Leri-Weill Dyschondrosteosis

Alternative Names
LWD
Dyschondrosteosis
DCO
Madelung Deformity

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities> Congenital malformations and deformations of the musculoskeletal system

Incidence per 100,000 Live Births
Unknown

OMIM Number
127300

Mode of Inheritance
Autosomal dominant

Gene Map Locus
Ypter-p11.2, Xpter-p22.32

Description
Leri-Weill Dyschondrosteosis (LWD) is a very rare hereditary disorder characterized by unusually shortened, bowed radius and ulna; Madelung deformity (abnormal deviation of the wrist toward the thumb side of the hand due to shortening of the radius and dislocation of the end portion of the ulna); unusually short lower legs; and mesomelic dwarfism. Sometimes, the humerus is also affected. In many cases, other abnormal features may also associated with LWD such as abnormal bony growths projecting outward from the surface of the shin bones (exostoses of the tibia); unusually short, broad bones in the fingers and toes; and abnormalities of the hipbone (coxa valga). Noticeably, the disease affects females more severely than males.

Molecular Genetics
LWD is transmitted as an autosomal dominant trait and is caused by mutations in the short stature homeobox (SHOX) gene. Normal SHOX gene product acts as a transcription factor for other genes and it is responsible for the development of the skeleton. The signs and symptoms of LWD appear by having one defective copy of the gene, however, the severity of the disease increases if two defective copies of the gene are present. SHOX gene is located on both the X and Y chromosomes in a region known as the pseudoautosomal region. Therefore, both genders have two functional copies of the gene in the normal cases.

Epidemiology in the Arab World
Djibouti
Laroche et al. (1978) described at least one case with Bessel Hagen's deformity and/or Madelung's deformity. No further details could be obtained by the time of editing this record.

Kuwait
Sabry et al. (1997) described the dysmorphic features in three sibs with congenital dyserythropoietic anemia type 1. Findings included growth retardation/short stature, congenital ptosis, abnormal tarsal bones, metatarsal duplication/hypoplasia, nail/phalangeal hypoplasia of fingers and toes, Madelung deformity, syndactyly of toes, and hallux valgus. The patients also showed a very low mitotic index of their peripheral blood lymphocyte cultures. Phenotypic heterogeneity was elicited amongst the three Bedouin sibs. Sabry et al. (1997) suggested that there exists an association between a subset of congenital dyserythropoietic anemia type 1 and a specific form of distal limb anomalies and that other traits, congenital ptosis and low mitotic index, could represent part of the syndrome profile.

References

Laroche R, Sirol J, Durand G, Burrel P, Lunven Y, Piriou A, Peyron JP. [Bessel Hagen's deformity, Madelung's deformity: cosmopolitan diseases (author's transl)]. Med Trop (Mars). 1978; 38(5):555-8. PMID: 311407

Sabry MA, Zaki M, al Awadi SA, al Saleh Q, Mattar MS. Non-haematological traits associated with congenital dyserythropoietic anaemia type 1: a new entity emerging. Clin Dysmorphol. 1997; 6(3):205-12. PMID: 9220189

Related CTGA Records

N/A

External Links

http://ghr.nlm.nih.gov/gene=shox
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=240
http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Dyschondrosteosis

Contributors

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