LETTERS TO THE EDITOR

Multiple pterygium syndrome: a relatively common disorder among Arabs

Multiple pterygium syndrome (MPS), also referred to as Escobar syndrome or pterygoarthromyodysplasia syndrome, is a rare, autosomal recessive disorder characterised by multiple congenital joint contractures, multiple skin webs, camptodactyly with or without syndactyly, distinct facial appearance with ptosis and antimongoloid eye slant, short stature, kyphoscoliosis, and vertebral segmentation anomalies. Approximately 60 cases have been reported from several countries in English language publications.

In Kuwait, during a community genetic survey at Farwania district hospital, serving a mixed Arab population of 400,000, we have ascertained 13 cases of MPS in six sibships in four Arab families. There were five males and eight females. Their ages ranged from soon after birth to 19 years.

Family 1. The parents are normal, first cousin Kuwaiti Bedouin with five affected children (two females and three males), who have been reported elsewhere, and six normal children (two males and four females). The oldest normal daughter has recently married a non-consanguineous Bedouin and has had an affected girl with multiple contractures and pterygia noted at birth. The manifestations in this family are typical of autosomal recessive MPS with marked intrafamilial variability. In addition, two of the affected children have had frequent admissions to paediatric wards for respiratory problems.

Family 2. The parents are normal, Palestinian first cousins who had two affected children, a 19 year old male and a 10 year old female, and five normal children (two males and three females). A male child died in the neonatal period because of congenital heart disease (not examined). The manifestations are typical of MPS but without ptosis.

Family 3. Two normal sisters were married to their normal first cousins who were brothers. One of the couples had three affected children. The oldest is 16 years old; she has severe respiratory impairment and recently developed pulmonary hypertension and heart failure. There were five normal children with families (four males and one female). The other couple had an affected boy and another, recently born, affected girl (not examined by us) in addition to two normal girls. Apart from the oldest girl, the children in this family had plastic surgery for their joints and webs in early life. This family is Palestinian of black African ancestors.

Family 4. The parents are normal, first cousin Kuwaitis whose first child (female) had congenital joint contractures, pterygia, and the typical facial appearance, as noted at the age of 7 months.

Our cases of MPS, briefly reported here, represent the largest series reported so far from one centre. It is noteworthy that they are not the only cases detected in Kuwait (population 2 million) where there is a well established community genetic service in three districts. The estimated minimum prevalence in the general population of Farwania district is approximately 1 in 31 000 (13 400 00) and, if the specific age group is considered, the prevalence would be much higher. This prevalence is high for a monogenic malformation syndrome and is similar to that of Bardet-Biedl syndrome in the Arabs of Kuwait.6 The finding of Thompson et al of a high proportion of Asian and Middle Eastern cases (including a case from Jordan) among 11 cases studied in UK is highly significant and is not a chance occurrence. Data from other centres in the Middle East may show that MPS is relatively common among Arabs in particular or even among other communities in the Middle East.

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Features of Turner's and DiGeorge's syndromes with X;22 translocation

We read with interest the paper entitled 'Features of Turner's and DiGeorge's syndromes in a child with an X;22 translocation' by Pinto et al (J Med Genet 1989;26:778–80) and would like to comment on it.

We agree that in this case the DiGeorge's syndrome (DGS) is the result of a 22q11 deletion. However, the hypothesis that a paternal meiotic accident plus adrenal hypoplasia (AHC) in one of the mother's X chromosomes was a coincidence is not convincing. The AHC gene is rare and no other case is mentioned in this family. Furthermore, as the authors quoted, "it is tempting to assume that the breakpoint in this t(X;22) is located at the region to which the AHC gene was assigned". This does not imply that the patient's mother is a carrier of the AHC gene. The authors concluded that only the abnormal X was inactivated.

If the replication study was mainly carried out on peripheral blood cells, available surviving lymphocyte cell lines necessarily come from clones with the abnormal X inactivated. This selective effect has been seen in females with X linked immunodeficiency diseases. In the other tissues, inactivation of the normal X, which usually occurs in the unbalanced t(X;A), would be sufficient to explain the association of DGS and AHC in this child.

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Marfan syndrome

Dr de Groote et al (J Med Genet 1990;27:82–5) present linkage data for Marfan syndrome using markers on
Features of Turner's and DiGeorge's syndromes with X;22 translocation.
S Gilgenkrantz and M Teboul

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Updated information and services can be found at:
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