Recessive Larsen Syndrome

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

OMIM Number
245600

Mode of Inheritance
Autosomal recessive

Description
Larsen’s syndrome is a rare, inherited defect of connective tissue formation. The syndrome characteristics include flattened facies (prominent forehead, depressed nasal bridge, and wide-spaced eyes), multiple congenital dislocations and foot deformities as a result of connective tissue mal-development during gestation. Cleft palate, hydrocephalus, and abnormalities of spinal segmentation that may lead to major spinal instability and spinal cord injury were found in some cases.

Molecular Genetics
There is evidence of both autosomal recessive and dominant inheritance in Larsen syndrome.

Epidemiology in the Arab World

Egypt
Knoblauch et al. (1999) reported two sisters with Larsen syndrome. They were born to clinically unaffected parents who originate from the South of Egypt and were first cousins. Further family history was non-contributory.

United Arab Emirates
In a United Arab Emirates family, Topley et al. (1994) described a brother and sister with Larsen syndrome born to parents related as first cousins. There was no family history of congenital anomalies and both parents, who are United Arab Emirates nationals, were phenotypically normal. Both children had many of the features of Larsen syndrome and most striking was their extremely short stature. However, in the second (female) child there was also diaphragmatic hernia. Topley et al. (1994) proposed that the syndrome in their patients may represent a new variant within the recessive group of Larsen syndrome associated with severe short stature, but not necessarily accompanied by a high risk of early death from respiratory or cardiac complications. Al-Kaissi et al. (2003) commented on the diagnosis of Topley et al. (1996) by saying that the later group emphasized shortness of stature and joint dislocations, but made little reference to the facial abnormality as a means of recognizing the syndrome.

References

Contributors
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