

Genetic identity

by Joanna Hartley on Wednesday, 07 January 2009

The unraveling of the human genome has revealed a unique and complex genetic epidemiology among Arab populations that will impact heavily on future healthcare policy in the region.

Scientific research into the human genome during the past 30 years has led medicine to a unique place in history, where for the first time the healthcare needs of individuals can be predicted before there are any signs of disease.

Genetic medicine is still seen as a frontier speciality, at least one step removed from day to day care, but its impact, especially among local populations in the Middle East, is set to have far reaching consequences, according to genetics experts.

Work carried out by the Centre for Arabic Genomic Studies (CAGS), based in Dubai, has discovered a complex and unique genetic epidemiology within the Arab world - influenced largely by the cultural preference for consanguinity that accounts for between 30% and 60% of marriages, depending on the country.

This trend for marrying first or second cousins, plus older parenthood and poor detection rates for hereditary conditions, has led to a high incidence of chromosomal disorders and congenital anomalies. These affect 8% of Arab populations compared with 3% of population's worldwide.

As a result genetic conditions account for more than 40% of all infant deaths in countries such as the United Arab Emirates (UAE), and cost Arab healthcare systems in terms of direct and indirect treatment costs around US\$ 13 billion per year.

It is a situation governments in the region have been making moves to tackle, but with limited evidence of success.

In the past eight years the UAE, Saudi Arabia and Qatar have all introduced pre-marital screening schemes for sickle cell disease and thalassaemia, both of which are common in the region. Thalassaemia affects around 8% of local populations and sickle cell anaemia is seen in around 1.5%.

However, Middle East genetics experts are warning that with the popularity of consanguinity growing stronger, the region should brace itself for even higher rates of genetic disorders in the future.

"Consanguinity is increasing," reveals Dr Ghazi Omar Tadmouri, assistant director of CAGS. "Despite education it has not stopped, it is a preferred choice, and so we expect genetic disorders will increase even more over time."

The cost of consanguinity

In view of this it is ever more important for healthcare professionals in the region to be fully aware of the unique and complex genetic epidemiology of Arab populations, Dr Tadmouri adds.

For a start marriage between relatives has led to a higher incidence of auto-recessive disorders in Arab countries than other parts of the world, where dominant disorders are more common.

Common auto-recessive conditions include cystic fibrosis, spinal muscular atrophy, congenital adrenal hyperplasia and phenylketonuria, which are all chronically debilitating and have the potential to lead to premature death.

Dr IC Verma, head of the department of genetic medicine at Sir Ganga Ram Hospital, New Delhi, India, which carries out genetic testing on Middle Eastern couples, says the situation is leading to increased need for families to get a handle on their genetic histories.

Although testing individual's for their own carrier status is common place, it is also advisable to obtain a thorough family history, so whole families can be preapred for the possibility of a child being born with a genetic disorder, Dr Verma says.

However, this is easier said than done, he admits. because the desire to be seen as suitable marriage material often prevents couples from revealing family members with a genetic disorder. "Genetic



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discrimination is a problem. People do not want to tell others I have this genetic disease in my family," Dr Verma says.

Yet, the link between consanguinity and auto-recessive disorders is unquestionable. It can be clearly seen in CAGS data, which is based on 2,000 pieces of genetic research carried out on the populations of the UAE, Oman and Bahrain. The data has been collated by CAGS in the past five years.

Figures presented by Dr Tadmouri at a conference on advanced genetic testing in Dubai two months ago, show that in Bahrain, where consanguinity is 39%, the prevalence of auto-recessive disorders is 45% of all genetic conditions.

Compare this with Oman, where the rate of family marriage is higher - hitting 45% - and auto-recessive disorders account for 55% of conditions. And the UAE, which has the highest consanguinity level of 47%, and an auto-recessive disorder rate of 58% of all genetic conditions.

The data also shows that a high proportion of children with genetic disorders have more than one condition. For example 47% of those diagnosed with sickle cell disease also have glucose-6-phosphate-dehydrogenase deficiency (G6PD), while 98% of those with cystic fibrosis have G6PD too.

Having two genetic conditions adds to the severity and complexity of symptoms and the treatments required, Dr Tadmouri explains. "Just cystic fibrosis with all the related problems is bad enough, then that person has anaemia," he muses.

"The average rate of death for a cystic fibrosis patient in Bahrain is 60%, higher than anywhere else because it is associated with another disorder." Dr Tadmouri adds.

Arab phenomenoms

On top of this there is evidence that the genetic mutations which lead to the same diseases are not in fact the same across the region. This means that the same presenting disorder, such as thalassaemia or diabetes, may actually be the result of a different genetic sub-mutation.

