PND. For this we strongly recommend that families should request for PND before pregnancy to avoid difficulties. Our PND results showed that 51.3% of cases were heterozygote, 25.2% homozygote normal and 23% homozygote affected.

We hope our results could help others planning to perform PND in other countries.

Aims and objectives in creation of National Molecular Medicine Network in IRAN

S. Zeinali,
National Molecular Medicine Network, Biotechnology Research Center, Pasteur Institute of Iran

The network now comprises twenty research establishments in the country. Part or all of these centers' activities are research based, particularly in molecular medicine. The aim were as follow: to enhance the integration of molecular medicine in the medical practice, to enhance the level of research in this field, to enhance the scientific collaboration, to educate young physicians in this field, to set research priorities, avoid unnecessary or duplicate activities, to promote public awareness on new technologies and etc. In mid 2001 the strategic plan was finalized and the plan has put forward a list of activities that must be implemented. Our immediate plans are to organize several purposely designed educational programs, both theoretical and practical (as workshops), to initiate the establishment of several necessary infrastructures for research and development such as HLA bank, DNA repository, stem cell bank, cell bank, Gene therapy (basics), Transgenics, Bioinformatics.

We are hoping this new initiative will enhance Iran's capabilities for better utilization of new advances in this field in the world.

Spectrum of beta globin gene mutations in west of Iran bordering Turkey, Iraq

P. Fouladi, M. Masrouri, V. Lotfi, M. Zeinali, Mo. Zeinali, R. Allpour, Z. Jamali, S. Azadeh, S. Zeinali
Medical Genetics Lab of Dr. Zeinali

Beta thalassemia is a major blood disorder in most Middle Eastern countries including Turkey, Iraq and Iran. Molecular analysis of β-globin gene mutation will help to plan for prenatal diagnosis (PND) and for population genetics studies.
We studied 410 chromosomes from affected and carriers of β-thalassemia from 6 provinces in the west of Iran namely West Azarbayjan (Az) (in the northwest), Kurdistan (Ku), Kermanshah (Ke), Ilam (I) (in the west) and Khouzistan (Kh) (in the south). We analyzed DNA samples for known mutations using ARMS/PCR method. Below are the number of chromosomes for each region, type and percentage of each mutation and number of unknown cases (e= codon; I = IVSInt; nd = not detected; del = deletion and II = IVSIInt).
Az: Total chromosome 134, of which 87/27% were known and 12/8% unknown: C5 (2/98%), Fr8/9(12/68%), C22 (nd), C30 (0/74), I-I(17/81), I-5(4/47%), I-6(1/49%), I-110(15/67%), -25del(nd), C36-37(5/22), C39(6/66), C44(nd), II-I(23/13%), II-745(2/98).
Ke: Total chromosome 45, of which 80% known and 20% unknown:
C5 (nd), Fr8/9(13/33%), C22 (2/22), C30 (nd), I-I(nd), I-5(2/27%), I-6(nd), I-110(nd), -25del(nd),
C36-37(15/55%), C39(8/88%), C44(nd), II-I(37/77%), II-745(nd).
Ku: Total no. of chromosome 54, of which 87% known and 13% unknown: C5 (1/85%), Fr8/9(18.51%), C22 (nd), C30 (nd), I-I(27/77%), I-5(3/70%), I-6(1/85%), I-110(9/25%), -25del(nd), C36-37 (15/55%), C39 (nd), C44 (3/70%), II-I (14.81%), II-745 (nd).
Kh: Total no. of chromosome 162, of which 94% known and 6% unknown:
C5 (1.9%), Fr8/9(11%), C22 (1.9%), C30 (1.9%), I-I(7.1%), I-5(36.3%), I-6(1.2%), I-110(8.4%),
-25del(1.2%), C36-37 (4.9%), C39 (0.6%), C44 (7.1%), II-I (14.9%), II-745 (1.2%).
I: Total no. of chromosome15, of which 100% known:
C5 (nd), Fr8/9(2/66%), C22 (nd), C30 (nd), I-I(6/66%), I-5(nd), I-6(nd), I-110(20%),
-25del(nd), C36-37 (6/66%), C39 (nd), C44 (nd), II-I (40%), II-745 (nd).
Our results showed that IVSIInt1 has the highest frequency in these regions except in Kurdistan. It is as high as 40% in Ilam to as low as 15% in Kurdistan and Khouzistan. IVSInt1 is considered to be a rare mutation in Iran but it is present with 28% in Kurdistan and 18% in Azarbayjan but not detected in Kermanshah. This is of particular interest since Kurdistan and Kermanshah are neighbors. This may indicate that they are more distinct by race than geography. These types of differences in beta-globin mutation can be seen when mutation distribution are compared across the country.

Spectrum of Genetic Disorders in Patients Referred to the Genetics Department of R.M.C. At Saudi Arabia: Cytogenetic and Clinical Experience for 8 Years

Mona O. El Ruby
Human Genetics Department, National Research Center, Cairo, Egypt

A genetic component is evident in the etiology of most human diseases. Genetic disorders pose a major health problem in the world in developed countries they represent the single largest cause of infant mortality loss while they are the 3rd – 5th cause of perinatal mortality in developing countries. Major efforts are directed towards lightening the burden of these disorders on patients, family and on the community at large. In this study over a period of 8 years, 5397 cases were examined and studied for the presence of chromosomal aberrations and / or identifiable malformations at the Human Genetics Department (HGD) of the Riyadh Medical Complex (RMC). Of the total 5397 karyotypes performed, genetic component was ascertained in 37.5% of the cases. Chromosomal abnormalities were detected in (22.1%). Down syndrome represented the highest chromosomal aberrations (15.4% of the total referred cases and 69.6% of the chromosomal aberrations) followed by trisomy 18 (1.1% of the total and 4.8% of chromosomal aberrations). Klinefilter syndrome represented the 3rd frequent chromosomal aberrations (1%, 4.4%) while Turner syndrome was diagnosed in 0.8% of the total cases and 4% of the chromosomal aberrations. Non chromosomal genetic disorders were ascertained in 9.61% of the cases; of which single gene inheritance was diagnosed in 7.26% (autosomal recessive accounted for 65.3% of the single gene disorders), multifactorial was diagnosed in 1.25% and sporadic cases in 1.1% of the cases. 5.7 % of the cases were unclassified genetic entities. Base line information on the frequency of genetic disorders and chromosomal aberrations, their classification and prevalence are the primary objectives of this study. The high frequency of genetic disorders at the Middle East level emphasizes the prerequisites for national surveys and further studies to evaluate the influence of consanguinity, dietary, health awareness, social, cultural and other environmental factors on the prevalence of congenital and genetic diseases in different communities.

Frequencies of the Beta-Globin Single Nucleotide Polymorphism (Snp): T to C Transition at Sixth Nucleotide of Exon 1

Zeinali, M. Moghadam, Z. Zeinali, S
Biotechnology Dept, Pasture Institute of Iran

Analysis of the association between genotype and disease phenotype in humans is at a critical point. Hundreds of rare disease showing mendelian patterns of inheritance in humans have been describes in the medical literature. The focus of variation analysis is now shifting from the identification of new markers to their typing in populations, and novel typing strategies are rapidly emerging. More than a million genetic markers in the form of single nucleotide polymorphisms are now available for use in genotype-phenotype studies in humans.

In this study we analysed 200 chromosomes from 100 patients who had come for prenatal diagnosis (PND). We screened their DNA samples for SNP at sixth nucleotide of exon 1 in ?-globin gene. We found that three genotype exist in our samples: In this study 200 chromosomes were screened for the two form of SNP T/C. 80 chromosomes were T and 120 chromosomes are C or C/T alleles are 15%, T/T alleles are 32.5% and C/C alleles are 52.5%.

Our results showed that though this polymorphisms is not highly informative but still can be use for PND.

Impact of Neonatal Size on Cardiac Dimensions and Functions in Infants of Diabetic Mothers

Nevein H. Abou El Soud*; Eitedal M. Dawoud*; Abd Allah El Foully**
*Basic Medical Sciences and Biochemistry Department, National Research Center, Cairo
**Abou El Rich Children Hospital, Cairo University

To determine the influence of neonatal size on various cardiac size dimensions (aortic root, left atrial diameter, interventricular septum thickness, right ventricle, pulmonary artery) and functions (left ventricular posterior wall, left ventricular end diastolic dimension, left ventricular end systolic dimension, end diastolic volume, end systolic volume, stroke volume, shortening fraction percentage, ejection fraction percentage) in infants of diabetic mothers (IDMs), we examined two dimensional –M-mode echocardiographic measurements in 46 IDMs [30 large for gestational age (LGA) and 16 average for gestational age (AGA)], together with 10 age and sex, AGA matched controls. When LGA were compared with AGA infants in the diabetic group, no significant differences in cardiac size dimensions were observed, in contrast to cardiac function related increase for LGA (ejection fraction percentage: 74.3± 8.5 vs 64.5±13.2, $p = 0.004$ and shortening fraction percentage: 38.6 ± 5.8 vs 33.6±7.2, $p = 0.01$).These data suggest that the increase in ventricular contractility and performance in LGA represent the effect of macrosomia rather than the effect of diabetes. While in comparing IDMs (LGA or AGA) with control, hypertrophy of interventricular septum and right ventricle outflow tract was observed, but no functional differences were detected. This suggests that the increase in cardiac size represents mainly the effect of diabetes and insulin disturbances rather than macrosomia. Body size assessed by body weight in relation to gestational age was correlated with cardiac size and function in LGA and with cardiac function only in AGA. While, when proportional body size (ponderal index) was used no relation was found in LGA group. Ejection fraction percentage was the best cardiac measure related to body size in LGA-IDMs, using multiple regression analysis with $p = 0.03$. We conclude that large neonatal body size in IDMs did affect the cardiac echo dimensions and functions leading to myocardial hypertrophy (involving right ventricular wall thickness, pulmonary artery and interventricular septum thickness), in addition to hypercontractility of left ventricle (judged by the increase in ejection fraction percentage and shortening fraction percentage). This could precipitate to heart failure; adding to the hazards of macrosomia.

Key wards: Diabetes- Echocardiography-Neonatal size.

Comparison of Intracytoplasmic Sperm Injection Outcome, Using Testicular Sperm Versus Seminal Sperm: Future prospects for Exclusion of Abnormal Embryos by Detection of Aneuploidy

Mariam A. Ghuloom
CMMS, Arabian Gulf University and IVF/Infertility Centre, BDF Hospital, Bahrain

Intracytoplasmic sperm injection (ICSI) has become the most effective treatment of male factor infertility since 1992. Although the fertilization rates have shown significant improvement by this technique over the last 10 years, the current implantation rates still remain lower than 35%. This report presents the outcome of ICSI procedures in two groups of infertile patient due to suboptimal semen parameters. In group I, 46 cases undergone ICSI with ejaculatory spermatozoa and group II, 42 cases with testicular spermatozoa. The aim was to analyze our compiled data retrospectively in terms of number of oocytes retrieved/puncture, fertilization rate/trial, morphological evaluation of embryos obtained and subclinical/clinical pregnancy success rates. Although there were no statistical significance differences in most of the above parameters between the two groups, higher clinical pregnancy rate was achieved in ICSI group with ejaculatory sperms. The incidence of early pregnancy loss was twice as high in group II (50%) than that observed in group I (23.5). Future prospects in investigating risk factors resulting in early embryo loss are considered. Detection and exclusion of aneuploid embryos by pre-implantation genetic diagnosis may have a great impact on improving ICSI implantation rate and pregnancy outcome.

Chromosome Damage in Nickel-Chrome Electroplaters

T. Sharma
Al Mahd Day Boarding School, Bahrain

Electroplating occupies a prominent position in the industrial world. Metal, plastic and rubber parts are plated to prevent rusting and corrosion, for appearance to reduce electrical contact, for resistance, as a base for soldering operations, to provide electrical insulation and to improve wearability. The type of electroplaters in the present study are the ones dealing with the Nickel-Chrome electroplating. Heparinized venous blood samples were collected from 68 electroplaters and equal number of matched controls. Genotoxicity was investigated by assaying mitotic index (MI), chromosome aberrations (CA), sister chromatid exchanges (SCE) and satellite associations (SA) in short term lymphocytes cultures. The mean frequency of MI was 5.77 ± 1.77, CA 3.64, SCE 6.40 ± 0.92, and SA 9.14, in the exposed group as compared to controls in which the mean frequencies of these parameters were 4.18 ± 0.85, 0.86, 4.18 ± 0.87 and 4.08 respectively. Smoking and alcohol consumption were found to be the confounding factors. Significant effect of period of exposure was observed. The present study clearly indicates that there is an increased risk of cytogenetic damage and various respiratory tract diseases, like bronchial asthma and intranasal ulceration.

Work Place Pollution Hazards: Cytogenetic Damage to Spray Painters Exposed to Organic Solvents

V. Kumar
Al Mahd Day Boarding School, Bahrain

Spray painting is a very common occupation throughout the world. It is used in various industries. Spray painters are exposed to a large range of organic solvents which pollute the work environment. Genotoxicity of organic solvents was investigating by assaying Mitotic Index (MI), Chromosomal Aberrations (CA), Sister Chromatid Exchange (SCE) and Satellite Associations (SA) in short terms lymphocyte cultures of 62 spray painters and 62 matched controls. The mean frequency of MI was 6.78 ± 1.55, CA 3.88, SCE 7.39 ± 0.65 and SA 12.28 ± 1.64 in the exposed workers as compared to controls in which the mean frequencies of these parameters were 4.32 ± 0.89, 0.94, 4.18 ± 0.67 and 3.59 ± 0.15 respectively. The differences in the values are statistically significant ($p < 0.05$). Smoking and alcohol consumption were found to be the confounding factors. Significant effect of period was observed. The present study clearly indicates that there is an increased risk of cytogenetic damage and various respiratory tract diseases, skin ailments like dermatitis etc. in the spray painters.

Advanced Laboratory Technology and Premarital Evaluation

Mohammed El Sawy
Paediatric Dept Ain Shams University

The last few decades have seen a dramatic decrease in the importance of environmental causes of ill health in developed countries. As a result, there has been a sharp increase in the relative importance of genetic disorders as causes of mortality and morbidity. Recent studies have shown that about 30% of admissions to, and 40.5% of deaths occurring in, paediatric hospitals are accounted for by children with genetic diseases or congenital malformation. Genetic diseases are almost always serious, are not curable and few are amenable to satisfactory modes of treatment. Thus prevention is of paramount importance. This includes genetic counseling for individuals at risk of having a child with a serious genetic disorder, coupled with prenatal diagnosis. Moreover, neonatal screening is mandatory for early intervention. The advanced laboratory technology and premarital evaluation will be discussed.

An Investigation on Chromosomal Abnormalities and Physiological Study of Mentally Retarded Male Children of Tehran

Bayat B, Sanati M1, Syeddena Y2, Khazab M.
National Research Center of Genetic Engineering and Biotechnology. Shahid Beheshti University. Tehran, Iran

Structural and numerical aberration is the major defects in mentally retarded individuals. This Abnormalities may affect physiological properties of individuals e.g. growth, circulation, respiration etc. In this study total number of 63 mentally retarded boys were randomly selected and were studied by cytogenetic and physiologic methods.

About 19% of the affected boys show fragile X ,5% have 21 trisomy, 35% have gap and breaks at least in one of the chromosomes and about 1.5% polyploidy are observed in total metaphase plates.

Physiological results show that weight and total length and the length of forearms of almost all the patients are statically reduced but length of all peduncle are reduced as well. Maximum and minimum blood pressure of all groups was decreased, compared to the normal samples. Hemoglobin's reduction was observed in samples from patients with fragile X syndrome.

Haplotype Analysis of Related ATM Markers Facilitate Prenatal Diagnosis in Iranian Ataxia Telangiectasia Patients

Behnaz Bayat
National Center of Human Genetic and Biotechnology, Tehran

Ataxia Telangiectasia is an autosomal recessive disorder in 1/40000 to 1/100000 in reported populations. There is 25% possibility for having an affected child when parents are carrier for ATM gene mutation. There is no cure available for this disease and prenatal testing is strongly recommended in prevention of this disease.

Although preference method is the direct mutation analysis of ATM gene, but large size of the ATM gene with 63 exons and the large number of possible mutation in patients considerably limit the facibility of mutations analysis as a choice in diagnosis. Indirect method is a better tool when parent are not carrier of founder mutation and pass different mutations to their children. Indirect molecular diagnosis using ATM related molecular markers facilitate prenatal diagnosis of AT children. In this study four molecular markers: D11S2179, D11S1787, D11S535, D11S1343 are genotype in 18 unrelated families from different region of IRAN. Those markers are amplified using extracted sequence primers from Gene Bank with their described PCR conditions. The amplified products were separated using denaturing PAGE gels, and the data were analyzed to detect their pattern of inheritance in each family. In all families segregation of alleles were recording to mandelian inheritance and affected chromosomes were distinguishable.

