LETTERS TO THE EDITOR

Multiple pterygium syndrome: a relatively common disorder among Arabs

Multiple pterygium syndrome (MPS), also referred to as Escobar syndrome¹ or pterygioarthromyodysplasia syndrome,² 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 anti-mongoloid eye slant, short stature, kyphoscoliosis, and vertebral segmentation anomalies. Approximately 60 cases have been reported from several countries in English language publications.³⁻⁵

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,⁵ 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 daughters. 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*⁴ 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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- 2 Temtamy S, McKusick VA. The genetics of hand malformations. *Birth Defects* 1978;3:155-6.
- 3 Hall JG, Reed SD, Rosenbaum KN, Gershanik J, Chen H, Wilson KM. Limb pterygium syndromes. A review and report of eleven patients. *Am J Med Genet* 1982;12:377-409.

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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 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

chromosomes 1 and 11 in six kindreds. We are concerned about the possibility of over diagnosis of Marfan syndrome within the kindreds presented. In the six families, one would expect 37 out of the 74 offspring who are at a 1 in 2 risk of inheriting the gene to be affected. Fifty-seven were in fact diagnosed as having Marfan syndrome; this is significant ($\chi^2=5.3$, $p<0.05$). The disparity is even more marked when families are sampled selectively. In family 4 a lod score of 0.92 was obtained using one of the probes, D1S7. However, out of 13 subjects at a 1 in 2 risk, only two were diagnosed unaffected.

Such methodological problems, we believe, may lead to false conclusions regarding exclusion of loci which may possibly be involved in Marfan syndrome.

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BOOK REVIEW

Third International Conference on Osteogenesis Imperfecta. Ed G Cetta, F Ramirez, P Tsipouras. (Pp 187.) New York: The New York Academy of Sciences. 1988.

This volume contains papers presented at the Third International Conference

on Osteogenesis Imperfecta (Pavia, Italy, 1987). Contributions on clinical, genetic, biochemical, and molecular aspects are presented, and aim to give a broad overview of current knowledge about this heterogeneous and complex group of connective tissue disorders.

Part I on nosology and genetics contains four papers discussing clinical classification and correlation of phenotype with biochemical and molecular pathology. The first paper by Silience outlines his generally used clinical classification and contains useful radiographs. This is followed by a different and somewhat conflicting classification by Maroteaux *et al* based on antenatal or postnatal presentation, while Beighton *et al* report linkage data on families categorised according to the Silience classification. It is clear from these papers that the biochemical and molecular heterogeneity within each defined clinical phenotype is considerable and not yet well understood.

Part II contains eight papers on biochemical defects detected in type I collagen, ranging from reports of abnormalities documented in single families to studies of substantial numbers of patients with all clinical types of osteogenesis imperfecta. No summary of collagen biochemistry is given, and a basic knowledge in this area is required in order to interpret the findings presented.

Part III contains three very good papers on the structure of the type I collagen genes, including an excellent review by Byers *et al* on the molecular basis of clinical heterogeneity. As in the biochemical papers, it is assumed that the reader will be familiar with molecular terminology.

Part IV on genetic counselling incorporates only two papers. The first by Sykes and Ogilvie concentrates on

the potential application to prenatal diagnosis of linkage with either COL1A1 or COL1A2 loci in dominantly inherited OI. The paper by Thompson *et al* gives excellent practical guidelines for counselling in cases of perinatally lethal and progressively deforming osteogenesis imperfecta.

Part V on management contains four papers, three of which report experience with orthopaedic treatment from units with a special interest in osteogenesis imperfecta.

As is inevitable in a book of this format, the style and quality of the papers varies considerably, and there is a certain amount of repetition between various chapters. The fact that English is not the first language of some authors detracts from a few of the chapters. It is difficult to define the readership for this book. Scientific investigators working in this field will inevitably find certain areas already out of date, and clinicians involved with osteogenesis imperfecta will find limited new information that is not already available to them. The lack of general introductory chapters on collagen biochemistry and molecular studies detracts from the value of the book for readers who are not already conversant with these aspects. Paediatricians who are occasionally faced with the task of clarifying the diagnosis and prognosis in affected neonates would find the clinical classification useful, despite the limitations imposed by the unresolved problem of heterogeneity in this group of disorders. Despite these reservations, this book would be a useful addition to postgraduate libraries, even if few clinicians or scientists wish to purchase personal copies.

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