Hypotrichosis-Lymphedema-Telangiectasia Syndrome

Alternative Names
HLTS

Record Category
Disease phenotype

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

Incidence per 100,000 Live Births
0-1

OMIM Number
607823

Mode of Inheritance
Autosomal recessive

Gene Map Locus
20q13.33

Description
Hypotrichosis Lymphedema Telangiectasia Syndrome (HLTS) is a rare disorder characterized, as the name suggests, by the triad of sparse hair, childhood onset-lymphedema, and telangiectasia. The telangiectasia is particularly prominent on the palms of the hands. Lymphedemas are manifested by the disturbances in the maintenance of proper vasculature, especially in the lower limbs and eyelids. Defects in hair follicle development lead to progressive scalp hair loss and the absence of eyebrows and eyelashes. Some patients have been known to develop renal functional abnormalities later on in childhood.

Diagnosis is based on the clinical features and upon molecular diagnostics for confirmation. Treatment is symptomatic and varies from patient to patient.

Molecular Genetics
Mutations in the SOX18 gene have been shown to cause HLTS. The SOX18 protein is a transcription factor that is involved in lymphangiogenesis, angiogenesis, cardiovascular development, and hair follicle formation. It is thus clear to see how defects in the proper functioning of this protein could lead to the phenotype of HLTS. Missense mutations in the gene tend to lead to a dominant mode of transmission, while nonsense mutations have been found to result in a recessively inherited phenotype.

Epidemiology in the Arab World

Jordan
Bastaki et al (2016) described a Jordanian child with HLTS. The boy was born to non-consanguineous healthy parents. An antenatal US had shown ascites with mild pericardial effusion and chylothorax. He was found to have alopecia and abnormal skin colour at birth. Developmental history was normal. Evaluation at 11-months of age revealed the presence of distinct craniofacial features, including microcephaly, periorbital swelling, and absence of eyebrows and eyelashes. He was found to have alopecia totalis, and multiple hemangiomas on the scalp, nape, over the left eyelid, on the back and the scrotal skin. Renal function appeared to be normal. Whole exome sequencing enabled the identification of a novel de novo mutation in the patient within the SOX18 gene, thereby confirming the diagnosis of HLTS.

United Arab Emirates
See Jordan > Bastaki et al, 2016

References
Bastaki F, Mohamed M, Nair P, Saif F, Tawfiq N, Al-Ali MT, Brandau O, Hamzeh AR. A novel SOX18 mutation uncovered in Jordanian patient with hypotrichosis-lymphedema-telangiectasia syndrome by Whole Exome Sequencing. Mol Cell Probes. 2016; 30(1):18-21. PMID: 26631803

Related CTGA Records
SRY-Box 18

External Links
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=69735

https://rarediseases.info.nih.gov/diseases/12827/hypotrichosis-lymphedema-telangiectasia-syndrome

Contributors

Pratibha Nair: 8.2.2017