Molecular Basis of G6PD Deficiency in Bahrain: An Extended Study

Al Momen NJ, Al Arrayed SS
Genetic Department, Salmaniya Medical Complex, Manama, Bahrain

Glucose-6-phosphate dehydrogenase (EC 1.1.1.49; G-6-PD) is the key enzyme of the pentose phosphate pathway and provides NADPH essential for a number of biosynthetic and detoxifying reactions. G6PD deficiency is one of the most common red blood cell abnormalities causing hemolytic anemia induced by ingestion of certain drugs and fava beans. In Bahrain, the G6PD deficiency is quite common with an estimated frequency of 26% of the whole population. In this study we have determined the molecular basis of G6PD deficiency in an extended patient/student sample. A total of 64 deficient subjects (48 males and 16 females) have been studied. By using a PCR-based DGGE and RFLP molecular detection methods, we found that 97% (62/64) of the subjects are bearing the G6PD Mediterranean variant (nt 563 C-T). It is interesting to note that 4 out of the 16 female deficient subjects are merely heterozgotes for the G6PD Med mutation while the others are homozygotes. Apparently effect of random X-chromosome inactivation might be involved in manifesting G6PD deficiency phenotype in these heterozygote females. In conclusion, this extended study confirms our previous observation that G6PD Mediterranean is the main variant causing G6PD deficiency in Bahrain.

Screening for Hypertrophic Cardiomyopathy

Dr. Wafaa Eyaid; Dr. Muayed Al-Zalbag; Doha Al-Nouri
King Fahad Hospital

We report on a Bahraini family with a history of six brothers and their sister who died at very young ages of Hypertrophic Cardiomyopathy (HCM).

HCM is an autosomal dominant disorder that is passed on from one generation to the next. The affected individuals die suddenly at a young age.

On examining this family pedigree, we identified number of individuals who were HCM symptom-free. However, the HCM has variable penetrance i.e individuals can carry the defective gene, but be disease free. Also, HCM is known to have a variable age of onset, which was the case with this family where we found a two month old baby boy to be affected while other members were symptom free till they are in their twenties.

Based on the above, we recommend that DNA testing be performed on all family members with a risk for having the disease. This will help their and subsequent generation ’s clinical management, especially in regard to pre-natal or pre-implantation testing.

Molecular Genetics of Mild Forms of Sickle Cell Disease in Bahrain

Al Momen NJ, Al Arrayed SS
Salmaniya Medical Complex, Manama, Bahrain

In this study we are reporting the molecular basis of mild forms of sickle cell disease in three Bahrainis. Two unrelated female students, presented through Student Screening Project, and a male genetic consultee having a child with sickle cell disease, were recruited for this study. All of the patients were asymptomatic and presented with no anemia at total hemoglobin levels of 12.7, 13.1, and 13.5 g/dl, respectively. HPLC of the whole blood revealed an atypical profiles showing HbA levels at 20.8%, 21.2%, and 43.8%, respectively. Sickle hemoglobin was presented in the HPLC chromatogram of all subjects at moderately elevated levels of 60.2%, 62.4%, and 42%, respectively. Hemoglobin A2 showed elevated levels at 8.9%, 7.3%, and 4.9%, while fetal hemoglobin (HbF) was elevated moderately at 7.9%, 6.8%, and 3.8%, respectively. Molecular investigations, by PCR-RFLP and PCR-DGGE methodologies, showed all of the patients as heterozygotes for the sickle cell mutation. Both students showed heterozygosity for the mild β^{+} mutation nt –88 (C-A). The third patient showed heterozygosity for the silent β^{++} mutation nt –101 (C-T). Mapping of the alpha globin genes revealed both of the students as homozygotes for the rightward deletion ($-\alpha^{3.7}/-\alpha^{3.7}$), while the third patient showed heterozygosity for the leftward deletion ($-\alpha^{4.2}/\alpha\alpha$). Further, haplotype analysis of the beta globin gene indicated all of the patients are bearing the Saudi-Indian haplotype of the sickle cell mutation. Presumeably, these molecular factors were interact synergistically to give such a mild form of sickle cell disease. In summary, molecular diagnosis might contribute significantly to resolve such an ambiguity in clinical presentation.

Congenital Absence of the Nose (A Case Report)

Hamedi AB. Akhlaghi F
Mashhad University of Medical Science

Background: Congenital absence of the nose is very rare condition. A review of the literature reveals that the previously applied terms, arhinia. The etiology of this condition is unknown. Arhinia is one of the presentations of malformation in the middle third of the face, often associated with central nervous system and somatic anomalies of different degrees of severity with a high mortality rate. About 20 cases have been reported in the medical literatures. The aim of presentation this case is be rarity and study on probable predisposing factors.

Case report: A male infant born without a nose was referred at the age of 8 hours from a hospital in quchan, north east of Iran to emergency nursery room in Ghaem hospital, Mashad, Iran for respiratory and feeding problems. The baby was full ter with birth weight of 2.5 Kg after a spontaneous vaginal delivery and Apgar scores were 8 in 1 min and 9 in 5 minute. He was the first child of healthy and unrelated parents. The clinical examination confirmed absence of the external nose. In addition there was a nasal bridge and slight hypertelorism. Vital signs were otherwise normal. He had complete absence of the nose with no nasal pits. Her psychomotor development was normal. Facial and brain C.T showed absence of nasal bone, cribriform plate and septal structures. – The mother was a healthy 16 years old primi gravida. She had no history of drug or chemical intake or exposure to radiation and had no significant illness during pregnancy.

Key Words: Nose-Congenital-absence.

Hereditary Nonpolyposis Colorectal Cancer: Identification of a Novel hMSH2 Gene Mutation in an Arab Family

Marafie M*; Motawy M**; Macdonald F***; Bell J[3]; Al-Awadi S*
* Centre for Medical Genetics, Kuwait. ** Cancer Control Centre, Kuwait. *** West Midlands Regional Genetics Laboratory, Birmingham.

Hereditary nonpolyposis colorectal cancer (HNPCC) is a highly penetrant autosomal dominant syndrome characterized by early onset colon cancer and a variety of extracolonic tumours such as endometrium, ovary, stomach, pancreas, small intestine, hepatobiliary and urinary tract. HNPCC accounts for around 5% of all colorectal cancers and germ-line mutations in the DNA mismatch repair genes (MMR) are responsible for this disorder. Several MMR genes (hMLH1, hMSH2, hMSH6, hPMS1, hPMS2) have been identified. The majority(90%) of the mutations have been reported in the hMLH1 and hMSH2 genes, and only 10% in MSH6. We offered genetic counseling to a cohort of patients attending the familial cancer clinics at both Kuwait Medical Genetic Centre and Cancer Control Centre, aiming to identify families with hereditary cancers. Here we report a highly consanguineous Arab family from Kuwait that fits the HNPCC syndrome. Pedigree analysis revealed the presence of 23 members of consecutive generations expressing colonic and extra colonic tumours, affecting both sexes at different young ages. DNA analysis were offered to 3 affected members. Mutational analysis of all exons of hMSH2 gene were carried out by PCR/SSCP test followed by direct sequencing of the fragment with a band shift. Sequencing showed a germ-line nonsense mutation Q76X in exon 2 of the hMSH2 gene in all 3 samples. Other members of the family are currently being investigated. Genetic counseling and screening for germ-line mutations in individuals of such genetically predisposed families will help in preventing additional tumours in the patients as well as in high-risk non-symptomatic mutation carriers.

Keywords
Colon cancer, hereditary nonpolyposis colorectal cancer (HNPCC), mismatch repair genes.

Oxoprolinuria: Biochemical Observations and Case Report

Naveed M.; Al-Hashmi H.; Taleb M.
Genetic & Thalassemia Center, Al-Wasl Hospital, Department of Health & Medical Services, Dubai, UAE

5-Oxoprolinuria (pyroglutamicaciduria), resulting from glutathione synthetase deficiency is a very rare autosomal recessive disorder characterized in its severe form by massive urinary excretion of 5-oxoproline, metabolic acidosis, hemolytic anemia, and central nervous system damage. The metabolic defect results in low GSH levels, presumably with feedback overstimulation of gamma-glutamylcysteine synthesis and its subsequent conversion to 5-oxoproline. We have come across a patient with persistent 5-oxoprolinuria, who presented with transient metabolic acidosis as a neonate; but remains asymptomatic with normal growth and development till eight-months of age.

Alpha-Thalassemia Genotype in Iran

Zarbakhsh, B; Zeinali, S; Karimi, M; Mohammadi, M.S.; Shafielyeh, E.; Rohi, S.; Sadeghi, S.
Pasteur Institute of Iran

In this study we wanted to see whether all a-globin gene mutation among iranian population, can be detected by the simple, rapid and inexpensive methods. Using PCR based tests for defining the mutations causing a-thalassemia, was our first try. We begun with the use of most famous primers mention in a lot of articles.though we used the published method, no convincing results were obtained. We Therefore changed the blood DNA extraction method and performing Hot Start and Touch Down PCR, However the resulting bands included non-specific fragments. Finally by preparing a new buffer Mix, we were able to amplify the target DNA using all mentioned primers. We performed both globin chain synthesis analysis and a-globin gene analysis by PCR in 85 subjects with microcytosis and normal HbA2, Hbf, and serum iron, the a/b globin chain synthesis ratio, were between 0.4 to 1.50.Sofar we have found the most common(about45%) being the heterozygous state for -a3.7Kb deletion.the homozygous condition for this pattern was about (-a4.2/aa) genotype. Also we report one case with the (-a20.5/aa) genotype. It could be interesting to examine the relationship between the type of mutation and the globin chain synthesis results and other blood tests.

Pregnancy Outcomes in Women with Pregestational Diabetes Compared with the Gestational Diabetic Mothers

Farideh akhlaghi; Monireh Pourjavad
Mashhad University of Medical science

Introduction: To compare pregnancy outcome in pregestational diabetes and gestational diabetic mothers.Diabete melitus is one of the most common and important metabolic disease in the pregnancy period.Despite considerable advances in the management of the pregnancy complicated by diabete melitus, embryo, fetus and the mother can experience serious complication of diabetes. Overt diabetes has a significant impact on pregnancy outcome directly to diabetes.The probablity of successful outcome for the fetus&infant and the pregestational diabetic mother are related somewhat to the well controlled of diabetes.We decided to study, compare the severity of maternal and perinatal complications in two group of mothers (Gestational and overt diabetic mothers).
Method: In Diabetic Research Center in the east of Iran(Khorasan) we have been studing prinatal care and pregnancy outcome of women with diabetes who were suffered for gestational and overt diabetes that reffered to this center.During pregnancy period and after delivery outcome of mother and neonates were studied.(Nov 2001-Nov 2002).
Results: The pregnant women suffering from overt diabete were the most studied population. 22% of pregnancies lead to abortion and the most incidence of its was seen in pregnant women suffering from type 2 diabete melitus. Preeclampsia was seen in 11% of cases that the most incidence of its was seen in gestational diabete. Fetal death was seen in 10% of these pregnant women that the most incidence of its was seen in patients with overt diabetes.The most common method for child birth was cesarean section in this study(43%).The most incidence of its were in pregnant women with overt diabetes.The most incidence of infant complications were seen in diabetic mothers with overt diabete before pregnancy.Complications in infant includes: hypoglycemia 22.2%, premature infant 11.1%, respiratory distress syndrome 13.8%, congenital anomaly 12.5 %, and macrosomia 6.9%.
Conclusion:In the diabetic mothers we would be recomended consultation and carefully prenatal care for prevention of complications in mothers and fetus.Use of folic acid during pregestational and pregnancy period is very nessesary for prevention of congenital anomalia. Metabolic care in pregnant mothers have be done carefully.Women with poorly controlled diabete may not expect a successful pregnancy outcome.

Key Words: Pregnancy-Diabet-Gestational

Three Cases with Limb Anomalies

S.N. Almadani; F. Afroozan; Y. Shafghati; M.H. Kariminejad
Kariminejad-Najmabadi pathology & Genetic center, Iran

Two patients with Tibial aplasia-Ectrodactyly syndrome. This pattern of malformation are included: split-hand/split foot, absence of Arms&Legs. Ours patient is 18 month old male with unrelated parents that suffered from Hypoplastic /absent tibia, split hands, Hypoplastic/absent carpals, Absent metacarpals & metatarsals, Oligodactyly, phalange& toes, Bilateral patellar hypoplasia and contracted knee joints. The second case is two years old male from unknown parents that suffered from Acromelia of upper limbs, Reduction deformity of arms, Absent hand, Hypoplastic/absent carpals, Absent finger/toe or oligodactyly, Absent metacarpals & metatarsals, Hypoplastic or absent Tibia.

The second syndrome is Fibular aplasia- oligodactyly-camptomelia that reported by Hecht & Scott (1981) is included absent hands, absent feet. Our case was one years old male from unrelated parents that suffered from Reduction deformity of arm, Absent hands, Oligodactyly, absent lower limb, Bowed and hypoplastic femur, Hypoplastic/absent fibula and tibia, Absent feet& toes, Syndactyly of toes.

Our two cases with tibial aplasia-Ectrodactyly are similar to patients that reported with this syndrome in references, but our case with fibular aplasia- oligodactyly- camptomelia may be similar to the femur- fibula- ulna complex (FFU syndrome) but ulna deficiency not seen in our case, And the otherhand in fibular- oligodactyly- camptomelia not seen any kind of femoral deficiency.

Distribution of the Clinically Important HLA Alleles Among Healthy Bahraini Population

Eman Farid, Samya Rostum, Aida Ali, Fadheela Al Alawi
Department of Pathology, Microbiology/Immunology Section, Salmaniya Medical Complex, Kingdom of Bahrain

HLA association with certain autoimmune and rheumatic diseases is well established. HLA which is located on chromosome 6, is considered as one of the inherited risk factors for some diseases. Ethnic origin plays a crucial role in the different distribution of HLA alleles among individuals. Kingdom of Bahrain is an island with a population of mixed Arabian and Persian origins.

To study the distribution of clinically important HLA alleles among healthy Bahraini individuals we studied 250 unrelated individuals out of 600 kidney or bone marrow transplant potential donors, during the period January 1996 to January 2002; in classI HLA B27 and B5 were selected, while in class II HLA DR2, DR3 and DR4. HLA B27 is known to be associated with ankylosing spondylitis, HLA B5 with Behcets disease, HLA DR3 and DR4 with type 1 diabetes, HLA DR4 with rheumatoid arthritis and HLA DR2 and DR3 with systemic lupus erythematosus. Among the studied group HLA B27 was only present in 1.6%, HLA B5(with its two splits 51 and 52) in 22%, HLA DR 2 in 20%, HLA DR3 in 14%, HLA DR4 in 12%.HLA B 27 is a rare allele in the Bahraini population, while the other studied alleles are moderately distributed.

A Study on Triple Maternal Serum Screen as a Tool of Prenatal Diagnosis in Bahrain

Das N.S; Hasan Sanad
Clinical Biochemistry Section, Department of Pathology Salmaniya Medical Complex, Kingdom of Bahrain

Eighty percent of children with Down syndrome and 90% of children with Open neural tube defect are born to women less than 35 years of age. Maternal triple serum screening during 16 18 weeks of gestation with maternal serum alpha-fetoprotein, human chorionic gonadotropin, and unconjugated estriol is considered as an important tool in prenatal diagnosis of these conditions. Though certain centers have initiated quad maternal serum screening with addition of dimeric inhibin type A, triple screen is most commonly used tool. Low maternal serum AFP, and uE3 and high HCG are considered as markers to suggest possible increased incidence of risk of Down syndrome. Calculation of Multiple of Median (MOM) and derivation of adjusted MOM is essential for evaluation of risk. Adjusted MOM evaluation requires information relating to 1) Patient's date of birth, 2) Current weight, 3) Due date, 4) The method used for determining the due date (US, LMP), 5) Date of last menstrual period, 6) The number of fetuses present (if known), 7) patient s race, 8) history of IDDM and 9) known family history of neural tube defects. Median of AFP, HCG and uE3 have to be assessed for the race. The present study evaluated median of AFP, HCG (MEIA method) and uE3 (Chemiluminescence method) for 16,17,18 wks of gestation in Bahrain population and presented along with comparison to median in western population.

Hemoglobin and Hematocrit Values of Newborns in Mashhad City, Iran

Gholam Ali.Mamouri; Vahid Nobahar
Mashhad University of Medical sciences, Pediatric department,Ghaem Hospital, Mashhad

Objective:Hemoglobin and hematocrit have been used routinely in the diagnosis of neonatal anemia and polycythemia.Our objective in this study include:determining the red cell values(Hb-,Hct) in the cord blood,compare these values with other reports,to address association between cord blood values with maternal hematological indices (Hb).

Method and material:A total 170 paired normal neonates and mothers were included in a cross sectional study,over 3 months in 2000.From the cord blood of these infants,with 37-42 weeks gestational age and birth weight 2.5-4 Kg.Hb and hemtocrit obtained by electronic coulter counter.All of these neonates,were born by vaginal delivery and were products of pregnancies without complications.

