Odontoonychodermal Dysplasia

**Alternative Names**
Fadhil Syndrome

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

**OMIM Number**
257980

**Mode of Inheritance**
Autosomal recessive

**Description**
Odontoonychodermal dysplasia is a rare disorder characterized by anomalies of the structures arising from the ectoderm. These anomalies include hyperhidrosis of the skin, hyperkeratosis palmaris, nail dystrophy, dry and sparse hair, facial erythema, peg-shaped incisors, and malformation of other teeth. Mild mental retardation was observed in some cases.

**Molecular Genetics**
Odontoonychodermal dysplasia is inherited in an autosomal recessive pattern and some cases are familial with parental consanguinity. There is no identification for the associated gene to date.

**Epidemiology in the Arab World**

**Lebanon**
Fadhil et al. (1983) reported seven affected individuals, in a total of 24 belonging to three inbred Lebanese sibships, with apparently an undescribed pure ectodermal dysplasia. For this condition, probably owing to the homozygous state of an autosomal recessive gene, Fadhil et al. (1983) suggested the name trichoolodontodermal dysplasia.

Megarbane et al. (2004) reported three Lebanese boys who were thought to have odonto-onycho-dermal dysplasia syndrome. The patients were two brothers and their maternal cousin. The parents and the other sibs in both families were normal. The first patient had at birth absent scalp hair and his fingernails were barely visible. Later on, he had widely spaced deciduous teeth. Hyperkeratosis of palms and soles appeared at around age four years. He was 17 years old when examined with noticeable dry hair and skin. Palmoplanter erythema and dystrophic nails were observed. The patient also had recurrent cutaneous dermatophytosis. His tongue had no papilla and there was light root resorption on molars. The brother of the first patient was 16 years old. At birth, he had normal hair and nails. He displayed the same physical features as his affected brother, but the palmoplanter hyperkeratosis was more severe, fingernail dystrophy less severe, and toenails normal, except for the fifth toenail. All mandibular teeth were deciduous, very compromised, and had different root resorption degrees. Also, there were four abnormally shaped permanent teeth germs. The third patient was the maternal cousin of the two brothers. At birth, he had almost no scalp hair or nails. His deciduous teeth were widely spaced. On examination, he was 18 years and presented the same clinical features of his cousins, but the whole presentation was more severe. He had three persistent maxillary deciduous teeth and two upper incisors were originally conical. A biopsy from his planter skin was examined histologically showing orthokeratotic hyperkeratosis, hypergranulosis, and mild acanthosis in the epidermis. A perivascular infiltrate was visible in the stratum papillare of the dermis. Glandular cells were few. Scanning electron microscopy examination of the third patient’s hair demonstrated some irregular hair with longitudinal depressions. Linkage analysis to X chromosome was performed for different members of both families, however, linkage analysis and lod-score calculations showed no
evidence for X-linkage. The three studied patients had neither telangiectatic nor sparse hair, eyelashes, and eyebrows. Those features were observed in previously described cases with odonto-onycho-dermal dysplasia syndrome. Therefore, Megarbane et al. (2004) suggested that those three patients either had odonto-onycho-dermal dysplasia syndrome with clinical differences, or a new but similar entity to the odonto-onycho-dermal dysplasia syndrome.

References

Contributors
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