Genetic Disorders in the Arab World: United Arab Emirates

Malformation Syndromes and Osteochondrodysplasias in the United Arab Emirates

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Several malformation syndromes are prevalent in the UAE. These syndromes tend to cluster in certain tribes and families where the incidence can be very high. One of the common syndromes seen in this country is Joubert syndrome. This autosomal recessive syndrome is characterized by agenesis of the cerebellar vermis, ataxia, hypotonia, oculomotor apraxia, neonatal breathing problems and mental retardation (Szttriha et al., 1999). Joubert syndrome has a birth prevalence of 1 in 5000 in the UAE. Over a period of 10 years, 30 children from 13 families with Joubert syndrome were seen. Most of these families originated from Oman. Homozygosity mapping was used to map two genes in these families to chromosomes 9q34.3 and 11p12-q13.3 (Saar et al., 1999, Keeler et al., 2003). A third gene was also mapped in a Palestinian family living in the UAE (6q23) and a mutation in the AHI1 gene, which encodes Jouberin protein, was found in that family (Dixon-Salazar et al., 2004).

Another commonly seen syndrome is Meckel syndrome. This autosomal recessive syndrome is characterized by occipital encephalocele, polydactyly and cystic dysplastic or polycystic kidneys. Meckel syndrome has a birth prevalence of 1 in 5000 in the UAE (Al-Gazali et al., 1999a) and is known to be common in Bedouins in Kuwait and Palestinian Arabs.

Another frequently seen syndrome in the UAE is Ehlers-Danlos Syndrome type VI (EDSVI). This autosomal recessive syndrome is caused by deficiency of lysyl hydroxylase enzyme and is characterized by progressive kyphoscoliosis, joint laxity and hypotonia in infancy and ocular fragility in late childhood. EDSVI was seen in 11 children from 7 consanguineous families, most of these families are Bedouins of UAE origin. A founder R313X mutation in the PLOD1 gene was found in these families (Giunta et al., 2004).

Other frequently seen syndromes in the UAE include Bardet-Biedel, Seckel syndrome, McKusick-Kaufmann syndrome, Cohen syndrome, Geroderma-Osteodysplastica, and Setleis syndrome (Al-Gazali et al., 2001a; Slavotinek et al., 2002; Chandler et al., 2003). In addition, several new syndromes have been identified and reported from the UAE. In some of these syndromes the gene has been localized and work is in progress to identify the mutations involved (Al-Gazali 1997; Al-Gazali et al., 1999a; Al-Gazali et al., 1999b; Al-Gazali et al., 1999c; Al-Gazali et al., 2000; Al-Gazali et al., 2002a; Al-Gazali et al., 2002b).

Osteochondrodysplasias

Osteochondrodysplasias are relatively common in the UAE. In a study of 38,084 births in Al-Ain Medical District, 36 cases of skeletal dysplasias were found; thus, a rate of 9.46/10,000 births (Al-Gazali et al., 2003a). Fibrochondrogenesis was found to be common in this population in particular in families of Omani origin. Another relatively common bone dysplasia in the UAE is Stuve-Wiedemann Syndrome (SWS) which has a birth prevalence of 0.52/10,000 births (Al-Gazali et al., 2003b). Over a period of 10 years, 24 children from 13 families were seen. Most of these families originated from Oman and Yemen. A founder mutation in the Leukemia Inhibitory Factor (653_654 ins T) was identified in these families (Dagoneau et al., 2004). Another fairly common bone dysplasia is Osteodysplasic Primordial Dwarfism type II which is common in families originating from Oman and Pakistan.

Other types of bone dysplasias seen in the UAE include: Ellis-Van-Creveld syndrome, chondrodysplasia punctata, short rib polydactyly syndromes, Jeune thoracic dystrophy, Dygve-Melchior-Clausen syndrome, and Larsen and Debuquois syndromes (Faivre et al., 2003; El Ghouzzi et al., 2003). In addition, several rare and new types of bone dysplasias have been identified and reported from the population of the UAE (Al-Gazali et al., 1996a; Al-Gazali et al., 1996b; Al-Gazali et al., 2001b; Al-Gazali et al., 2003a; Al-Gazali et al., 2003b; Bayoumi et al., 2001).