Focal Dermal Hypoplasia

**Alternative Names**
FDH
FODH
DHOF
Goltz Syndrome
Goltz-Gorlin Syndrome

**Record Category**
Disease phenotype

**WHO-ICD**
Congenital malformations, deformations and chromosomal abnormalitie > Other congenital malformations

**Incidence per 100,000 Live Births**
2-5

**OMIM Number**
305600

**Mode of Inheritance**
X-linked dominant

**Gene Map Locus**
Xp11.23

**Description**
Focal Dermal Hypoplasia (FDH) is a congenital disorder characterized by defective development of ectodermal and mesodermal tissues, in association with defects in various organs. Typical dermatological features of the condition include localized patches of hypoplasia, which also appear to be depigmented, inflammation, reddening, blistering and irritation in affected areas, presence of papillomas in sensitive areas like gums, tongue, armpits, genital organs, and anus, hyperkeratosis in the palms and soles resulting in excessive sweating, and patchy hair loss. In addition, patients may have dental problems (malformed teeth, and cavities), webbed digits, syndactyly, spinal curvature, protruding lower jaw, hearing loss, facial deformities, and ocular abnormalities (anophthalmia, coloboma, and strabismus). The disorder can also affect the stomach, intestines, heart, lungs, and kidneys. About 15% of affected patients also show mental retardation. With rare exceptions, FDH is seen only in females. This is because the condition is lethal in males, and affected males die in utero.

Diagnosis is based on the clinical symptoms of the condition. The lack of pigmentation and abnormal sweating seen in FDH are useful indicators in differentiating it from other ectodermal dysplasias. Affected female patients can be properly managed with the help of dermatological treatments, dental work, orthopedic surgery, and if needed, respiratory therapies. With treatment, a normal life expectancy is possible. However, as mentioned before, the disease is lethal in males and most affected male fetuses either do not reach term or are stillborn.

**Molecular Genetics**
FDH is an X-linked dominant disorder, which explains the lethality observed in males. The locus responsible for this condition was identified as the Xp11.23 locus, which contains the gene for Microphthalmia with Linear Skin Defects as well as Aicardi Syndrome. The exact gene responsible for causing FDH, however, is the PORCN gene, the protein product of which modulates the processing of Wnt proteins. It is not clear exactly how mutations in the PORCN gene affect dermal tissue development, although it is known that improper secretion of the Wnt proteins interferes with normal tissue development.

**Epidemiology in the Arab World**

**Jordan**
Al-Anazy and Zakzouk (1997) reported the case of a 46-year-old Jordanian male who complained of progressive left nasal obstruction and hearing loss for five years with a history of multiple surgical removal
of dental cysts. Clinical and radiological investigation revealed a cystic mass occupying the left maxillary sinus protruding to the nasal cavity, calcification of the falx cerebri and bifid ribs. On these findings Gorlin-Goltz syndrome was confirmed.

**Oman**
Sawardekar (2005) conducted a study to establish the prevalence of major congenital malformations in children born during a 10-year period in Nizwa Hospital. Of the 21,988 total births in the hospital, one child was born with Goltz Syndrome. This was the only X-linked dominant syndrome identified in the population. Sawardekar (2005) hinted for a possible genetic contribution in this child.

**Saudi Arabia**
See: Jordan > Al-Anazy and Zakzouk, 1997.

**References**

**Related CTGA Records**
N/A

**External Links**
http://www.emedicine.com/DERM/topic155.htm
http://www.healthline.com/galecontent/goltz-syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=2092
http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Focal%20Dermal%20Hypoplasia

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