Results:The mean Hb and Hct were(15.8g/dl,48%) with the range of 11 to 21.4 g/dl, 34% to 63% respectively.There were significant differences between males and females values(16.02±1.4 versus 15.34±1.8g/dl).Of total newborns,97 were male and remainder female.Polycythemia (Hct>63 %) was not found in any infants,but 11.7% of newborns had Hb less than 14 g/dl.Mean hemoglobin of mothers were 12.32g/dl and 18% had less than 11g/dl.There was not any correlation between cord Hb and parity,birth weight and maternal age.By using pearsons correlation indices there were not direct association between cord Hb and maternal Hb.However more anemic neonates were born from mothers with low Hb (Hb<11g/dl) relation to mothers with Hb>11g/dl (29 % vesus 8.2%). Conclusion:we were found that mean Hb in our newborns were less than values reported in the most literature and also no correlation was found between maternal Hb and cord Hb.

Genomics in a Global Context: A Programmatic Approach to Avoiding A Genomics Divide

Abdallah S. Daar
University of Toronto Joint Centre for Bioethics

Inequities in global health are amongst the most crucial issues in bioethics today. A number of political, economic and social developments could either improve or worsen the disparities that are the manifestation of this inequity.

Human development, including in the area of health, has historically often been linked to specific scientific advances. The 2001 UNDP World Development Report entitled "Making new technologies work for human development" concludes that high-technology can be made relevant and useful to poor countries, as long as risks are well managed.

The sequencing of the human genome is a major scientific achievement. It has raised much hope amongst medical researchers, investors and the public. Because powerful tools can be dangerous, many people are also concerned about the ethical implications of applying genomics knowledge. This concern has been heightened by conflating genomics and genetically modified (GM) crops. In the process, and with the unsatisfactory handling of the debate on GM foods, the potential of genome-related biotechnologies for improving the health of people in developing countries has received very little attention or emphasis.

Over the recent past we have worked with the World Health Organization and other international organizations to look at both the potential of these technologies to improve world health and their ethical implications. We have developed programs at the University of Toronto Joint Center for Bioethics that have begun systematically to address the various issues and approaches that might, in the long run, reduce the likelihood of the development of a genomics divide. This work is strengthened by the recent designation of our centre as a WHO Collaborating Centre in Bioethics, and our growing links with international organizations such as PAHO, the African Centre for Technology Studies, The Indian Council of Medical Research and the Chinese Academy of Sciences.

Genomics and related biotechnologies do have the potential to reduce global health inequities by providing developing countries with better, cheaper and more robust means of preventing, diagnosing, ameliorating and treating major diseases burdening their populations,. However, this can only happen if enlightened and objective analysis, public engagement, science and technology policies and specific investments are marshaled towards this specific goal. Additionally, in areas such as genomics, it may be necessary critically to re-examine the mission and approach of "traditional" bioethics.

The talk will (1) highlight international developments that focus on the potential role of genomics for the improvement of world health; (2) provide an overview of the work of our programs and (3) describe the results of some of the major research projects.

Birth Defect Registry, the Hungarian Experiences

Andrew E. Czeizel
Foundation for the Community Control of Hereditary Diseases, Budapest, Hungary

The Hungarian Congenital Abnormality Registry was established in 1962 based on the obligatory notification of cases with structural birth defects, i.e. congenital abnormalities by obstetricians (in Hungary all deliveries take place in hospital and birth attendants are obstetricians), paediatricians (working in general and specific surgical, cardiological, orthopedic, etc. institutions), by pathologists (autopsy was obligatory for all infant deaths and usual for stillborn fetuses) and by all other physicians. Later the data of malformed fetuses from electively terminated pregnancies were also obtained. The task of the Registry has been to determine as reliably as possible the baseline occurrence: total prevalence rate of different congenital abnormalities (1) to detect temporal and/or spatial increases (the so-called clusters), (2) to help plan medical and social services for affected persons, and (3) to estimate the public health importance of different defects so that resources can be properly allocated. The quality control of the Registry is based on the proportion of misdiagnoses, completeness of notification and the use of a pathogenetically oriented classification. The outcome evaluation indicated the different quality of recorded data in lethal, severe and mild defects. In 1980 the first task of the Registry was extended to a population based case-control surveillance system. This system is appropriate for the postmarketing surveillance of drug teratogenicity. For the improvement of congenital abnormality diagnoses, to get informed consent from the parents of cases to record personal data, to have a communication with parents and to provide material for research.

Primary Prevention of Some Congenital Abnormalities

Andrew E. Czeizel
Foundation for the Community Control of Hereditary Diseases, Budapest, Hungary

Experimental studies and epidemiological observations indicated the role of micronutrient deficiency in the origin of structural birth defects, i.e. congenital abnormalities (CAs) particularly of neural tube defects (NTD). Two intervention studies showed the efficacy of a folic acid-containing multivitamin and folic acid (4 mg) alone in the prevention of NTD recurrence. However, about 95% of women who deliver infants with NTD have no previous NTD pregnancies, thus the first goal of the Hungarian randomised double-blind controlled trial and two-cohort controlled study was to determine the efficacy of this method in the reduction of first occurrence of NTD. The pharmacological dose (more than 1 mg) of folic acid may have some adverse effects thus it cannot be recommended for the population at large or without medical supervision. Thus the Hungarian trials tested the preventive effect of a physiological dose (0.8 mg) of folic acid as a component of a micronutrient combination. Finally, the third goal of the Hungarian trials was to reveal all possible adverse and beneficial effect of folic acid-containing micronutrients. The pooled findings of these trials indicated that approximately 92% of the first occurrence of NTD may be prevented by the use of the micronutrient studied during the periconceptional period. In addition a significant reduction was found in the occurrence of cardiovascular CAs particularly ventricular septal defect, urinary tract's CAs, especially the obstructive CAs of pelviureter junction and limb reduction CAs. The available data suggest that the cause of NTD and other CAs is an interaction between genetic predisposition caused by inborn errors of folate and/or homocysteine metabolism and deficiency of certain micronutrients mainly folate. Thus micronutrient deficiency can trigger the genetic predisposition or a good diet and periconceptional supplementation of folic acid and other micronutrients can neutralize the genetic predisposition. There are three options for the use of micronutrients in the primary prevention of NTD and other CAs; diet rich in micronutrients, periconceptional supplementation and food fortification. These new approaches offer a breakthrough in the primary prevention of CAs. Obviously the primary prevention of CAs is better than secondary prevention, i.e. termination of pregnancy after the diagnosis of serious fetal defects.

Preimplantation Genetic Diagnosis as Frontier of Early Detection: Are we ready to implement it in the GCC Countries?

Hamza Eskandarni

Preimplantation genetic diagnosis (PGD) is nowadays becoming an important theme at various specialized and advanced centers for assisted reproduction technology. It is, therefore, considered to be at the forefront of genetic screening when contemplating early detection for inherited disorders or abnormalities. However, with PGD still at an early stage of development, it is an appropriate time to consider the potential uses of this technique in the GCC countries for detecting common genetic disorders at the start of new life. Moreover, the implication of PGD in the region should have a tight scrutiny for reasons that will be addressed. Nevertheless, PGD raises troublesome ethical and legal issues, which is likely to be controversial.

The allele frequencies of ST14 (DXS52) in hemophilia A and normal population in Iran

Azimifar Babak, Zeinali Sirous, Seiedna Yousef
Biotechnology Research Center, Pasteur Institute of Iran

Hemophilia A is an X-linked bleeding disorder affecting approximately 1 in 5000 males births worldwide. The disease is caused by defect in the function or production of blood coagulation factor VIII (FVIII). In majority of hemophilia A patients, no common mutation is present or when present is not easily identifiable. This is partly due to large size of FVIII gene, its many exons and great variety of mutations described. The limitation has been overcome by the use of linkage-based analysis using polymorphic DNA markers. The multiallelic locus DXS52 (ST14) is an extragenic DNA marker linked to the human FVIII gene and has been found to be highly polymorphic in different populations. The DNA sequence spanning ST14 VNTR was amplified using PCR. The PCR products were subjected to electrophoresis on a 1% Agarose gel. 13 different alleles (ranging in size from approximately 700 to 3000 bp) were observed in 160 x-chromosomes in DNA samples from Iranians. The rate of observed heterozygosity is 75 %. We found ST14 VNTR to be highly polymorphic in our population and in conjunction with intragenic markers could provide an effective way for linkage analysis of hemophilia A families.

Keywords: VNTR, Hemophilia A, DXS52, Factor VIII, PCR.

Mutations of the Glycine Receptor beta-subunit Result in Hypereplexia

Meyer BF, Hejazi N, Yamani S, Ozand PT, Carroll P, Al Ahmed S, Al-Hamed M, Yousef N, Abu Amero S and Kambouris M
AmGene, Research Centre, KFSH&RC, Department of Genetics, Research Centre, KFSH&RC, Department of Neurosciences, KFSHRC

Hyperekplexia (HEK) is an autosomal dominant disorder characterized by abnormal startle response and neonatal hypertonia (major form), or only the abnormal startle response (minor form) due to dysfunction of the glycine receptor (Gly-R). Glycine, an inhibitory neurotransmitter in the brainstem and spinal cord has been implicated in the regulation of muscle tone. The Gly-R comprises four ligand binding alpha-subunits and one structural beta-subunit encoded by the GLRα1-4 and GLRβ genes respectively. HEK in humans has until recently been exclusively associated with mutations of the GLRα-1 subunit. However, murine studies have indicated that the HEK phenotype results from mutations of either the GLRα or GLRβ. This study used four Saudi Hyperekplexia families. Genotyping of these families was conducted using microsatellite markers closely linked to the glycine receptor alpha 1 (GLRA1) (D5S209, D5S119, SPARK-1), glycine receptor alpha 3 (GLRA3) (D4S408, D4S2417, D4S2431) and glycin.

Localization of the Gene for a Novel Autosomal Recessive Neuromuscular Disease Featuring Tremulous and Myoclonic Dystonia with MRI White Matter Alterations

M. Kambouris, S.A. Bohlega, D. Trabzuni, B.F. Meyer
AraGene, Research Center – KFSHRC

A novel autosomal recessive neuromuscular disorder characterized by tremor of juvenile onset, dystonia, myoclonus, spasticity, preserved cognitive cerebellar and peripheral nervous system functions with diffuse white matter alterations in brain MRI is segregating in two consanguineous sibships, parts of one extended family. Central conduction times for visual, motor and sensory systems are all prolonged. Lysosomal, peroxisomal, mitochondrial and other metabolic abnormalities were excluded. Homozygosity mapping localized the offending gene to Hsa 17p13.1-13.3 with LOD=3.89 at theta=0. The expected homozygous genotype in all affected individuals was for markers D17S1298 and D17S1537. Additional mapping is currently performed for narrowing of the linkage interval with microsatellite markers and Single Nucleotide Polymorphisms (SNPs). The linkage interval boundaries at present (5 million bp at the physical map, contig #NT_010692.7) are defined by the presence of heterozygosity for a distal SNP in affected individuals within the P2RX1 gene and genetic recombination for the proximal marker D17S974. Positional candidate genes screened for pathogenic sequence alterations and subsequently excluded are: MYO1C (Myosin 1C); GABA (A) receptor-associated protein; VAMP2 (Synaptobrevin 2, vesicle associate membrane protein 2); CHRN (Nicotinic Cholinergic receptor,) E & B1 subunits; P2RX1 (Purinergic receptor P2X, ligand-gated ion channel 1); SRR (Serine racemase) and PITPN (phosphotidylinositol transfer protein). Additional positional candidates are presently screened by Mutation Detection Enhancement (MDE) Heteroduplex analyses and sequencing. The unique phenotype and localization to 17p13 are consistent with the identification of a novel neuromuscular disorder.

Novel Mutations underlying Nephrogenic Diabetes Insipidus (NDI)

Carroll P, Al-Mojalli H, Al-Abbad A, Al-Hassoun I, Butt AI, Rajab M, Al-Hamed M, Kambouris M and Meyer BF
AraGene, Research Centre, KFSHRC, Department of Genetics, Research Centre, FSHRC
Department of Pediatrics, KFSHRC

NDI is an inherited disease characterized by inability of the kidney to concentrate urine upon stimulation with vasopressin. Mutations of the vasopressin type 2-receptor (AVPR2) (Xq28) or aquaporin-2 (AQP2) (12q13) result in NDI. AQP2 mutations have been associated with autosomal dominant (AD) and autosomal recessive (AR) inheritance. Two of 16 NDI families were selected for analysis. Family 1 (apparently X-linked) had 3 severely affected males, normal parents and 2 clinically normal females. Family 2 (apparently AR) had 2 severely affected males, 1 affected female and normal distantly related parents. The coding regions of AVPR2 and AQP2 were screened for mutation by heteroduplex analysis of obligate carriers. No heteroduplexes were detected in AVPR2 and further analysis was by direct sequencing. For AQP2 heteroduplex patterns were able to exclude involvement in both families. Closer examination of family 2, showed that the mother was mildly symptomatic and the daughter had less severe disease than

Molecular Characterization of a Novel Autosomal Dominant Hyaline Body Myopathy (HBM) in a Large Saudi Family

Bohlega S, Abu-Amero SN, Lach B, Wakil SM, Kambouris M, Carroll P, Al-Omar R, Al-Sayed Y, Hodgkinson C, Bharucha V, Cupler EJ and Meyer BF

Institution: AraGene, Research Centre, KFSHRC, Department of Neurosciences, KFSHRC; Department of Pathology and Laboratory Medicine, KFSHRC; Department of Genetics, Research Centre, KFSHRC ; Prince Salman Center for Disability Research

HBM is a rare congenital myopathy with unique histopathological features. We describe HBM with autosomal dominant inheritance in a three generation Saudi family. Eight of 15 children, their mother and maternal grandmother were affected. Five of nine patients presented with non-progressive, proximal and distal weakness, no bulbar, facial or ocular weakness, but significant wasting and loss of subcutaneous fat. The other four patients showed early, progressive scapuloperoneal weakness with early loss of ambulation. Muscle biopsies showed discrete sub-sarcolemmal hyaline bodies (HB) in approximately 20% of type 1 skeletal muscle fibres. A whole genome scan of approximately 7 cM resolution and linkage analysis with GeneHunterPlus resulted in a maximum LOD score of 3.01 at D14S1280.). High-density mapping surrounding the linked locus was performed with 8 additional fluorescently labeled microsatellite markers. Multipoint parametric linkage analysis showed that the linkage region with a maximum LOD score of 3.01 extended from D14S742 to D14S608 with a peak NPL score of 3.75 at D14S608. The MYHC genes MYH6 and MYH7 map to the region between D14S742 and D14S1280. Sequence analysis of the coding regions of MYH7 revealed an A to T transversion at nucleotide position 23611 (X5288.9) resulting in a histidine to leucine amino acid change at residue 1901 (H1901L). Sequencing of an additional 130 chromosomes from random normal individuals in the population showed only the wild type A nucleotide at position 23611 (X5288.9). Pathogenicity of the H1901L mutation most likely results from disruption of MYHC assembly.

Two Founder Mutations are Responsible for Papillon Le-Fevre Syndrome in Saudi families

Ullbro C, Pedersen K, Boumah CE, El-Samadi S, Wakil SM, Al Ahmed S Yousef N, Kambouris M and Meyer BF
ArsGene, Research Centre, KFSHRC; Department Genetics, Research Centre, KFSHRC; Department of Dentistry, KFSHRC

We genotyped 23 Saudi PLS patients using 10 microsatellite markers (D11S901, D11S4187, D11S4147, D11S1795, AFM207ya5, D11S1354, D11S4197, D11S4082, D11S1780, D11S931) surrounding the cathepsin-C gene on chromosome 11. All 23 patients showed homozygosity at D11S1354 and D11S1780 which defined a region of approximately 600kp. At D11S1354 and D11S1780 genotypes and haplotypes indicated the presence of a strong founder effect. At D11S1780, three genotypic sub-groups 183/183 (18 patients), 173/173 (4 patients) and 189/189b (1 patient) were identified. Direct sequencing of cathepsin-C exons from individuals representing each of the three genotypes was performed. The most common genotype (183,183) was associated with 815G to C mutation in exon 6 resulting in an arginine to proline change at amino acid 272 (R272P). Patients with the 173/173 genotype all carried a G300D mutation in exon 7 of cathepsin-C. The 189/189 genotype was restricted to one family which was not identified. Patients from this study represent the largest group of PLS families studied at the molecular levels and show the presence in Saudi Arabia of two founder populations of PLS patients. This has provided an opportunity to explore phenotype/genotype correlations within these founder groups.

