



Furlong Syndrome

Alternative Names

FS

Marfanoid Disorder with Craniosynostosis, Type II

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

610168

Mode of Inheritance

Possibly autosomal dominant

Gene Map Locus

9q33-q34

Description

Furlong syndrome is usually associated with craniosynostosis, normal intelligence, delayed fontanel closure, hypertelorism, ptosis of the eyelids, cleft palate, aortic root anomalies, mild skeletal dysplasia, and normal height.

Molecular Genetics

Furlong syndrome is thought to be caused by mutations in the transforming growth factor, beta receptor I (TGFBRI) gene.

Epidemiology in the Arab World

Lebanon

Megarbane and Hokayem (1998) described a 16 and half-year-old boy with craniosynostosis and marfanoid habitus. He was the first child of

healthy non-consanguineous parents. His psychomotor development was delayed from the beginning. At the age of six months, he was treated for a right inguinal hernia and omphalocele. Physical examination, echocardiography, and total body skeletal radiography were performed to allow a clear case differentiation. He displayed the following features: normal intelligence, muscular hypotrophy, hypoplasia, ptosis of the eyelids, microretrognathism, long thorax, thin limbs, camptodactyly and clinodactyly, kyphoscoliosis, cubitus and hallux valgus, flat foot with long toes, enlarged aortic root, biconvex vertebra, and enlarged diaphyses. Chromosomes were normal (46, XY). According to the findings, the patient had features of two types of craniosynostosis with marfanoid habitus; type I (Shprintzen-Goldberg Craniosynostosis Syndrome) and type II (Furlong Syndrome). Due to the absence of consanguinity in healthy parents of the Lebanese patient and of two previously studied male patients, Megarbane and Hokayem (1998) suggested autosomal dominant inheritance. However, X-linked recessive inheritance could not be excluded.

References

Megarbane A, Hokayem N. Craniosynostosis and marfanoid habitus without mental retardation: report of a third case. *Am J Med Genet.* 1998; 77(2):170-1.

Contributors

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