Tooth Agenesis, Selective, X-Linked, 1

Alternative Name
STHAGX1
Hypodontia/Oligodontia, X-Linked, 1

Record Category
Disease phenotype

WHO-ICD
Disorders of tooth development and eruption; Anodontia

Incidence per 100,000 Live Births
N/A

OMIM Number
313500

Mode of Inheritance
X-linked dominant

Gene Map Locus
Xq13.1

Description
Tooth agenesis is a rare developmental dental anomaly in humans. It is characterized by the absence of six or more teeth, lack of development of maxillary and mandibular alveolar bone, problems in tooth eruption and exfoliation. Several forms of syndromic and non-syndromic oligodontia exist. STHAGX1 (Tooth Agenesis, Selective, X-Linked, 1) is a non-syndromic, heritable form of tooth agenesis. Non-syndromic tooth agenesis displays a broad phenotypic heterogeneity, even within the same family.

Diagnoses can be established through clinical examination and intra-oral X-ray. Treatment is given via a multidisciplinary team and is usually very costly. The simplest treatment involves minor prosthodontic interventions like a fixed partial denture.

Molecular Genetics
STHAGX1, as the name suggests, is transmitted in an X-linked dominant fashion. Mutations in the EDA gene, located on the X chromosome, are causal for this condition. The EDA gene is involved in the normal embryological development of the ectodermal appendages, including hair, skin, teeth and sweat glands.

Several mutations in the EDA gene have been identified and reported in the literature in patients with STHAGX1. These include large deletions, insertions, small in-frame deletions, and nonsense and missense mutations.

Epidemiology in the Arab World

Saudi Arabia
Gaczkowska et al., (2016) described an Egyptian male child with sporadic non-syndromic oligodontia. The 7-year old boy was born to non-consanguineous parents. He exhibited oligodontia of both deciduous (lacking lower central incisors, all lateral incisors and left upper first molar) and permanent dentition (lacking 11 teeth). His anterior hairline was low and peculiar with a frontal upsweep. The patient was found to carry a mutation in the EDA gene.

References
Gaczkowska A, Abdalla EM, Dowidar KM, Elhady GM, Jagodzinski PP, Mostowska A. De novo EDA mutations: Variable expression in two Egyptian families. Arch Oral Biol. 2016; 68:21-8. PMID: 27054699

Related CTGA Records
Ectodysplasin A

External Links
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=99798

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