Localization of Genes for a Novel Disease Characterized by Intestinal Lymphangiectasia to 6p21-22 and 13q14.2-21.1: Evidence for Genetic Heterogeneity

M. Kambouris, S. Shabib, A. Al Mehaidib, H. Halabi, B.F. Meyer.
ArabGene, Research Center KFSHRC

Intestinal lymphangiectasia is the dilatation of the intestinal lymphatic system vessels and is characterized by protein losing enteropathy, steatorrhea and lympyhopenia. Four consanguineous (all first cousin marriages) unrelated nuclear families affected with a novel autosomal recessive disorder characterized by intestinal lymphangiectasia were identified. Disease onset is early (~2 years) and clinical features include lymphedema, protein losing enteropathy, malabsorption, growth failure, generalized edema, diarrhea and recurrent chest and skin infections. None of the known hereditary intestinal lymphangiectasia and/or lymphedema disorders seem to correlate with the inheritance pattern and clinical symptomatology seen in these families. Homozygosity mapping with microsatellite markers was performed. For family 1 (four affected), a maximum LOD score of 4.3 at theta=0 was obtained for markers D6S2439 & D6S1017 identifying an area of homozygosity by descent localizing the disease gene to 6p21-22. A maximum linkage interval of 30 KcM was defined by the presence of recombinants at the distal D6S2439 and heterozygosity at the proximal D6S424.0 markers. Linkage to 6p21-22 could not be established for at least two of the other three families indicating genetic heterogeneity. Affected members from families 2 (three affected) and 3 (two affected) share an area of homozygosity at 13q14.2-21.1 within a 30 KcM linkage interval defined by recombinant events at the proximal D13S325 and the distal D13S1320 markers resulting in a combined LOD score of 4.35 at theta=0 for the homozygous interval containing markers D13SD788 & D13S1492. The unique lymphangiectasia phenotype the autosomal recessive pattern of inheritance and the unique localization to 6p21-22 and 13q14.2-21.1 are consistent with the identification of a novel lymphangiectasia disorder exhibiting genetic heterogeneity. The similarity in clinical symptomatology and the genetic heterogeneity demonstrated suggests that the different genes involved might be components of a common biochemical and/or signaling pathway. Related candidate genes mapping within the linkage intervals at chromosome 6 and 13 are being screened for mutations.

Rapid Detection and Identification of C. Jejuni and C. Coliform Chicken Sample by Real Time PCR

Abu Halaweh Marwan; Bates, John2; Patel Bharat K. C1
Microbial Research and Discovery Unit, School of Biomolecular and Biomedical Sciences, Griffith University, Nathan, Brisbane, Australia 4111 and Public Health Microbiology, Queensland Health Scientific Services, Queensland Department of Health, Coopers Plains, Brisbane, Australia

We have developed a two-stepped Real Time PCR assay, in a LightCycler™, which detects, identifies and differentiates the pathogenic, C. coli and C. jejuni, from all other members of the family Campylobacteriaceae. In the first step of the assay, the continuous monitoring of Fluorescence Resonance Energy Transfer (FRET) signal is acquired from the of two adjacent fluoroprobes, [probe Cy5 (5'-Cy5-AGGTGTGCATGGTTGTCGTTGTCG-PO4-3') and probe FITC (5'-GTGCTAGCTTGCTAGAACTTAGAGA-FITC-3')], which hybridise to the target 681bp amplicons of the 16S rRNA gene (E. coli position 1024 to 1075). These amplicon are produced by the primer pair, F2 (ATCTAATGGCTTAACCATTAAAC, E. coli position 783) and Cam-Rev (AATACTAAACTAGTTACCGTC, E. coli position 1464). As expected, an increase in the fluorescence was observed with both C. jejuni and C. coli but not with any other Campylobacter species. A Tm of 65 o C was obtained from the disassociation of the fluoroprobe from the target DNA confirming the specificity of the assay.

Identification of Genes Responsible for Coronary Heart Diseases in Nigerian Population

Sokefun O.
The bioscience research center, faculty of science,lagos state universty, ojo PMB 1087 Apapa, lagos Nigeria

Positional cloning is now widely used for the identification of genes defects that are the basis of inherited diseases. A necessary first step for positionla cloning is the mapping of the gene locus that co - segregates within families with a particular disease or trait. which allow for the allocation of a specific chromosomal position to the responsible gene.

Although mapping was initially developed for monogenue traits, it has now become the most widely used strategy to allocate factors involved in the etiology of complex diseases. the most powerful technique currently available is linkage analysis with highly polymorphic microsatelite markers, which involves an examination of the entire geneome with a set of evenly spread markers. This types of study is usually referred to as a genome scan.

Currently, in conjunction with a few partners in Europe we are interested in mapping the genetic factors responsible for the multifactorial diseuse hypertension which leads usually to coronary heart dieseases if not properly managed. The study involves the analysis of large numbers of phenotypically well characterised families. For this project a total of 100 families totaling about 2670 probands with two or more affected siblings each due for investigation to identify genetic factors involved with susceptibility. The study is based on isolated populations to take advantage of the restricted genetic heterogeneity in these populations.

Three Bahrainis Families with Retinitis Pigmentosa, Ataxia, Neuropathy, Cerebella Hypoplasia, Mental Retardation, and Hypogonadotrophic Hypogonadism

Fatima Neama, Fouad A.M Ali, A.Jabbar Al.Abasi, Ghazi Al.Mahroos

The classic association of ataxia, neuropathy and retinits pigmentosa has been reported earlier as (NARP syndrome OMIM no. 551500) However, hypogonadotrophic hypogonadism was not describe with the above syndrome, but in (boucher-Neuhauser syndrome OMIM no. 215470) the association of spinocerebellar ataxia, hypogonadotrophic hypogonadism and chorioretinal dystrophy has been recognized by Boucher and Gibberd (1969).

We describe three separate families from Bahrain with five consistent features. These include, ataxia, neuropathy, mental retardation, hypogonadotrophic hypogonadism, and retinitis pigmentosa. The parents of these families were consanguineous and all of them have similarly affected sibling.

In conclusion our patients could be a spectrum of either or a newly diagnosed syndrome.

Multifactorial Disorders in Saudi Arabia

Arjumand S. Warsy; Mohsen A.F. El-Hazmi
Department of Biochemistry, College of Science & College of Medicine, King Saud University, Riyadh, Saudi Arabia

Multifactorial disorders are a group of frequently encountered genetic defects that require contributions from both genetic and environmental factors for their development. These defects are polygenic in nature and several genes contribute to the genetic susceptibility. Hence these disorders are frequently referred to as "complex traits". Some of these defects are present at birth while others develop later on during adult life. This latter group contributes significantly to morbidity and mortality in the human population.

The aim of this investigation was to determine the prevalence of adult-onset multifactorial genetic defects in the Saudi population. An extensive National investigation was carried out over a period of three years in which adult Saudis were screened from different regions of Saudi Arabia utilizing W.H.O criteria for the diagnosis of diabetes mellitus, hypertension, overweight and obesity. The group screened included 14,500 adult males and females (males= 6,162; females= 8,498).

The results showed the presence of insulin dependent diabetes mellitus in 0.23% and 0.3%, non insulin dependent diabetes mellitus in 5.63% and 4.53%, hypertension in 5.39% and 3.65%, overweight in 27.23% and 25.21% and obesity in 13.05% and 20.26% of the total Saudi males and females, respectively. Analysis of the data from different regions showed significant differences in the prevalence of these disorders in the different regions of Saudi Arabia. In general, the males had a higher prevalence of diabetes mellitus, hypertension and overweight in most of the regions, while obesity was of more frequent occurrence in the females. Each of these disorders showed a significant positive correlation with age ($p < 0.05$). Investigations showed a strong correlation between overweight and obesity and the development of diabetes mellitus and hypertension in both male and female groups. Several biochemical abnormalities were observed in these patients, particularly hyperlipidaemias and elevation of leptin and Lp(a).

This paper will present an overview of multifactorial disorders in the Saudi population and will discuss steps necessary to decrease the prevalence of these disorders in this population.

Genetics of Obesity

Arjumand S. Warsy; Mohsen A.F. El-Hazmi
Department of Biochemistry, College of Science & College of Medicine, King Saud University, Riyadh, Saudi Arabia

Obesity is a multifactorial disorder with genetic and environmental factors contributing to its development. Studies of concordance of obesity in families, twins, and adopted children and their parents imply that genetic factors make a substantial contribution and as much as 80% of variance in BMI appears to be ascribable to genetics. Nevertheless, development of obesity is absolutely dependent on the availability of sufficient food and on its consumption and utilization implying that the influence of genotype is powerful but subtle. Hence, the environmental factors shape the genotype to produce a certain phenotype.

Over the last couple of decade interest in the prevalence and distribution of obesity in different populations has gained considerable momentum due to the association of obesity with a number of complications such as development of diabetes mellitus, hypertension and cardiovascular disease. At the same time, considerable efforts have been directed towards the understanding of the genetic factors contributing to the development of obesity. Several genetic loci have been implicated and a long list of loci is now available, where each gene is believed to have a small but significant effect on the development of obesity. These include: leptin, neuropeptide Y, leptin receptor, melanocortin receptor, insulin and carboxypeptidase E. The contributions from leptin and its receptor are Significant.

This presentation will discuss the physiology of weight gain with various models and will outline the genetic factors contributing to the development of obesity.

National Genetic Blood Disorder Project (Student Screening) (1999-2002)

Shaikha Al Arrayed
Genetic department, Salmaniya Medical Complex, Ministry of Health, Bahrain

The inherited disorders of hemoglobin are the most common genetic conditions in humans. These include sickle cell disease (SCD), thalassemias (thal), and glucose 6-phosphate dehydrogenase deficiency (G6PD). These diseases cause serious health problems in the Arab countries in general, and in Bahrain in particular.

The student-screening project was initiated in 1999, and continued for the subsequent four years, with the aim of raising the awareness among the public. This paper presents the results of four years of screening (1999-2002).

Material and Method: It is a collaborative project between the Ministry of Health, Ministry of Education, and Bahrain Hereditary Anemia Society. The plan was to screen all the students in the 11th grade (2nd Secondary). A total of 7000 students were targeted every year. In the first year, the project took 10 months for planning, education, blood samples collection, laboratory testing, data processing, distribution of cards, data analysis, and reporting. During the subsequent years the project took 4-5 months every year. Consent for screening was taken from parents, and the response rate was 80-85% every year. The students were fully informed about these diseases through educational sessions. During the first year the total number of students screened was 5656. While the number was 5677, 6978, 5883 during the second, third and fourth year respectively. The total number of students screened until now is approximately 24,000 students. The number of schools involved was 38. During the first year, 120 educational lectures were given at schools, on the subsequent years the teachers and students manage to arrange the educational sessions and they requested assistance from the educational committee when needed.

More than 30,000 informational booklets were distributed annually. Technician visited the school to withdraw the blood samples and these were tested on the same day. High performance liquid chromatography (HPLC Variant machine, from Biorad) was used to perform the hemoglobin electrophoresis. The blood samples were also tested for blood grouping, and G6PD testing. A coded form was designed with the demographic information of each student. At the end of the campaign each student received a card with the lab results. Reports were also sent to schools M.O.H and M.O.E officials.

Results: Comparing the prevalence results during the years 1999-2000-2001-2002. The study showed that the Prevalence rate of Sickle cell disease among these students was 1.3%, 1.3%, 1.09%, 1.09% respectively. Sickle cell trait was found in 13.8%, 13.8%, 14.1%, 14.2% respectively. While Beta thal trait was found in 2.9%, 3.6%, 2.6%, 3.7% respectively, and G6PD in 23.2%, 23.4%, 21.9%, 20% respectively. The study included reports on the prevalence of these diseases by region, and by school. It also showed the level of hemoglobin S, F, A2, and H in each group.

Conclusion: We conclude that this project is vital for the community in order to raise the awareness. The Educational and screening campaigns which have continued for the last 18 years have showed its effect on the decline trend of the sickle cell diseases prevalence among Bahrain newborns.

Evaluation of the National Neonatal Screening Program in UAE. Analysis of eight years experience

Al-Hesani II, Salah M., Osman H., Rashid N., Saade D. and Al-Hassani A.

Newborn Screening no longer refers only to the screening tests themselves, but encompasses all the elements essential for every neonate to have access to a screening system that is optimal in terms of quality and performance. In UAE, the program started by screening for PKU in January 1995 and then Congenital hypothyroidism and sickle Cell diseases were introduced in January 1998 and January 2002 respectively.

Objectives: To evaluate the progress of the program, in addition to determine the incidence of PKU, Congenital Hypothyroidism and Sickle Cell diseases in UAE.

Material and Methods: Blood is collected on the fifth day by heel prick onto to the filter paper S&S 903. TSH and phenylalanine are assayed by Delfia fluorometric kits and the same filter papers are tested for Sickle Cell disease by HPLC system. For the evaluation, we used the coverage (%uptake), timeliness of screening program indicators (age sampling, time of delivery of the specimen to lab, time taken by the lab to produce the result, age of recall and age at treatment initiation) and unsatisfactory specimen quality against the international standards. Also we used our data to determine the recall rate, the apparent sensitivity, specificity, positive predictive value and relative incidence rates for PKU, CH and Sickle cell diseases.

Results: The coverage was good for most districts but is still low in the whole UAE. Timeliness indicators approximates the international standards except for delivery of specimens to the laboratory. Our protocol produced a high recall rate for congenital hypothyroidism and an acceptable recall rate for PKU. There was a satisfactory apparent sensitivity, specificity and positive predictive value for congenital hypothyroidism and PKU.

Conclusion: Screening for CH, PKU and Sickle Cell disease is worthwhile and the performance of the national screening program in the UAE is steadily improving and we aim to watch with international standards soon.

National Congenital Abnormality Registry (NCAR) in the UAE

Al Hosani H.; Salah M.; Farag II; Saade D.

Infant mortality rate in UAE has declined sharply from 11.4/1000 live births in 1990 to reach 8.4/1000 live births in 2000. The rate reached levels comparable to those of the more developed countries. However, congenital abnormalities accounted for about 75% of infant deaths in 2000 and the proportion increased from the previous 30% in the late 1980s.

CAs represent a special category of disorders with their earliest onset (birth) and the limited chance for complete recovery. Thus, there is only one optimal solution, it is the prevention. The prevention can be based on the real knowledge, i.e. the knowledge of causes. Etiological studies, however need appropriate CA datasets including baseline prevalence of different CA entities NCAR started at the national level since January 1999.

Objectives of NCAR: (1) The determination of baseline birth prevalence of different congenital abnormality entities; (2) To establish a priority list of preventive actions and measures.

Material and Methods: (1) Sources of cases: The goal of NCAR is to record all liveborn infants (from birth till the age of one year) Affected with CA. The main reporting sites of CAs are obstetrical inpatient clinics, pediatric inpatients and centers of Maternal and child health; (2) A special notification form was used to collect the desirable data related to the malformed case; (3) Coding: CAs as defined in the XIVth chapter of the 9th version of the International classification of Diseases (ICD), codes 740.0-759.9 with slight modification in multiple CA; (4) CAs were classified into isolated and multiple CA category; (5) Epidemiology studies (e.g. consanguinity, parity, family history, maternal age, sex, gestational age, birth weight, nationality, birth prevalence of different CAs against the international rate, etc*coding and diagnosis of CAs were critically evaluated in the department of Maternal and Child health at Ministry of Health, Abu-Dhabi.

Results: The total prevalence of reported cases with CAs was 7.1-15 per 1,000 live births. However, there was an evident difference in the prevalence rate in reported cases with CAs among the seven emirates due to underreporting of cases in some emirates. CAs of Cardiovascular system represented the most prevalent anomaly.

Conclusion: The high protection of infant mortality due to CA cannot be explained by the high absolute birth prevalence of CA in the UAE. It may be connected with the significant reduction of infectious diseases, nutritional deficiencies and environmental causes.

Spectrum of Genetic Disorders in Patients Referred to the Genetics Department of R.M.C. at Saudi Arabia: Cytogenetic and Clinical Experience for 8 Years.

Mona O. El Ruby
Human Genetics Department, National Research Center, Cairo, Egypt

A genetic component is evident in the ethiology of most human diseases. Genetic disorders pose a major health problem in the world In developed countries they represent the single largest cause of infant mortality loss while they are the 3rd - 5th cause of perinatal mortality in developing countries. Major efforts are directed towards lightening the burden of these disorders on patients, family and on the community at large In this study over a period of 8 years, 5397 cases were examined and studied for the presence of chromosomal aberrations and /or identifiable congenital malformations at the Human Genetics Department (HGD) of the Riyadh Medical Complex (RMC). Of the total 5397 karyotypes performed; genetic component was ascertained in 37.5% of the cases. Chromosomal abnormalities were detected in (22.1%). Down syndrome represented the highest chromosomal aberrations (15.4% of the total referred cases and 69.6% of the chromosomal aberrations) followed by trisomy 18 (1.1% of the total and 4.8% of chromosomal aberrations. their classification and prevalence are the primary objectives of this study. The high frequency of genetic disorders at the Middle East level emphasizes the prerequisites of national surveys and further studies to evaluate the influence of consanguinity, dietary, health awareness, social, cultural and other environmental factors on the prevalence of congenital and genetic diseases in different communities.

Eastern Mediterranean Approach to Genetic Disease Primary Prevention & Control

O M N Khatib
Regional Adviser, NCD/EMRO/WHO

Available data indicate that the health burden of genetic disorders is as significant in some Member States of the Eastern Mediterranean Region as industrialized world. Some of these disorders, particularly Haemoglobinopathies, are extremely common in this Region.

Recognising the increasing importance of congenital and genetically determined disorders in the Region, RO undertook a umber of initiatives to assist Member States in establishing programmes and implementing activities to prevent and control these disorders and improve health care services to the individuals and families affected. The epidemiological situation of hereditary disorders was initially reviewed and regional task force, set up by WHO in 1993, identified specific priorities for prevention based on the needs of countries of the Region. Subsequently, a Regional consultation on Community Genetics Services was held in the Regional Office in 1994 at which preventive strategies were discussed.

