

A Rare Case of Lujan Fryn Syndrome

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Case Report

Abstract: The Lujan-Fryns syndrome is a X-linked syndrome, affecting predominantly males. The diagnosis is based on the presence of the clinical manifestations. There is no specific treatment for this condition. Patients need special education and psychological follow-up, and attention should be given to diagnose early psychiatric disorders.

Keywords: Rare, Lujan Fryn Syndrome, clinical manifestations, follow-up.

Introduction

The Lujan-Fryns syndrome(LFS) is a X-linked syndrome with mental retardation and marfanoid habitus, affecting predominantly males. The prevalence is not known for the general population. This syndrome is associated with distinct facial dysmorphism, long narrow face, maxillary hypoplasia, small mandible and prominent forehead, tall marfanoid stature, long slender extremities, and behavioural problems. The diagnosis is based on the presence of the clinical manifestations¹. Complete or partial agenesis of corpus callosum, ascending aorta aneurysm and ventricular septal defects are manifestations of LFS, and thus brain MRI and echocardiogram should be part of the routine evaluation². Additionally, the high prevalence of psychopathological alterations in these patients suggests the need for psychiatric evaluation at the time of diagnosis^{2,3}. The exact etiology of the disorder is unclear³. Prenatal testing is not possible¹. There is no specific treatment for this condition. Patients need special education and psychological follow-up, and attention should be given to diagnose early psychiatric disorders¹.

Case

We present the case of a 8 year old boy who was brought to us with complaints of fever and cough since 15 days. Patient was admitted and evaluated. All routine investigations were done. He was born out of a third degree consanguineous marriage by normal vaginal delivery at term. Antenatal and perinatal history was uneventful but he had delayed developmental milestones, walking independently only after 3 years. He required assistance for carrying out his routine activities. On clinical examination patient had dysmorphic craniofacial features like macrocephaly with a prominent forehead, long narrow face, maxillary hypoplasia, small mandible, long nose with high and narrow nasal bridge, high arched palate, receding chin and large ears (fig 1). Patient had hypernasal speech, pectus excavatum, tall marfanoid status with long slender extremities, hyperextensible long digits (fig 2). He had mild mental retardation and was extremely shy with intermittent episodes of aggression. Other mental and behavioural abilities such as problem solving, reasoning was preserved. Eye examination was normal and genitals were of normal size. His height was 135cms, arm span was 138cms(fig 3), weight 28 kg and head circumference 53cm. Echocardiography revealed small sized ventricular septal defect with mild pericardial effusion. MRI of brain, ultrasonography of abdomen were normal. Serum homocysteine levels were within normal limits. The patient was treated with antipyretics and routine antibiotics. He improved within a week and was discharged and advised regular follow up for behavioural abnormalities.



Figure 1



Figure 2



Figure 3

Discussion

The disorder is inherited in an X-linked manner, and is attributed to a missense mutation in the MED12 gene. There is currently no treatment or therapy for the underlying MED 12 malfunction, and the exact etiology of the disorder remains unclear³. Kindred have been studied in which mental retardation and marfanoid clinical features are present in several individuals. The pedigree is consistent with X-linked recessive inheritance⁴. Behavioural and psychiatric disorders like attention deficit hyperactivity disorder, autism, schizophrenia are frequently associated with this syndrome⁵. Cardiovascular defects like ventricular septal defect and aortic root dilatation have been reported in a few cases and hence echocardiography should be part of the investigations⁶. Close attention and specialized follow-up care, including neuropsychological evaluation methods and therapies, and special education, should be

given to diagnose and prevent psychiatric disorders and related behavioral problems.³

References

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