"There is diversity at a molecular level. With thalassaemia there are 250 mutations worldwide. In the UAE there are 50 mutations which pose different problems, some are very mild and some very severe," Dr Tadmouri reveals.

This can create problems in diagnosis, he adds, as there are no testing kits at present able to pick up all the subgroup mutations for the same disease, which in practice may actually require a different approach to treatment and care.

Furthermore, despite the common cultural practice of family marriage, and the close geographical proximity of countries in the region, genetic differences between Arab populations are surprisingly varied.

CAGS data shows that across all Arab populations there are 898 genetically linked disorders, some of which have never been identified outside of the region.

Within this there are 451 genetic disorders that are only seen in the UAE, Oman and Bahrain, and within this there are high numbers of disorders that are unique to each country - there are just 30 common to all three.

"A small fraction of these disorders are common in the three populations, while a large number of genetic disorders are country specific indicating a remarkable heterogeneity in the populations in the region," Dr Tadmouri explains.

Of the 451 about 119 are "deadly", he adds. "By the time these diseases are reported, those patients have died in the first hours, or days, or weeks, or in their early years."

Brave new world

All these discoveries will have a significant impact on the way both governments and healthcare professionals will have to approach screening and treatment regimens in the future.

Preventing genetic disease is obviously a key objective. Biochemical pre-marital tests are in place in most Middle Eastern countries, but in most they are limited to testing for sickle cell anaemia and thalassaemia.

However, newer chromosomal tests can pick up at least nine disorders, says Dr Sanjida Ahmed, director and researcher at Eastern Biotech and Life Sciences, Dubai.

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The current programmes need to be extended to ensure as many disorders as possible can be detected, she believes.



"In the end, the Ministry of Health says they are at the initial stages of this (pre-marital screening). They are conveying the message to people first, and will wait until people are familiar with this procedure before they will do it more in the future," Dr Ahmed tells *Medical Times*.

Yet, there are doubts among screening experts that pre-marital testing in Arab countries will ever have a significant impact on cutting incidence rates of genetic disorders, unless the option to terminate affected foetuses is also available.

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Research published in the British Medical Journal (BMJ) in October 2005 shows that a pre-marital screening scheme for thalassaemia launched in Iran in 1996 had no impact on cutting incidence rates in newborns.

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That was until it was made legal, in 2001, for affected foetuses to be terminated up to 15 weeks, which resulted in a 70% drop the following year in new cases.

"Experience shows that if (post-pregnancy) options are not made available to carrier couples, such programmes will not be effective in reducing the burden of genetic diseases, as many carrier couples go ahead with their planned marriage," writes lead author Professor Lihadh Al-Gazali, from the clinical genetics department of paediatrics in the faculty of medicine and health sciences at UAE University.

But termination, which is illegal in most Arab countries unless the mother's physical health is at risk, is unlikely to be made lawful due to its uneasy footing within Islamic principals, Professor Al-Gazali adds.

So, geneticists are looking at other options. Screening whole families is a less threatening and more cost effective option, according to Dr Verma. "Where there is a child with thalassaemia, then screen the extended family, and that is a cost effective way of doing it," he says.

The approach is particularly suitable for populations with high levels of consanguinity and clustering of rare genetic diseases, advises Professor Al-Gazali.

Screening of newborns is another important arm of healthcare management for genetic disease, which has been implemented for a limited number of conditions in the UAE, Saudi Arabia, Oman and Jordan - including phenylketonuria, congenital hypothyroidism, G6PD, cystic fibrosis and sickle cell anaemia.

Data from Saudi Arabia suggests that 50% of genetic disorders picked up at birth are manageable. Treatment can prevent death and substantially improve quality life, says Dr Ahmed, whose company carries out newborn screening for a number of local maternity units. Genetic technologies can now pick up more than 50 hereditary disorders known as 'errors of metabolism', at birth, she adds.

"Early detection is important as these (disorders) can be controlled and completely cured, such as lactose intolerance, all they need is to have lactose free milk," Dr Ahmed says.

Pre-implantation genetic diagnosis, however, is proving one of the most popular approaches to genetic detection in Arab countries, according to a study carried out by Professor Al-Gazali of UAE residents.

Yet, she warns that the procedure, which involves assisted reproductive technology, is still in its early stages of development and has many limitations.

"It is complex, time consuming and expensive, and only available at a small number of centres and for a limited number of genetic conditions," she adds.

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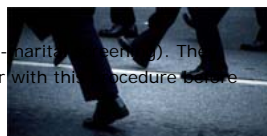
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