There is an urgent and growing need for systematic national and international initiatives and activities to reduce the health burden of congenital and genetic disorders. This was originally initiated at a meeting of a Working Group convinced at the EMRO of WHO in November 1995. in December 1999 a group of WHO advisors met in Cairo to review the global epidemiology of congenital and genetic disorders, identify evidence based interventions for their prevention and control, and recommended approaches for incorporating appropriate interventions into primary care at country level, and suggested coordination among regional centers.

The control of genetic and congenital disorders is a rapidly expanding field. There is a clear need for public health professionals and clinicians with knowledge and expertise in the prevention and control of these diseases. Coordination of preventive genetic counseling activities, among regional centers is needed. There is inadequate quantitative information about genetic counseling. Interested EM countries should recruit a national task group, and development of country plan, epidemiological studies to support planning and develop systems to start collecting background data on preventive genetic counseling services. Because of wide differences between EM Countries, the appropriate approach for such coordination is to implement a programme of work intended to support an organized approach to control of congenital disorders in any setting. A corresponding programme of selected experts, and representatives, in a long-term relationship. Such an approach may be recommended for consideration to all EM Countries.

Therefore it is now necessary to evaluate the present and likely future magnitude of congenital and genetically determined disorders within the Region, and to develop appropriate structures for their own prevention and control within the health care systems.

Study of Hemogloninopathy on 10000 Anemia Patients in Mazandaran Reference Laboratory 1997-2001

S, Abedian; S, Ghaseml; M Davari
Head of Laboratory, Iran

Background and purpose: Hemoglobinopathies is the most common genetic disorder in Mazandaran province of Iran.

In this study 10000 Anemia patients evaluated in 5 years.

Material and Methods: This study was prospective descriptive. H1 cell counter used for measuring of CBC, HbA2 measured by Ion exchange Chromatography. Sickeling test used for Hbs and also used. Electrophoresis on cellose acetate (PH=8.6)

Results: of this study shoed that from whole patients, 104 person contained HbF, 3015 contained increased HbA2, 189 contained sickle cell, 31 HbH, 1 HbE and 3 persin contained constant spring.

Conclusion: Due to research, there are rare hemoglobinse such as: HbE,C, CS,*. In Mazandaran province, that it need more research and investigation for providing of information bank (gene mutation).

The GCC Genetic Defects Registry: an Online KFSH & RC Initiative

Ali Al-Sanousi; Edward B. De Vol
King Faisal Specialist Hospital & Research Centre, Saudi Arabia

Healthcare will change more in the next 10 years than in the past 1000 years as a result of information technology. Rapid developments in electronic technology, the Internet, wireless computers, together with all forms of emerging biomedical informatics technology like genomics, nanotechnology and biomedical artificial intelligence, will radically alter the way health care organizations do biomedical research in the next decade. These driving forces of change will put new demands on health care leaders and researchers.

It is thought by many that Saudi Arabia has a large number of genetic diseases which are encountered more frequently than in western countries. This probably is due to consanguineous marriages that have been the custom for many years and to founder effects. Reliable data are scarce, however, for determining their prevalence. It has been reported that in some instances it may be 40-80 times higher than any other country in the west (Rashed et al 1994, 1998, and Ozand, 1998).

The national genetics defects registry will serve as a tool to maintain data and other information on patients with genetic defects and provide a scientific basis for various other research projects. It is anticipated that this effort will provide invaluable data for public health and genetic disease prevention programs of the Kingdom. So far, this particular information has not been available, except scattered among many databases of individual clinicians and scientists. This paper explores the initiative of the King Faisal Hospital & Research Center (KFSH&RC) for establishing a national genetic defects registry which will be expanded to include all the GCC countries. The goals are ultimately to significantly improve the quality of health care performance and enhance public health and preparedness. Through the intelligent application of Information and Internet technology, healthcare organizations can achieve lower costs, reduce medical errors, enhance outcomes and deliver health anywhere, while lowering.

This paper will enlighten the vision of the health care professionals attending the first GCC Genetic Conference in Bahrain in regards to the first web-based genetic defects registry technologies and how can we invest in medical informatics to take advantage of the existing strong ties among all GCC countries. The presenter will examine highlights of some success stories such as the GCC Cancer registry, the National Diabetes Registry, the KFSH Congenital Heart Disease Registry, all of which reside in the Biostatistics, Epidemiology, and Scientific Computing Department of the Research Center.

Keywords: Genetics, Genomics, Birth Defect, Registry, Technology, GCC, Biomedical, Informatics, Patients, Quality, GCC, and KFSH & RC

Premarital screening and aspects of relevance in Saudi Arabia

Mohsen. A F.EL-Hazmi
Professor & Chairman of Medical Biochemistry Department, Director Of W.H.O
Collaborating centre, College of Medicine, King Saud University, Riyadh, Saudi Arabia

Population screening Programmes are of value for prevention of genetic disorders that occur at an high frequency. The premarital screening is of particular significance to limit the birth of new affected children and can be offered for both, the common and the rare defects.

The major advantage of the premarital screening Programmes lies in the fact that the birth of an affected child is prevented, when couples found to be carriers of a defective gene are counseled and consequently adopt a decision not to marry.

In Islamic countries, other means of control, particularly prenatal diagnosis is associated with the dilemma of abortion of the affected fetus. Abortion is not acceptable from the Islamic point of view, except in special circumstances. The In Vitro Fertilization followed by Preimplantation genetic diagnosis, has significant value in primary prevention, but still a newly born field with several technical difficulties and lack of expertise in the region.

In Saudi Arabia, a recently adopted premarital screening programme at the national level was formally introduced. It is perceived that this programme will provide a major step towards prevention and control of genetic disorders among Saudi population. However, being voluntary, it requires a comprehensive and efficient awareness programme and the co-operation among all concerned at the levels of the individuals, the families and the community at large.

This presentation will highlight the major features of the premarital screening programme and discuss the pros and cons of this preventive measure in the Saudi community.

A framework for Ethics in Relation to Community Genetics in Islamic Countries

Mohsen.A F.EL-Hazmi; Argmand.S.Warsy
Professor & Chairman of Medical Biochemistry Department, Director Of W.H.O Collaborating centre, College of Medicine & Department of Biochemistry, College of Science, King Saud University, Riyadh, Saudi Arabia

Community genetics, is an area of medical disciplines, that responds to the health demand in relation to human genetics. As it is concerned with the synthesis of community ecology and population genetics, it has a significant impact on both on the individual and the society. With a wide view, the community genetics services involve

Programmes such as genetic screening, general preconceptional and prenatal consultations, of genetic management and congenital anomalies, and public education on genetic and related ethical issues

Scientific advancement and technical development in the field of laboratory diagnosis and medical intervention augmented the success of Community Genetics and highlighted variety of practical applications. Collateral several ethical issues linked to religious believes, life style and traditions prevailing in different communities came to light. Of relevance is the right of the fetus and infant to live, inbreeding marriages, comparative family size, life style among family members, cloning and preimplantation genetic diagnosis. The analysis of human genome and theories arisen from connection of human genome components to the pattern of inheritance and social life style, have led to global requirements to outline legal aspects and ethical principles in relation to diagnosis, prevention and health care. It is strongly believed that with special consideration given to Community Genetics, it is essential to weigh the pros and cons of the new technological advances and to establish the ethical guidelines, wi

The populations of Islamic/Arabic communities have several unique features, mainly due to their strong religious beliefs, and fairly respected traditions, which are strongly embedded in the practice of their daily life. These include large family size, combined family system, consanguineous marriage, which occur at an high prevalence. Thus ethical and genetic counselling issues have to cater for these aspects in the populations. In this regards, Islamic teachings cover the complete code of life and include ethical, moral and juridical issues. It specially emphasizes non-maleficence (i.e. avoiding and preventing harm to all), beneficence (i.e. to provide benefit or advantage to all) and justice (i.e. fairness and equity for all), confidentiality and shouldering medical responsibility.

This presentation will discuss the main issues of community genetic services and guidelines on ethics in relation to the services in Islamic Countries.

An Outline for Community Genetic Services for the Gulf- the Saudi Example

Mohsen.A F.EL-Hazmi
Chairman and Professor Of Medical Biochemistry, Director Of W.H.O Collaborating centre, College Of Medicine, King Saud University, Riyadh, Saudi Arabia

Community Genetic Services cover a wide range and multifaceted genetic- related requirements of the individuals, the families and the community. A penta-axial SETRR model appropriate to these demand includes Services, Education, Teaching, Registry and Research, is entertained as follows: (a) Provision of services that must reach the entire population in a country in order to provide equitable health services to all citizens. This involves establishment of genetic clinics and genetic centres, well-trained and knowledgeable family doctors, nurses and social workers and consultants; (b) education for the health care personnel, including the family doctors as partners in these services, (c) teaching to improve knowledge of the medical and non-medical personnel, patients, their families and the general public at large; (d) preparation of the Registry of genetic diseases in the population & (e) research to identify the molecular pathogenesis of the genetic diseases, establish more definitive methods for the identification and diagnosis of the genetic defect, establish appropriate management strategies and identify ways and means of prevention and control.

With the model in view, Community Genetic Services programme in Saudi Arabia were initiated during the early 1980s. The programme was initially directed towards blood genetic disorders, where these disorders occur at a high frequency in several regions of Saudi Arabia. A Tetra-step pyramid model was established with local, regional, national and international levels as a collaborative programme. The programme incorporated a National Working Group and W.H.O collaborating centre.

More recently, other genetic diseases were included and additional bodies i.e. the co-ordinating National Committee was conceived at the Ministry of Health, Riyadh. Consequently, several Programmes were adopted to improve genetic services through: (1) establishment of specialized centers and genetic clinics for care and management of patients; (2) provision of learning and training opportunities for health personnels by way of courses, workshops and conferences and (3) adoption of the health care personnel, scientists, patients, parents and the general public about genetic disorders by holding doctor-doctor, doctor-patient and patient- patient meeting.

This is a continuous process and new ideas and inputs from different related disciplines has been incorporated in order to achieve the ultimate goal of prevention and control. In this presentation, the Saudi model will be presented and proposals will be discussed to accomodate this model to the Gulf Region.

Genetic Metabolic Disorders of Saudi Arabia

Aida Al Aqeel
Consultant Paediatric Metabolist, Geneticist and Endocrinologist

Middle Eastern cultures are tribal and heavily consanguineous. Marriage between cousins has been part of the culture for millennia leading to "founder" effect and a large number of autosomal recessive disease. These ethnic groups did not mix with their neighbors, therefore, most of these disorders are either rare by Western standards or are unknown

A review of our files of the Armed Forces Hospital documented more than 150 varieties of neurodegenerative diseases among 2,000 children; 27 of which constitute more than half of these files.

In conclusion, our Saudi community is quite unique, because of the unique disorders seen in this community and the high prevalence of the genetic disorders especially neurometabolic disorders. The early recognition of these disorders is important to initiate early treatment especially in cases of organic academia and aminoacidemia to prevent neurologic crippling, and in lysosomal storage disorders to initiate early bone marrow transplantation or enzyme replacement therapy.

Medical Genetics: A physician Approach

Aida Al Aqeel,
Department of Paediatrics, Riyadh Armed Forces Hospital and King Faisal Specialist Hospital & Research Centre, Riyadh, Kingdom of Saudi Arabia

Medical genetics deals with the study of genetics of human diseases, with special emphasis on the diagnosis, counseling and management issues. We reported an autosomal recessive multicentric osteolysis in a Saudi Arabian family with distal arthropathy of the metacarpal, metatarsal and interphalangeal joints, which eventually progressed to the proximal joints and resulted in ankylosis and generalized osteopenia. In addition, they had large, painful to touch palmar and plantar pads and mild dysmorphic facial features including proptosis, a narrow nasal bridge, bulbous nose and micrognathia.

Using a genome-wide search for microsatellite markers from 6 members of this family, localized the disease gene to chromosome 16q12-21 with a LOD score of 4.59. Haplotype analysis with additional markers narrowed the critical region to 1.2 cM between markers D16S3032 and D16S3140 and identified the matrix metalloproteinase 2 (MMP-2, gelatinase A, collagenase type IV, EC 3.4.24.24) gene as a disease candidate. All affected individuals were homozallelic for a nonsense mutation (TCA>TAA) in codon 244 of exon 5, predicting the replacement of a tyrosine residue by a stop codon in the first fibronectin type II domain (Y244X) leading to no MMP-2 enzyme activity in serum and/or fibroblast of affected individuals.

Conclusion: The discovery that deficiency of this well characterized gelatinase/collagenase results in an inherited form of an osteolytic and arthritic disorder provides invaluable insights for the understanding of osteolysis and arthritis and the in vivo function of MMP-2.

Haematopoietic Stem Cell Transplantation for Inherited Disorders in the Sultanate of Oman

David Dennison; Jalil Ur Rehman, Nadeem Nusrat; Fehmida Zia, Salam Al Kindi; Krishnaratnam Kannan; Anil Pathare, Shahina Daar; S Muralitharan; Brian Christie; Ayesha Al Maamari; Shoaib Al Zadjali; Leonard Brown
Department of Haematology, Sultan Qaboos University Hospital, Oman

Allogeneic haematopoietic stem cell transplantation (SCT) has become well established as curative treatment for a variety of inherited haematologic and immune disorders. Bone marrow transplant programmes in countries where the prevalence of such disorders is high would therefore find hereditary diseases to be a major transplant indication. This has been the case in the Sultanate of Oman. Between June 1995 and December 2002, a total of 75 patients underwent SCT in the Sultan Qaboos University Hospital. Genotypically HLA matched siblings were used as donors in 73 patients, a phenotypically HLA identical parent for one and a HLA haplo-identical father for one patient with severe combined immunodeficiency syndrome. Of the 65 patients, 37 (57%) were transplanted for inherited disorders. Specifically these disorders included the following: homozygous beta thalassemia 27 (73%), sickle cell anemia 2 (5%), Fanconi anemia 2 (5%), chronic granulomatous disease 2 (5%), Glanzmanns thrombasthenia 1 (3%), hemophagocytic lymphohistiocytosis 1 (3%), Blackfan Diamond syndrome 1 (3%) and severe combined immunodeficiency syndrome 1 (3%). With the exception of one patient with sickle cell anaemia who died of sepsis, and one patient with homozygous beta thalassemia who rejected her graft and become transfusion dependent again, all other patients 95% (35/37) are alive and well and cured from their disease 3 months to 7 years post transplant. Apart from providing cures for these patients, the transplant programme in Oman has opened up avenues in research for the basic biology of inherited disorders unique to this region and has led to the development of newer tests for the screening of these diseases.

Anatomical and Developmental Study of Congenital Anomalies of Nervous System in Fetus and Newborn in Zeinab University Hospital in Mashad from 2001 to 2002

M. Pourjavad
Mashad Medical School, IRAN

Objective: Anomalies of the CNS are among the most prevalent congenital defects,yet the reason of their occurrence has not been well understood. Our purpose in this study was to consider the anatomic malformation and the explain the possible embryonal reasons for anomalies of nervous system in humans,in mashad.

Design: In this study, we have taken into consideration the anatomic, statistical condition and embryonal explanations in infants who have been born by vaginal or abdominal deliveries during one year in hazrat zeinab university hospital.

Materials and methods: During one year 3697 vaginal or abdominal deliveries were performed,of which 21 cases sufferred from nervous system anomalies and some other craniofacial defects.all cases were sent to the anatomy department and were fixed in liquid fixative. Weight, length and the type of anomaly with some of its anatomic descriptions were recorded information.. Pictures were also taken from most of the cases.

Results: In this study,anencephaly (0.35%), hydrocephaly (0.16%), meningocele (0.05%) were seen that in most cases were accompanied by craniofacial anomalies (separation of eyes, cataract, deformity in head and face).

Conclusions: Considering social and experimental problems, probably one of the agents producing these defects is addiction and abuse of narcotic substances in critical stages of nervous system formation.other influencing factors can be malnutrition, vitamin deficiencies and some medications.in molecular aspect, another factor involved in nervous system defects is sonic hedge hog morphogene which is inhibited by different conditions and factors and therefore, pirotduces these anomalies. Anatomal and histochemical studies of these defects are underway.

Key words: nervous system defects,anencephaly,hydrocephaly,sonic hedge hog morphogene

Declining Trends in the Incidence of Sickle Cell Diseases in Bahrain

Shaikha Al-Arayyed, A Aziz Hamza, Bema Sultan*, D. K. Shome, J. P. Bapat.
Genetic Department, Salmaniya Medical Complex, *Ministry of Health, Kingdom of Bahrain.

Introduction: Genetic blood diseases are frequent in Bahrain. Previous neonatal screening in 1986 showed that the incidence of sickle cell disease (SCD) is 2.1% and that of sickle cell trait (SCT) is 11%.In 1984 the Ministry of Health (MOH)instituted a campaign for the prevention of these disorders. Measures included community education together with screening programs (antenatal, newborn and school screens) and premarital counseling. The incidence has been falling gradually during the years. A premarital study done in 1995, showed SCD incidence of 1.6%. Students screening done in the years 1999, 2000,2001,2002 showed incidence figures of 1.2%, 1.3%, 1.09% and 1.09% respectively.

Objectives: To update the national data on the incidence of sickle cell disease among the newborns and to compare with the previously available data.

