X-Linked Mental Retardation, with Marfanoid Habitus

Alternative Names
Lujan-Fryns Syndrome

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

OMIM Number
309520

Mode of Inheritance
X-linked dominant with higher penetrance and greater expressivity in males

Description
X-linked mental retardation with marfanoid habitus syndrome is a congenital disorder that is characterized by mental retardation in association with marfanoid features, distinct craniofacial appearance and behavioural problems. Marfanoid features describe the tall and stooped posture with long, hyper-extensible fingers and toes. The distinct craniofacial appearance for this disorder includes large forehead, long and narrow face, maxillary hypoplasia, long nose with high and narrow nasal bridge, short and deep philtrum, thin upper lip, and highly arched palate. Behavioural features are emotional instability, hyperactivity and shyness. A hypernasal voice and generalized hypotonia are often present. The syndrome affects males more than females and it may occur more frequently among mentally retarded patients and psychiatric patients. Therefore, X-linked mental retardation with marfanoid habitus syndrome should be considered in the differential diagnosis of schizophrenia. Almost the marfanoid features become apparent after puberty. Secondary sexual development and testicular size is normal. There is no specific treatment for this condition, but patients require specialized education and psychological follow-up.

Molecular Genetics
X-linked mental retardation with marfanoid habitus syndrome is a development disorder of genetic origin. It is transmitted as X-linked dominant manner and it affects predominantly males. The cause of this syndrome is unknown. However, it is suggested that a mutation in a structural connective tissue gene can cause the disorder because two related patients with venricular septal defect and aortic root dilatation have been described.

Epidemiology in the Arab World

Lebanon
Megarbane and Chammas (1997) reported a Lebanese young adult male with a marfanoid habitus, a severe mental retardation, and a peculiar face. He was the son of healthy non consanguineous parents. He was evaluated at birth to have a long face and slender members. The psychomotor development was delayed from the beginning. When he was seen at the age of 20 years, he was very thin man with general muscular hypotrophy and a centripetal mild obesity. His voice was hyper nasal and he was sociable and jovial despite some nervousness. Craniofacial appearance included triangular thin face, large forehead, a narrow pointed chin, slightly rotated and protruded ears, a deep philtrum, a small mouth, and a highly narrow arched palate. A kyphoscoliosis with a right hump was noted. His fingers were thin with clinodactily of two fingers bilaterally and on both feet, the second toe was hammer-shaped. Genitalia were delayed in development. Echocardiography revealed mitral regurgitation and skeletal radiography displayed the absence of mandibular angles, vertebral bloc of D3-D4, and malformation of extremities. Hormonal evaluation suggested primary testicular failure because of the high levels of FSH, LH, and prolactin. Studying the family history revealed that his younger sibling presented the same
dysmorphic features and died at two months from incorrigible heart disease. He also had a normal sister. Some findings existed in the patient, and they are absent in the ones with Lujan-Fryns syndrome like, the absence of mandibular angles, the osseous malformations, the toes positioning, the hormonal involvement, and the severe mental retardation. Also, the phenotype of the patient was presented from birth, while in Lujan-Fryns syndrome it was reported to be diagnosed only rarely before puberty because of varying phenotype with age. Therefore, Megarbane and Chammas (1997) described the case to be a diagnostic challenge.

References

Contributors
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