Geroderma Osteodysplastica

Alternative Names
GO
Gerodermia Osteodysplastica
Osteoplastic Gerodermia Walt Disney Dwarfism
Premature Senility Syndrome
Bamatter's syndrome
Bamatter-Franceschetti-Klein-Sierro Syndrome
Walt Disney Dwarfism

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
231070

Mode of Inheritance
Autosomal recessive

Description
Gerodermia osteodysplastica is a rare, autosomal recessive developmental disturbance of connective tissue characterized by changes in the skin suggesting precocious aging, wrinkled, and lax skin with reduced elasticity which is more marked on the dorsum of the hands and feet. The prematurely aged appearance has been likened to the dwarfs in Walt Disney's 'Snow White.' The bones are osteoporotic and susceptible to fractures, particularly the vertebrae, which show compression with anterior wedging and biconcavity.

Molecular Genetics
Scientists suggest the collagen genes COL3A1 and COL5A2 could be candidate genes for gerodermia osteodysplastica and wrinkled skin syndrome. Mutation in COL3A1 causes Ehlers-Danlos IV (EDS IV), while mutation in COL5A2 gene produce EDS type I and II phenotype.

Epidemiology in the Arab World

Kuwait
Al-Torki et al. (1997) reported the first Arab family with gerodermia osteodysplastica in which there were two affected female siblings. The parents were second cousin healthy Kuwaitis with Bedouin ancestors. A deceased brother of their paternal grandfather has been reported by the family to have a similar phenotype. The affected siblings had a prematurely aged face with loose and wrinkled skin, joint laxity/dislocation, and osteoporosis. In addition, the patients had ear anomalies and an abnormal EEG, not previously reported in association with gerodermia osteodysplastica. Al-Torki et al. (1997) noticed a more pronounced phenotype in the younger sib with the presence of delayed motor milestones, hypotonia, brachycephaly and kyphoscoliosis, and abnormal EEG records. On the other hand, the elder girl had pes palmaus.

Morocco
Lustmann et al. (1993) presented four individuals with Gerodermia Osteodysplastica in a Jewish family from Morocco confirming the autosomal recessive inheritance of the disorder. Lustmann et al. (1993) discussed in detail abnormalities of the mandible and mandibular teeth associated with Gerodermia Osteodysplastica. Lustmann et al. (1993) also described three previously unreported findings: a) enlarged funnel-shaped mandibular lingula; b) extension of the mandibular premolar and molar roots below the inferior dental canal, and of the second molars into the lower border of mandibular cortical bone; and c) hypercementosis of the maxillary incisors and mandibular molars surrounded by a radiolucent halo in several teeth.

Palestine
Al-Gazali et al. (2001) reported three patients for a Palestinian consanguineous couple,
residents of the United Arab Emirates, who also have one normal child. The parents were symptomatically normal and had a history of miscarriages at five months and two months. All children had similar dysmorphic facial features consisting of broad and prominent forehead, hypotelorism with epicanthal folds, prominent bulbous nose, flat malar region, and large protruding ears. All had wrinkling of the skin more marked on the dorsum of the hands, feet, and abdomen; hyperextensibility of the joints, particularly of the hands; and aged appearance. Intrauterine growth retardation, subsequent failure to thrive, developmental delay, and variable degree of osteoporosis was also present in all of them. The older children developed progressive prognathism. In view of the similarities and overlap between geroderma osteodysplastica, wrinkly skin syndrome, and cutis laxa and developmental delay, Al-Gazali et al. (2001) suggested that all these syndromes represent variable manifestation of the same disorder.

Syria
Al-Gazali et al. (2001) reported two children for a Syrian consanguineous couple, residents of the United Arab Emirates, with no family history of any significant problems. Both children had similar dysmorphic facial features consisting of broad and prominent forehead, hypotelorism with epicanthal folds, prominent bulbous nose, flat malar region, and large protruding ears. They had wrinkling of the skin more marked on the dorsum of the hands, feet, and abdomen; hyperextensibility of the joints, particularly of the hands; and aged appearance. Intrauterine growth retardation, subsequent failure to thrive, developmental delay, and variable degree of osteoporosis was also present in both of them. In view of the similarities and overlap between geroderma osteodysplastica, wrinkly skin syndrome, and cutis laxa and developmental delay, Al-Gazali et al. (2001) suggested that all these syndromes represent variable manifestation of the same disorder.

United Arab Emirates
[See also: Palestine > Al-Gazali et al., 2001; Syria > Al-Gazali et al., 2001].

References

Contributors
Ghazi O. Tadmouri: 16.4.2006
Ghazi O. Tadmouri: 2.5.2005
Ghazi O. Tadmouri: 16.4.2005