Material and Method: Target Group: All Bahraini newborns delivered at the (MOH) maternity hospitals for a period of three months from February to April 2002. Automated high performance liquid chromatography (Variant, BioRad) was used with the beta-thalassemia short program to analyze cord blood samples. A questionnaire was filled for each new born. This included demographic data, parental age and consanguinity history. These forms were coded for computer analysis.

Results: Two thousand Bahraini newborns constituted the study population. Eighteen were affected with SCD with an incidence of 0.9%. SCT was found in 16%. SCD was found in 1.92% of the fathers and 1.3% of the mothers whereas SCT was detected in 13% of the fathers and 14% of the mothers. Glucose 6-phosphate dehydrogenase deficiency was found in 18% of the male newborns, and 10% of the female newborns. Other parameters: (a)Gender: male to female ratio among the sample group was 1.1. (b) Age: The paternal age distribution showed that 87% of the fathers had children between 25-44 years of age. Mothers were younger with 82% falling in the age group of 20-39 years. (c) Consanguinity rate was found to be 20% (12.5% first-cousins and 7% far relatives).

Conclusion: Bahrain has for the first time recorded a less than one percent birth rate of babies with sickle cell disease. If this trend continues, Bahrain could become the first country in the Gulf region to eliminate this disease.

Pharmacogenetics and Pharmacogenomics: Perspectives towards individualized drug therapy?

R. Krishnamoorthy, Koumaravelou K, Gerard N, Adithan C *, Srivastava A**
INSERM U458, Hopital Robert Debre, Paris, France. *JIPMER, Pondicherry, India. **CMC Hospital, Vellore, Tamil Nadu, India.

For both clinicians and patients it is clear that one drug does not fit all and one dose does not suit all. Individual variability in both therapeutic efficacy and drug toxicity is common in the clinical setting as well as in the clinical trial during the development process of a new drug. Some subset of patients gain no benefit from therapy while others experience adverse drug reactions. A number of factors contribute to such interpatient variability in drug response which include gender, age, nutrition, disease condition and environmental components, even presumptively trivial. Importance of the involvement of genetic factors and ethnic differences in such variability is more and more recognised and gain particular importance in the post genomic era. Our human genome consists of 3 billion bases and we are all identical by 99.9 %. The difference of 0.1 % translates into potentially 3 million base changes and a significant percentage of such changes contribute to the

Correlation of a Novel Perforin Gene Deletion Mutation and Flowcytometric Detection of Perforin in an Omani Patient with Familial Hemophagocytic Lymphohistiocytosis: Implications for Rapid Diagnosis

Dennison D, Muralitharan.S, Christie B, Wali.Y, Zachariah M, Romana M, Mamaari A, Al Said B, Mamaari S, Belushi T, Krishnamoorthy R and Al Lamki Z.

Familial Haemophagocytic Lymphohistiocytosis (fHLH) is an autosomal recessive disease of early childhood manifested by hypercytokinemia, organ infiltration of macrophages and activated lymphocytes, and characterized by a fulminant clinical course. The molecular mechanism appears to be a disorder in the regulation of apoptosis. This is evidenced by the fact that approximately 40% of patients with fHLH reported worldwide, have perforin gene mutations. We report in this paper, a unique 12 bp deletion mutation in the exon-3 of the perforin gene in a 2 month old Omani boy who was diagnosed for fHLH. Using direct DNA sequencing, the 12 bp deletion was found in a homozygous state in the patient and confirmed to be heterozygous in both parents. The deletion mutation completely removes four codons (codon 284 to 287) but maintains the reading frame in the membrane attack complex domain of the protein. Flowcytometric analysis of the intracellular perforin protein in the patient's NK cells showed reduced levels. The positive correlation of decreased intracellular perforin in our patient implies that flowcytometry may be of use in the rapid diagnosis of patients suspected to have fHLH. More studies however need to be done on patients with other perforin gene mutations to determine the true utility of this test.

Myogenetics, France and the Middle East

J. Andoni URTIZBEREA* ; Djamel GRID, BTR-Généthon**
* Institut de Myologie, Hôpital de la Salpêtrière, Paris, France. ** Institut de Myologie, Hôpital de la Salpêtrière, Paris, France

Nowadays neuromuscular diseases comprise at least 130 different conditions characterized by their high phenotypic and genetic heterogeneity. Since the discovery of the gene encoding the dystrophin by reverse genetics in 1987, the majority of myopathic or neuropathic entities so far described on clinical and pathological grounds have been elucidated from a genetic view point. The so-called limb-girdle muscular dystrophies for instance, irrespective of their mode of inheritance, are caused by over 15 distinct genes many of which do not share any functional relationship. In hereditary sensory motor neuropathies, polymorphism is even worse with more than 26 disease causing genes for both axonal and demyelinating forms.

Neuromuscular disorders should also be regarded as a public health issue especially in endogamic populations. In some countries of the Gulf region, it represents up to 25% of all referrals for genetic counselling. In the long-run, identification of genetic defects in appropriately investigated patients will be a prerequisite to any curative clinical trials.

European scientists, notably those from France, played an important role in sorting these genes out. This was achieved by fruitful interactions between geneticists and clinicians, and between groups from Europe and isolated clinicians spanning the Middle East up to the sub-Indian continent. However, a number of logistic and biotechnicological hurdles make these collaborations sometimes difficult. DNA extraction and storage as well as sophisticated diagnostic tools such as muscle immunostainings, are not available everywhere. Conversely and due to growing financial contraints, a number of European laboratories are becoming reluctant to process DNA samples for routine analysis after the gene of a rare disease is found. That is why transferring the technology to developping countries is an urgent need for both sides. It has been done successfully for Duchenne muscular dystrophy and spinal muscular atrophies (SMA) but many more orphan diseases are still on the waiting list.

In parallel, mapping projects are still under way in a couple of ultra-orphan conditions: distal SMA, Brown-Vialetto-Van Laere syndrome, congenital myasthenic syndromes, merosine-positive congenital muscular dystrophies, and many others. For all, international cooperation is required to pool informative families and fuel collaborative linkage studies.

In the regional and demographic context of myogenetics in the Gulf, building up an alliance of geneticists and patients/families for rare diseases would be an important step forward. Given the paucity of laboratories and expertise on the one hand, and the extreme variety of genetic diseases on the other, some regional cooperation is an absolute necessity.

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Wolfgang Trautwein
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Attitudes of Medical Personnel to Ethical Issues in Iran

D.D. Farhud [1,2], N. Nikzat [2] ; H. Sadighi [2]
1. WHO Ethical Committee Geneva, Switzerland. 2. Dep. of Human Genetics, School of Public Health, Tehran Univ. of Medical Sciences, Tehran-Iran

A WHO meeting of experts in genetics was convened in Geneva, Switzerland, in december 1997, to review proposed international guidelines on ethical issues in medical genetics and genetic services. The medical application of genetic knowledge must be carried out with due regard to the general principles of medical ethics; *Autonomy, Beneficience, Non-malficience, Proportionality* and *Justice*.

Human genetics with its advances, specially in the last two decades, has created new ethical, legal and penal issues.

This study was carried out with the purpose of obtaining points of view of 756 physicians, nurses, midwives and common people, with regard to ethical principles in medical genetics. The study was performed by questionary method and the descriptive and analytical assesment was accomplished on the results. The results showed that the application of these ethical principles in health care, have been observed with different views.

According to these views, the principles were categorized based on priority of acceptance: 1- Proportionality; 2- Beneficience; 3- Justice; 4- Autonomy and 5- Non-maleficience.

Analytical assesments suggest that a number of personal, cultural and social variables were taken in to account and the relationship between negative views and the variables were assessed.

Autonomy, with age and marriage statues, have shown statistical significance. More adults than middle aged, and more singles than married individuals, responded negatively. For beneficience, the variable of profession has shown statistical significance. Non-malficience and proportionality showed to be not statistically significant by the variables. Residents of Tehran produced more negative responses to justice than those living in other cities.

Mutation Detection and Prenatal Diagnosis of Patients With Cystic Fibrosis (CF) in Iran

P. Derakhshandeh-Peykar [1,2], M. Rahmani[2], M. Atai[2], D.D. Farhud [1,2]
1Dept. of Human Genetics, School of Public Health, Tehran Univ. of Med. Sciences, Tehran, Iran. 2. Genetic Clinic, Valie-Asr Sq. 16, Keshavarz blvd. Tehran, Iran

Cystic fibrosis (CF), the most common severe lethal autosomal recessive disorder in whites, is caused by mutation in the CF transmembrane conductance regulator gene (CFTR) on chromosome 7q31. The carrier frequency among Caucasians is approximately 1 in 25, with an incidence of approximately 1 in 2500 live birth. The gene for CF spans approximately 250 kb and contains 27 exons. CF patients have two defective alleles and may either be heterozygous for different mutations, or homozygous for one of the mutations.

Since the identification of the gene responsible for CF, more than 900 mutations was described in CFTR gene of patients affected by cystic fibrosis, but the prevalance of the CF shows a geographical and ethnical variations in the world. The ΔF508 mutation in CFTR gene accounts for over 70 % all mutant CFTR alleles in the Europeans to 20 % in the Asians.

The DNA samples of 24 individuals, who were carrier for CF, partly with their affected children and their chorion villus samples (CVS) from the pregnant women, have been tested for five common mutations: ΔF508, G551D, G542X, W1282X and N1303k. The study was performed by using the ARMS method for mutation detection of CF gene. The ΔF508 mutation was found only in one couple with first cousin marriage and three times in other partners of related/non related couples (28%). One of these couples were heterozygous for two different mutations (ΔF508 and G551D). Results from this study revealed the following frequencies: ΔF508: 28%, G551D: 5%, G542X: 5%, W1282X: 0%, N1303k: 0%.

Photoanthropometric Investigation of Facial Structures in Iranian Children with Down Syndrome and Normal Control

Farhud D.D.[1]; Mahmoudi M.[2]; Derakhshandeh-Peykar.P.[1] Stengel-Rutkowski S.[3]
(1) Dept. of Human Genetics, School of Public Health, Tehran Univ. of Med. Sciences, Tehran, Iran. (2) Dept. of Anatomy, School of Medicine, Univ. of Babol, Iran. (3) Genetic Diagnosis and Counselling Department in the Munich Child Center,Institute of Medical Genetics, University of Munich, Germany

The photoanthropometric method suggested firs by Stengel-Rutkowski et al. (1984) was used to study the facial features in 136 Iranian children with Down syndrome, aged 4 to 14 years. Nineteen parameters were investigated and compared to an age related control group of 100 normal Iranian children. The obtained measurements were related to reference values in the same faces. The normal range was defined by age related index values between the 20th and 80th percentile in the collective of normal Iranian children. Five parameters were considered as characteristic facial traits of Iranian children with Down syndrome by index values outside these percentiles in 50% of the studied collective: low midface; narrow, upslanted palpebral fissures and short, anteriorly rotated ears. Twelve parameters were considered as additional facial traits by index values outside these percentiles in 30% of the studied collective: broad inner canthal distance; prominent nose root, short nose back; everted nasal base; long nasolabial distance, forwards inclined integumental upper lip; narrow mouth fissure; high, prominent chin; high-set, narrow ears and narrow conchae. These results contribute to an objective definition of facial traits in children with Down syndrome in a homogeneous ethnic population.

Deletion Analysis and Prenatal Diagnosis in Iranian Spinal Muscular Atrophy Patients Type I- III

P. Derakhshandeh-Peykar[1,2], M. Esmaili[1,2], M. Rahmani[2], M. Atai[2], J. Ghassemi[2], D.D. Farhud[1,2]
(1) Dep. of Human Genetics, School of Public Health, Tehran Univ. of Medical Sciences, Tehran-Iran (2) Genetic Clinic, Valie-Assr Sq. 16, Keshavarz blvd. Tehran-Iran

Autosomal recessive spinal muscular athrophy (SMA) is after cystic fibrosis, the second most common fatal monogenic disorder and after Duchene muscular dystrophy is the first common severe neuromuscular disease in childhood. The disease is characterized by degeneration of anterior horn cells leading to progressive paralysis with muscular athrophy. Depending on the clinical type (Werdnig-Hoffman=type I, intermediate form=type II and Kugelberg-welander=type III), SMA causes early death (type I) or increasing disability in childhood (type II,III). All three types of autosomal recessive SMA map to chromosome region 5q13.1. Homologous deletions in exon 7 and 8 of the survival motor neuron (SMN) gene have been described in >90-95% of SMA patients.

The aims of this study were to screening the deletions of SMA gene (exon 7 & 8) in iranian patients for prenatal diagnosis of SMA. We have studied 26 families with SMA types I-III, partly with their affected children and their chorion villus samples (CVS). DNA deletion genotypes were determined by PCR-RFLP analysis amplifying exons 7 and 8 of SMN (survival motor neuron). Results released the homozygous deletions of exon 7 and 8 of the SMN gene in 23/26 (90%).

The data support that homozygous absence of SMN exon 7 & 8 is strongly associated with SMA. The percentage of homozygous deletions in the study is almost as high as that reported by oder researchers. This method is useful, fast and effective for gene diagnosis and prenatal diagnosis of SMA.

Sickle Cell Disease Among Bahraini Children

Hussein Al Mukarreqr

Aim: To evaluate the common manifestation of sickle cell disease in children to know its behavior among Bahraini Population.

Methods: One year records of the admission at pediatric department had been reviewed retro respectively.

Result: Total number of admission over one year in the pediatric department were 4552, sickle cell disease patients were 552 (12.1%) percentage. There were 60% male and 40% female and the age distribution were 0-4 years 23.9%, 5-9 years 33.5% and 10-14 year 42.6%. Vasoocclusive crises represent 69.3%, fever 11.7% hyper hemolysis/ severe anemia 9.1%, acute chest/pneumonia 6.5%. Gastroenteritis 2.7% Osteomyelitis 2.2%. The cerebrovascular accident, sequestration and hand food syndrome were each less than 1%.

Conclusion: Vasoocclusive crises is the commonest manifestation of sickle cell disease where other complication were relatively less common in comparison to other SCD haplotypes.

Endocrine complications of B-thalassemia Major in Bahrain

Najat Mahdi, Akbar Mohsin, A.Jabbar Al.Abbasi, Shaikha Al.Arrayed

Objectives: To find out the frequency of endocrinopathies among Bahraini B-thalassemia patients.

Methods: 105 patients with B-thalassemia major were studied with age range of 6 months-40 years. All of them are followed up in the pediatric hematology clinic and are on regular blood transfusion. Those patients with serum ferritin level above 1000ng are on regular desferal therapy.

Results: We found that the commonest endocrinopathy is growth retardation followed by hypogonadism, hypoparathyroidism and impaired glucose tolerance test.

Conclusion: Patients with thalassemia have to be investigated regarding endocrinopathies as many of them suffer from these complications.

Clinico Hematological profile with Children of Sickle cell Disease in Bahrain

Hussain Al Mukareq

Aim: To determines the clinical and hematological parameter of children with sickle cell disease in Bahrain which could be used as base line reference value.

Patient & Methods: Retrospective review of ~50 files of children with sickle cell disease attending pediatric clinic at SMC, they were all asymptomatic.

Results: A total of 70 files reviewed and 20 were excluded and the remaining 50 enrolled in the study. The study population divided into 2 group according to age: Group I 1-7 year and group II 8-15 year. The male is predominating in both group 52% and 68% respectively, there is no difference in the mean hemoglobin. Although the MCV and MCH is lower in both groups yet it is lower in group I. The retics counts is higher in group II (4.9% compared 3.9%). The mean WBC and platelets counts were both high in group I. The HbS is higher in group II (78.4% VS 65.8%), the HBF higher in group I (25.9% VS 17.7%). The G6PD deficiency was higher in group I (42.1% VS 35.2%). The jaundice and splenomegaly were higher in group II(84% and 56% respectively).

Conclusion: (1) The low MCV and MCH in both group could be explained by the CO inheritance of alpha thalassemia or iron deficiency anemia. (2) HbS, jaundice and splenomegaly is higher in older age children; (3) The G6PD deficiency is a common inherent among patients with sickle cell disease.

Hemoglobin H disease in Bahrain

Hussain Al Mukareq

Objectives: To study the clinical and Hematological behavior of Hb H. disease among Bahraini children.

Patient Methods: 26 children aged 1-17 years were retrospectively studies. The diagnosis of Hb H disease confirmed by Hb electrophoreses and HB H inclusion bodies, clinical assessments based on the severity of anemia, requirements for blood transfusion and splenic enlargement and growth para meters.

Result: There were 11 male and 15 females and the mean age at diagnosis was 6.7 years. The mean Hb was 8.1g/dl and the mean Hb H level was 17.4%. Thirteen (50%) required blood transfusion. Seventeen (65.4%) had average growth and nine (34.6%) were below average. There were no significant thalassaemic bone change or splenomegaly.

Conclusion: Hb H disease in Bahrain children is phenotypically mild to moderate.

The Genetic services at the Ministry of Health in the Kingdom of Bahrain (1984-2002)

Shaikha Salim Al Arrayed

Hereditary disorders are common in Bahrain, as in all other Middle Eastern countries. The hereditary disease program in Bahrain started in 1984 by the opening of a genetic unit at Salmaniya Medical complex. In 1991 a National Committee was formed by Ministerial order to study the genetic diseases, and to put a plan aiming at controlling these diseases, together with improving the standard of management and treatment of patients suffering from such diseases.

The unit performed epidemiological studies on the common genetic diseases. These included studies on blood genetic diseases, congenital abnormalities, metabolic disorders among newborns, chromosomal abnormalities, cystic fibrosis, and on some of the frequently seen genetic syndromes.

Regarding the Health education program: Many educational booklets and leaflets were prepared and distributed to increase awareness of the public. Many educational sessions were given to different groups including medical, paramedical and public. These educational venues included: symposia, lectures, school health education campaigns, educational exhibitions and competitions.

The Genetic unit and the Maternal and Child health department worked jointly to establish the service of premarital counseling in 1992. Training courses were organized for all family physicians. A specific risk assessment sheet was prepared.The service started in 1993 on a voluntary basis. During the first few years only 20% of the married couples seeked premarital counseling. In the year 2000 The Shura council approved a recommendation to issue a mandatory decree to make premarital counseling a necessity by law, as part of legal requirement in the marriage documentation. The Bahrain ministers' cabinet approved that in 2001. The couples seeking advice are counseled, and they have the freedom to accept that advice or not. Couples at risk have to be referred to Genetic clinic.

Students screening: The project started in 1998 as collaborative project between MOH, MOE. It was partially funded by NGOs. The targeted group was students at high schools (11 grade students). It is now running in its fifth year. Nearly 25000 students had been screened. An ID card with the results had been issued to each student. The project proved to be very popular among students and families, and it resulted in tremendous raise in the awareness among the community.

Genetic registers: Two separate registers were established. The first is a birth defects register that was established in collaboration with WHO in 1999. This register covers infants born with anatomical birth defect. The second is a registration of all patients with genetic diseases referred to the genetic unit.

Genetic laboratory: The Cytogenetic laboratory was established in 1999, and it serves all the hospitals and clinics in Bahrain. The molecular genetic laboratory was established in 2000. It performs molecular testing on genetic blood diseases such as sickle cell diseases, alpha thalassemia, beta thalassemia and glucose six phosphate dehydrogenase.

With the support and the encouragement of health planners, we are looking for the success of these prevention programs in controlling the genetic diseases in the country.

Sever Multiple Synostoses, a New AD Syndrome, in a Large Iranian Pedigree, Exclusion of the First Locus on Chromosome 17q21-q22

D. D. Farhud [1];Tayebeh Rezaie [2]; Ali Nasiri [3]; Malek Demirtas [4]; Nurten Akarsu [4]; Mansoor Sarfarazi [2]

(1) Department of Human Genetics, School of Public Health, Tehran University of Medical Sciences, Tehran, Iran. (2) Surgical Research Center, Department of Surgery, University of Connecticut Health Center, Farmington, CT, USA. (3) Dept. of Orthopaedics, School of Medicine, Tehran University of Medical Sciences, Iran. (4) (TUBITAK) DNA & Cell Bank and Gene Research Laboratory, Hacettepe University, Child Health Institute, Sihhiye, Ankara, Turkey.

Multiple synostoses syndrome (SYNS) is an autosomal dominant congenital limb malformation presenting with carpal-tarsal synostosis, joint ankylosis, symphalangism, conductive deafness, and spinal and craniofacial abnormalities. A locus (SYNS1) has previously been mapped to chromosome 17q21-q22 for this phenotype and disease causing mutations have been described in the Noggin gene. Overlapping phenotypes, such as proximal symphalangism (SYM1) and tarsal-carpal coalition syndrome, have also been mapped to the same region raising the possibility that different phenotypes within this spectrum may result from the same genetic defect. In this study, we report a large Iranian kindred with severe carpal, carpometacarpal, tarsal, and tarsometatarsal synostoses, brachydactyly, humeroradial synostosis, and proximal symphalangism. The findings are mostly compatible with Pearlman syndrome, but considerable overlap also exists with other multiple synostosis phenotypes. Genetic linkage analysis of this family has excluded the locus on chromosome 17q21-q22, thus providing evidence for genetic heterogeneity and, possibly, a new basis for reclassification of this group of phenotypes.

7- Carpenter syndrome in eight Arab patients,Dominant inheritance suspected

Shaikha Al Arrayed

Carpenter syndrome is rare autosomal recessive syndrome. The diagnostic criteria are acrocephaly, Poly dactyly and syndactyly of fingers and toes. Some patients have congenital heart disease hypogonadism, mild obesity and frequent mental retardation. Thirty - five cases had been reported worldwide, since 1961. We are reporting eight cases with carpenter syndrome, four alive and four diseased form two families. In both families parents are consanguineous. All patients have typical features of carpenter syndrome. All the patients have mild mental retardation. None of the patients have congenital heart disease. The karyotypes in all of them are normal. The father in one family has some of the syndrome features such as, hepertelerism, acrocephaly, epicanthusfold, but without polysyndactyly, and with normal intelligence.

Conclusion: In this family the consanguinity supports autosomal recessive inheritance. The face that the father shows some of the syndrome features may indicate either condominant inheritance or dominant inheritance.

Familial Chromosomal Abnormalities

Shaikha Al Arrayed
Genetic department, Bahrain

Introduction: chromosomal abnormality can be inherited and can cause recurrent fetal wastage and abnormal children. Method: cytogenetic studies were performed on Bahraini patients suspected of having chromosomal abnormalities. Result: we are reporting on ten families inheriting different chromosomal abnormalities.

Family No 1: one child and sex abortions. The chromosomal analysis showed interesting results. Both wife and husband have different chromosomal abnormalities. The wife has 45,XX,der (13;14)(q10;q10). The husband has 46, XY, inv (3) (p25q21). Their child is normal. He inherited the abnormal chromosomes from both parents.
Family No 2: have three mentally retarded children with 47 chromosome Trisomy 21.
Family No 3: a male baby has 46, XY, rec(8), dup p.inv (8) (p23.1q24.2). he inherited the abnormal chromosome from the mother.
Family No 4: the patient is married to her first cousin. She is 4p3D2A1L1. both wife and husband have the same balanced reciprocal translocation 46XX,t (6;10) (q15;q21.2) for the husband. Their retarded baby girl has the same chromosomal abnormality.
Family No 5: the family has one anencephalic stillbirth and an abortion. The husband has apericentric inversion of the Y chromosome.
Family No 6: the family has two children with trisomy 21, the mother has mild Down syndrom features.
Family No 7: four children are mentally retarded with fragile X syndrom. The father is the transmitting parents.
Family No 8: mentally retarded child with fragile x syndrom. The mother and five of her sisters are carriers for fra X. the grandfather is the transmitting parent.
Family No 9: father karyotype is 46.XY, inv.per. (2) (p12;q14). He has balanced pericentric inversion of chromosome 2. his first child is abnormal with trisomy13.
Family No 10: the father has 46,XY/46,XY,rea (2). The family has three abnormal mentally retarded children and two abortions.

Conclusion: this indicates that inherited chromosomal abnormality is not a rare condition, and that cytogenetic studies should be done for any family with abnormal children and with recurrent fetal wastage.

Student Screening Project: The management of School Campaign

Mariam Al Mulla Harmas
Consultant family physician, School health coordinator, Ministry of health, Bahrain

Introduction Hereditary blood disease (HBD), being common and chronic, are the major problem of our community. As reported in 1996-1998 statistics, hereditary blood disease (HBD) were ranked the second highest among the ten common disorders recorded at Salmaniya medical complex, Bahrain.

Project Proposed Hence the ministry of health and education in collaboration with the Bahrain National Hereditary Anaemia society (BNHAS) and the Rotary club of Manama established the National project for the prevention of HBD in 1999.

Target Population: 11th grade students attending government secondary schools and the private schools.

Project Description: The project consists of three phases.
Phase 1: Mass media education about the HBD, the importance of the premarital counseling and blood testing was done through a structured campaign that targeted school teachers and students of all secondary schools. This was followed by another educational session to all grade 11 students by the trained students.
Phase 2: With the parents consent, a team of laboratory technicians collected blood samples from the students of the secondary school. The community response was very positive and encouraging (nearly 85%). Hemoglobin electrophoresis, blood grouping and G6PD activity were tested.
Phase 3: After analyzing the blood samples the professionals from the team identified the carries who are at major risk of giving birth to diseased children if they get married to a carrier or diseased person. Every student is given a personal card about the blood results and a specific booklet about their illness (if it is positive).
All the cards carry a common message "if you are a carrier of diseased, make sure to be engaged to normal person"

Outcome of the Project: This project has raised the community awareness to the problem of the hereditary blood disease and the importance of premarital counseling.

The Control of Thalassaemia in Cyprus - Some Ethical Considerations

Michel Angistiniotis
Cyprus

Programs of community control of genetic disease have proven to be successful in limiting new affected births, the Thalassaemia programs being the main examples.

The successful programs were characterized by planning and implementation of various components, such as health education, screening for carriers, counseling and prenatal diagnosis. In the implementation of these components bioethical issues arose which must be considered as new methods of prevention are adopted.

The bioethical issues which are universally accepted, independent of culture are:

- Informed choice
- Free choice

Full information implies a public educated in genetic principals as well as being well informed on the nature of the disease to be prevented.

Informed choice also depends on the quality and timing of genetic counelling given to individual couples. Freedom of choice starts from freedom to choose to be screened and then having alternative solutions to choose from. The couples before marriage have the most choices i.e. avoiding marriage, donor sperm from non-carrier, prenatal diagnosis, pre-implantation diagnosis. If the program included antenatal clinic screening then a woman whose risk is recognized when already pregnant has the least choices i.e. either to have prenatal diagnosis, or to take the chance and keep the pregnancy or to terminate.

Planning needs to keep all these choices freely available without directional counseling, but cultural, religious and legal considerations in each country many lead to difficult choices.

Goals, strategy and achievements of the World Alliance of Organizations for the Prevention of Birth Defects

Ysbrand Poortman
Sweden

This presentation will provide information on the goals, strategy, activities and achievements of the World Alliance (WA).
This Alliance is a unique joint venture of scientific groups, international discipline bound societies and patient alliances with the aim to contribute to the reduction of birth defects by a structural approach.
The WA started in 1994 on the initiative of the March of Dimes in New York and decided to focus on developing countries and the exchange of information and experience.
The activities of the Alliance are based on the exertions of its members.
The Alliance organises, as a rule in conjunction with major regional congresses, annually a specific theme bound conference leading to position papers, policy statements or declarations. Such meetings took place in Berlin, The Hague, Rio de Janeiro, Geneva, Cape Town, Kiev, Amritsar, Riadh, Matsue, Amsterdam, Vienna, Johannesburg and Toronto.
Some conclusions and recommendations of these meetings read as follows:

- need for official/governmental recognition of the burden imposed by genetic disorders and birth defects
- need for political will and commitment
- improvement of epidemiological knowledge
- education of the public and health officials in genetics
- organization of genetic services in a comprehensive and integrated manner with roots at the primary healthcare level
- build or improve peri natal and pre natal services
- build or extend genetic services based on individual family well- being and public health
- encouragement of the formation of parent/patient organizations.

Genetic Alliance of Parent/Patient organisations

Ysbrand Poortman
Sweden

Increasingly patients and their families form local, regional and international networks. They are organised disease bound and subject (genetics, handicap/disability) bound.
This presentation will explain the policy and activities of the genetic & research oriented groups in general and present examples of the achievements on some of these groups.
They focus on healthcare, research, ethics and information/education and wish to collaborate with medical and scientific groups. They strive internally at empowerment of the families involved to enable them to cope with the disease related problems and externally at influencing healthcare policies on the national and global level.
They contribute to the setting up of disease specific consortia, networks of expert centres, protocol development, rare disease programs, national awareness campaigns and public debate on genetic topics and dilemmas.
Increasingly they realise their role and (potential) influence in the political arena and towards science, industry, financiers and society at large.
These groups have recently formed an International Genetic Alliance which communicates with WHO and UNESCO. The charter, the policy and program of the alliance will be presented. They have their bi annual meetings in conjunction with those of he Global Life Sciences Forum.
Its is regretted, although there is understanding for the (geographical/cultural) reasons why, that Arab countries are so far weakly represented.

The European Network on Epidemiology of rare disorders (NEPHIRD)

Ysbrand Poortman
Sweden

Epidemiological studies of rare disease are hampered by the rarity of the condition, by under or inaccurate and late diagnoses, by lack of money to establish adequate coordinating networks. In the past many epidemiological studies were small scale, rather local, divers in outcome and with limited reliability due to uncertainties in diagnosis, and difficulties in classification and therefore of doubtful scientific quality.

Epidemiological data on the incidence and prevalence of disorders are the basis for the estimation of the impact for society, policymaking and charting overall healthcare needs (e.g. hospital facilities, rehabilitation, adaptations, technical aids). Information of this nature is also important for scientific groups and for industry embarking on drug exploration and ultimately for pharmaceutical trials.

NEPHIRD (Network of Public Health Institutions on Rare Diseases) is the eponym for a major project funded by the European Union.

Countries involved are Belgium, Croatia, Denmark, France, Germany, Ireland, Lithuania, Luxembourg, Malta, the Netherlands, Norway, Portugal, Spain, United Kingdom, Sweden, Finland and Armenia.
The main objectives of this project are:

* to develop a model for epidemiological data collection at European level
* to identify the ongoing national activities on rare diseases in the participating
* to establish a European network for epidemiological and health care data collection on rare diseases

In the long term, a strong collaboration is expected between the various research centres, policy institutions and health service delivery centres dealing with rare diseases.

This concerted effort will create new ideas and knowledge and will facilitate the development of new drugs, treatment protocols, and diagnostic procedures. Recommendations on data base standards, institutional framework and health policy action on data collection and service delivery are the expected outcomes of the study.

Collaboration with similar projects or networks outside Europe is highly appreciated.

Current Approaches to the Treatment of Thalassaemia

Michel Angastiniotis
Cyprus

The classical treatment of Thalassaemia major has been that of regular blood transfusions and chelation. New treatment modalities are being developed aiming to improve effectiveness, meet the needs of adult patients, deal with complications, but also to if possible to replace transfusions.

In the field of iron chelation, the issues currently being discussed are the possible cardioprotective effect of Deferiprone, the advantages and indications of combination therapy with Desferrioxamine, the assessment of cardiac iron by magnetic resonance imaging and the expected new iron chelating agents which are expected to be marketed in the next two years.

A major area of research is the search of effective modulators of HbF production. After a period of disappointment with drugs that had some effect in Thalassaemia intermedia but not on transfusion dependent thalassaemia major, hope has been renewed by the discovery of more promising compounds. These are short chain fatty acids, methyl transferase inhibitors and histone deacetylase inhibitors which at the laboratory level at least are producing better results.

The role of anti-oxidants is still being explored and at present they seem to be a promising adjuvant to treatment due to their cellular protective effect.

Bone Marrow Transplantation is now accepted treatment for suitable candidates who have a related histo-compatable. The role of matched unrelated donors, cord blood stem cells, intrauterine transplants are al being explored.

In gene therapy, some breakthroughs have been reported but practical realization still seems very far away.

A multidisciplinary approach, especially of adult patients is now the accepted norm, so that the multiple organ pathology and the psychosocial issues can be effectively addressed.

Rantes Promotes Growth and Survival of Human First Trimester Forebrain Astrocytes

Moiz Bakhiet
Arabian Gulf University

The role of alpha and beta chemokines in the promotion of the ontogenetic development of the brain was examined. Rantes gene and protein was preferentially expressed in human foetal astrocytes in an age-dependent manner. Astrocytes from 5-week-old brains showed high proliferation and reduced survival, while 10-week-old astrocytes exhibited opposite effects. These effects were suppressed by anti-Rantes or anti-Rantes receptor antibodies and enhanced by recombinant Rantes. To persuade biological activity, Rantes induced tyrosine phosphorylation of several cellular proteins and nuclear translocation of STAT-1. IFN-gamma was required for Rantes effects since Rantes induced IFN-gamma and only 10-week old astrocytes expressed IFN-gammaR. Antibody-blocking of IFN-gamma reversed Rantes effects which suggests that cytokine/chemokine networks are critically involved in brain development.

Neonatal morbidity in infants of diabetic mothers in relation to maternal diabetic control and some selected social factors

Nevein H. Abou El Soud, Eitedal M. Dawoud, Nabila A. El Lithy, Ehsan B. El Sheikh
Basic Medical Sciences and Biochemistry Department, National Research Center, Cairo, Egypt

Objectives: To investigate the influence of maternal glycemic control and some selected social factors on neonatal morbidity in infants of diabetic mothers (IDMs).

Design: A randomized, group-comparative, cross-sectional study. Patients - 40 infants of diabetic mothers, 26 of mothers with class B through D and 14 with gestational diabetes. Setting - Abou El Rish Children Hospital. Intervention - Neonatal birth weight, serum glucose, bilirubin, calcium, hemoglobin, leucocytic count, hematocrite value were measured, blood gases when necessary. In addition to assessment of neonatal respiratory distress syndrome, heart failure, congenital malformations, traumatic injuries and neurological complications. Maternal blood glucose was estimated just after delivery, social data was collected (maternal and paternal age, level of education, smoking, parity and consanguinity). Main outcome measure: neonatal birth weight, serum glucose, bilirubin, calcium, and hemoglobin.

Results: The study population was divided into two groups according to maternal blood glucose values: uncontrolled group, 17Ιwith mean blood glucose > 110 mg/dl and controlled group, 23 with mean blood glucose < 110 mg/dl. The degree of maternal glycemic control appeared to affect perinatal outcome. Significant differences were observed for the incidence of respiratory distress syndrome (RDS) 70 %, macrosomia (4.01±1.03 vs 3.65± .63), septic infections (17.6 %) and traumatic birth injuries (17.6%) with P<0.05 in uncontrolled group. Maternal complications mainly hypertension and pre-eclampsia significantly increased as well by the maternal uncontrolled diabetes (P<0.05). There was positive correlation between increased parity and development of RDS (r =. 28,P=. 03), anemia (r=. 26,P=. 007) and macrosomia (r=. 41,P=. 004). Duration of diabetes and development of prematurity (r=. 31,P=. 02) and small for gestational age (r=. 2,P=. 03). Maternal glycemic control was related to the level of mother education (P=. 04) and father education (P=. 02). In addition to increased risk of neonatal polythycemia with maternal low education level and exposure to passive smoking, while, consanguinity of parents increased the risk of neonatal hypoglycemia.

Conclusion: These data suggest that maintaining maternal blood glucose level <110 mg/dl may serve to reduce several major forms of morbidity in the infant of diabetic mothers and that maternal low education level, older age and high parity together with, consanguinity and passive smoking represent other risk factors for neonatal morbidity in these infants.

Key Words: neonatal morbidity / infant of diabetic mother / maternal glycemic control.

Spectrum of Inborn Error of Metabolism in Saudi Arabia

Moeen Al-Sayed
Geneticist and Metabolist, Department of Medical Genetics, King Faisal Specialist Hospital And Research Center – Riyadh

Autosomal recessive genetic diseases are relatively common in Saudi Arabia. Contributing factors include large family size, extensive consanguinity and relative homogeneity of the Saudi gene pool. Among these disorders, inborn errors of metabolism (IEOM), which are by large autosomal recessive disorders, are particularly evident. Inborn errors of metabolism are usually the result of an enzyme defect or cofactor deficiency and should be seriously considered when any of the following symptom complexes are present in a patient's phenotype: catastrophic neonatal presentations, recurrent biochemical disturbances, liver disease or dysfunction, neurological symptoms or features; myopathy or cardiomyopathy, and signs of a storage disease.

More specifically, with advances in medical services in Saudi Arabia and especially at KFSH & RC in Riyadh, inborn errors of metabolism have moved from virtually unknown entities that were often mislabeled as sepsis and resulted in death, to well-known diseases that are considered in the differential diagnosis of any sick neonate.

This presentation will give an account of the various metabolic disorders in Saudi Arabia, their clinical presentation and methods of diagnosis incidence, prevalence and distribution of IOM in Saudi Arabia will be addressed. The presentation will also discuss the economic and social impact of IEOM and efforts towards primary and secondary prevention in Saudi Arabia.

Primary amenorrhea, infantile uterus, Alopecia, Diabetes mellitus, Intracranial calcification in two sisters: A new syndrome

Laila Ali Akbar Bastaki, Sadika Ali Al-Awadi Kamal Kamel Naguib
Kuwait Medical Genetics Centre, Maternity Hospital Al-Showaikh, Kuwait

Here in we are reporting a Kuwaiti female with masked face, coarse acromegalic facial features, tall stature, primary amenorrhoea, absence of secondary sexual characters, alopecia, scanty eye lashes, absent eye brows, diabetes mellitus and pyramidal and extra pyramidal disorders. She has similar mildly affected younger sister but free of the neurological symptoms, while her father has late onset Parkinson disease. On investigation, she had mild mitral regurge,visualized ovaries and small infantile uterus, CT head showed calcification of the basal ganglia, small hypodense lesion in the left half of the pituitary gland. Hormonal profile showed high LH & FSH, normal progesterone, normal E2, high para thyroid hormone, low deoxycorticosterone and normal adult growth hormone. Fasting blood sugar was high. Both visual and hearing assessments were normal while the IQ assessement was low in the proband and average / below average in the sister. Chromosomal study was normal 46,XX. These combinations have no match

Key words:
Alopecia, primary amenorrhoea, Hypogonadism, Diabetes mellitus, Consanguinity Mental, Retardation, Parkinson's disease

Genetic resistance against HIV infection and Aids

Alexander Voevodin

Abstract Not Available

Challenges of New Genomics: What is the Human Genome Project, and what challenges is it posing on health systems?

Naeema Aziz
Ministry of Health, Bahrain

The most revolutionary project of 21st century, the Human Genome Project (HGP), which began in 1990, is a government-funded project to map all of the human genes (30,000 total) on the 46 chromosomes and determine the sequences of the 3 billion chemical base pairs that make up human DNA. The project originally was planned to last 15 years, but effective resource and technological advances have accelerated the completion of the first working draft of Human Genome announced in June of 2000, with analysis published in February 2001.

The current debate is whether this genetic revolution is truly knocking our door and posing big challenges on the healthcare systems globally or its just hype. Advocates are promoting the possibilities and potential held by this powerful tool of science and medicine. Industry, which stands to make huge profits, is firmly behind the speedy transfer of discoveries from the laboratory to the clinical setting. One thing is certain: Genetic testing is upon us and its tide is growing stronger. Developing countries are lagging far behind in knowledge about the challenges of "New Genomics" an outcome of Human Genome project. What are the implication of HGP for our health system and how we should prepare for genetic testing tomorrow, will be discussed in this presentation.

Genetic Disorders among Arabs

Ahmad Said Teebi
Department of Medical Genetics, King Faisal Specialist Hospital and Research Center, Riyadh, SA

The Arabs are genetically diverse. The major factors that contributed to their diversity include the Semitic tribes migrations from the Arabian Peninsula to The Valley of Tigris & Euphrates in 3500 BC and to the East Mediterranean region in 2500 BC, the Islamic expansion in the 7th century AD, the Crusade wars and the recent migration dynamics. These events had resulted in the admixture of the original Arabs with other populations extending from east and south Asia to Europe and Africa. The demographic characteristics of Arabs nowadays include high rates of consanguinity; which is not a religious prescription but a tradition, large family size with an average of more than four children per family, and a rapid population growth. As a consequence of their inbreeding and due to other factors, there is a high frequency of autosomal recessive disorders together with increased frequencies of homozygosity of autosomal dominant traits such as familial hypercholesterolemia and X-linked traits such as G6PD deficiency.

Autosomal recessive entities that are prevalent among Arabs include: Hemoglobinopathies with variation in the incidence of specific types as seen in the frequencies of -thalassemia and sickle cell traits. Occurrence of rare traits (such as Hb-O) also exists in Arabs.

Neuromuscular disorders including Werdnig-Hoffman syndrome (all over), severe childhood autosomal recessive muscular dystrophy (Sudan/Tunisia), other neuromuscular disorders (all over), neonatal Schwartz-Jampel (UAE), and autosomal recessive ataxias (North Africa). Familial Mediterranean Fever (Jordan and Palestine) with an incidence similar to that in the Sephardic Jews and Armenians of 1:1500.

Metabolic disorders including homocystinuria and organic aciduria (Saudi Arabia), Wilson disease (all over), and congenital chloride diarrhea (Arabian Peninsula). Phenylketonuria have an incidence probably similar to those in Europe and North America but with different patterns of mutations that are also different within Arab countries, with many rare and unique mutations.

New syndromes and variants. More than 250 disorders have been described among Arabs, the majority of which are autosomal recessive. Among those, Sarjad-Sakati syndrome is relatively common in the Arabian Peninsula.

Cystic fibrosis originally thought to be extremely rare was often found in atypical presentations and mild forms. The incidence is close to that of Europe. The common European mutation (F508) has an incidence of 0-40% in various Arab communities. Many unusual or new mutations were detected.

Other relatively common autosomal recessive disorders include xeroderma pigmentosa (Egypt/ Tunisia), osteopetrosis syndromes including osteopetrosis with carbonic anhydrase II deficiency (Arabian Peninsula/Tunis) and nesidioblastosis of the pancreas (Arabian Peninsula). Common malformation syndromes include Bardet-Biedl, and Meckel syndromes

(Bedouins in Palestine and the Arabian Peninsula). There are several examples of disorders limited to certain isolates or large kindred's such as cystic fibrosis in a Qatari tribe, new type of Ehlers-Danlos syndrome with arterial tortuosity in Saudi Arabia and Qatar, neurogenic arthrogryposis congenita in a large Palestinian kindred, Bardet Biedl syndrome in some Bedouin tribes, cerebrotendinous xanthomatosis in the Druze community, and metachromatic and Krabbe's leucodystrophies in some Palestinian villages. Nonsyndromic sensori-neural deafness with Connexin 26 mutations is frequently diagnosed in Palestine, Arabian Peninsula and Tunisia. More work is required to further characterize the prevailing genetic disorders in each geographic location together with their mutations in order to plan for appropriate screening and testing.

Autosomal dominant disorders frequencies are probably similar to those seen elsewhere except for an increased frequencies of homozygosity for familial hypercholesterolemia in Lebanon and among the Bedouin in Kuwait.Among the X-linked disorders, G6PD deficiency has increased frequencies in maes and females. The development of Arab Genetic Disease Database (www.agddb.org) has been created and further development is in progress.

Bahrain Birth Defects Register – Prevention and control of congenital abnormalities

Shaikha Al Arrayed
Genetic department, Bahrain

Congenital anomalies have become a decisive part of child health care, and the right to be born healthy is an important criterion for better way of growing up. Birth defects are a structural defect that is present at birth whether diagnosed at that time or not. Most birth defects occur in ordinary families without known risk factors. The registry allows us to identify groups who may have a higher risk, and discover causes

It will answer question such as:
Are birth defects increasing locally? As the registry tracks rated and trends.
How will the Birth defect programs help?

Prenatal care: Study findings can be incorporated into clinical care, with appropriate interventions for recognized high-risk groups
Education: Woman can be warned of potentially hazardous exposures, and reassured about those found to be safe
Public policy: once identified, harmful exposures can be regulated or removed, reducing the threat to public health
Reducing health care costs: Preventing birth defects will save millions of Dinars in treatment cost each year.

The DBR should be suitable for the country; it has to be designed according to WHO requirements and to be comparable with International register (Clearing house of birth defect. Euro cat)

The Bahrain Birth defect register started in 1999, detailed features will be presented.

لمحات عن دور مجلس وزراء الصحة في مكافحة الأمراض الوراثية في دول مجلس التعاون لدول الخليج العربية

د. توفيق بن أحمد خوجة
المدير العام للمكتب التنفيذي لمجلس وزراء الصحة لدول مجلس التعاون لدول الخليج العربية

شهدت السنوات القريبة الماضية وبفضل الله عز وجل ثم التقدم العلمي والطبي وتحسين الوضع الاقتصادي والاجتماعي انخفاضا كبيرا في معدلات الإصابة والوفيات لكثير من الأمراض المعدية وسوء التغذية على المستوى العالمي والخليجي منه. غير أنه قد طفت على السطح مشكلات مهمة كالأمراض المزمنة, إضافة لما نحن بصدده الآن وهو مجموعة هامة من الأمراض تلعب الوراثة فيها دورا أساسيا أو كليا كالتشوهات الخلقية وأمراض الدم الوراثية والأمراض الاستقلابية وغيرها.. وصارت هذه المجموعة من الأمراض الوراثية أحد المشاكل المهمة التي تواجه كل طبيب في عمله.. لكن التقدم الهائل والسريع في مجال الوراثة البشرية عموما والجزئية خصوصا و الهندسة الوراثية والتوصل لخريطة الإنسان الجينية قد مكن من معرفة آلية ونوعية الوراثة لهذه الأمراض والتي تم تصنيفها ضمن مجموعات منها ما هو ناتج عن صفات سائدة أو متنحية أو محمولة عن طريق الجنس أو نقص الخماتر أو تداخل عوامل وراثية وبيئية , ومن هنا فإنه المواكبة الفعلية لهذا التقدم العلمي صار مهما لحل لغز آلاف الأمراض التي تصيب أو يمكن تعتري الانسان. كما صار التعرف على الأصحاء من حاملي الأمراض الوراثية أهم هذا ومجال للباحثين المختصين لتجنيب نقل المرض أو الإعاقة لذريتهم.

ومن هنا نشأت:

- فكرة إنشاء مراكز الاستشارة الوراثية للوقاية من انتشار هذه الأمراض وتجنيب الأبناء الإصابة بها.
- فكرة إنشاء عيادات الأمراض الوراثية للمعالجة المبكرة لتقليل وتخفيف الإعاقة والمعاناة.

والمكتب التنفيذي كان له قصب السبق في هذا المجال من خلال تأكيده:

- أولا على التعرف على حجم المشكلة خليجيا.
- وثانيا وضع البرامج المطلوبة لمكافحة.

وعلى هذا الأساس **تشكلت لجنة من دول المجلس** لاقتراح برنامج وقائي وعلاجي وتأهيلي لهذه الأمراض لتنفيذه مرحليا واقتراح خطة للبحوث والدراسات في هذا المجال. وكانت أولى اجتماعات هذه اللجنة (كويت ١٩٩٥م) حيث تم مناقشة أكثر الأمراض الوراثية انتشارا في دول المجلس (أمراض الدم الوراثية- أمراض التمثيل الغذائي – نقص إفراز الغدة الدرقية)..... **وكانت أهم توصيات الاجتماع:**

- تشكيل لجان وطنية على مستوى دول المجلس لمكافحة الأمراض الوراثية.
- إنشاء مركز الاستشارات الوراثية على مستوى دول المجلس.

وصدر قرار أصحاب المعالي وزراء الصحة بدول المجلس رقم (١) في مؤتمرهم التاسع والثلاثين (عام ١٩٩٥م في جنيف) لتفعيل توصيات الهيئة التنفيذية في المبادرة بتشكيل اللجان الوطنية وانشاء

مراكز الاستشارات الوراثية وتطوير أساليب التعريف والتوعية بهذه الأمراض ... كما صدر قرار أصحاب المعالي الوزراء رقم (١٤) في الاجتماع الثاني والأربعين (عام ١٩٩٧م في أبو ظبي) بتفعيل أعمال اللجنة الخليجية المختصة وإعداد برنامج للوقاية من هذه الأمراض وتأهيل للخدمات المتوفرة في كل دولة والتنسيق والتعاون فيما بينها في هذا المجال... حيث تابعت اللجنة في اجتماعها الثاني (مسقط ١٩٩٧م)ما تم تنفيذه وشددت على الانتهاء من إنشاء اللجان الوطنية ومراكز الاستشارات الوراثية ودعم المسوح الصحية في هذا المجال وتعميم ودعم عيادات الأمراض الوراثية وتجهيز مختبراتها بما يحقق الأهداف المرجوة منها وتطوير برامج توعية صحية للتعريف بهذه الأمراض.

وفي اجتماعها الثالث (الكويت ١٩٩٨م) أكدت اللجنة على أن تكون اللجان الوطنية للأمراض الوراثية في وزارات الصحة هي المسئولة عن كل الأنشطة المتعلقة بالبرنامج.

وفي توصيتها رقم (٣) في اجتماعها التاسع والأربعين (١٩٩٨م) تأكدت الهيئة التنفيذية لدول المجلس بإنشاء السجلات الوطنية للأمراض الوراثية. كما دعت اللجنة في اجتماعها الرابع (الكويت ٢٠٠٠م) لمواصلة استكمال إنجاز التوصيات على المستويات الوطنية، ومواصلة دعم الدول لها لتحقيق ذلك. وأوت الهيئة التنفيذية في اجتماعها الثامن والخمسين (٢٠٠٣م) دول المجلس بتجميع وجدولة وتصنيف البيانات التي تم تسجيلها عن الأمراض الوراثية والتشوهات الخلقية وذلك لتقدير حجم المشكلة بدول المجلس واقتراح خطة لمكافحة المناسبة. كما أكد الاجتماع المذكور على أهمية توفير التجهيزات المختبرية الملحقة بعيادات ومراكز الأمراض الوراثية وتوفير القوى العاملة الفنية المؤهلة والمدربة بتشغيلها. حيث بادرت معظم دول

المجلس بتنفيذ هذه التوصيات أو معظمها ومواصلة دعم الجهود التي تبذلها اللجنة الخليجية المختصة في هذا المجال.
كما صدرت قرار أصحاب المعالي الوزراء رقم (١٥) في الاجتماع الرابع والخمسون (عام ٢٠٠٣م في أبو ظبي) والذي بحث الدول الأعضاء على الاهتمام بالاشتراك في الاستراتيجية المقترحة لمكافحته.

وتأكيدا على الأهمية التي يوليها دول المجلس لهذا الموضوع كانت التوصية بعقد مؤتمرا خليجي للأمراض الوراثية والذي قامت مملكة البحرين بجهود مشكورة بالإعداد المناسب لعقد هذا المؤتمر واستضافته هذا العام فيها.

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