

## Case Reports

# Lujan-Fryns Syndrome

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### Introduction

The Lujan-Fryns syndrome or X-linked mental retardation with marfanoid habitus syndrome is an X-linked form of mental retardation associated with tall, marfanoid stature, distinct facial dysmorphism and behavioural problems<sup>1</sup>. Few cases have been described in the literature. Individuals with Lujan syndrome may have behavioural disturbances ranging from shyness to frank psychosis<sup>2</sup>. Our index case had a marfanoid built, mental retardation and attention deficit hyperactivity disorder. Similar findings were noted in his 11 year old sister.

### Case Report

We present the case of a 14 year old boy and his 11 year old sister who were brought to us with the complaints of mental retardation and hyperactivity. The 14 year old boy was the oldest of four children born out of a consanguineous marriage. His height was 149cm (Z score 1.65), arm span 152 cm, weight 28 kg (Z score 2.87) and head circumference 50 cm. He had a tall thin build, long face, long thin nose, maxillary hypoplasia (figure 1) and long hyperextensible digits (figure 2).

He had mild mental retardation and was aggressive, impulsive and defiant. He was hyperactive with a short attention span. He had hypernasal speech. His eye examination was normal and cardio-respiratory examination revealed scoliosis and pectus carinatum deformities. Genitals were of normal size. Echocardiography, ultrasound study of the abdomen and MRI brain revealed no abnormality. Serum homocysteine level was normal (11.6 micromols per litre).

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**Figure 1: Tall thin build, long arms and long face**



**Figure 2: Long thin fingers**

He was healthy normally delivered at term. Antenatal and perinatal history was unremarkable, but he had delayed developmental milestones, walking independently by 3 years of age. He could not carry out activities of daily living independently and required assistance. He was on antipsychotics for the last 2 years and had some improvement. There was no constipation or genitourinary problem.

His 11 year old sister also had a thin build, long face, long nose and long fingers.



**Figure 3: 11 year old girl with tall thin stature, long face and long arms**

Her height was 132 cm (Z score 1.85), arm span 133 cm, weight 21 kg (Z score 2.55) and head circumference 47 cm. She was less aggressive and talkative than her brother but had hyperactivity, impulsivity and mild mental retardation. Her birth history was uneventful but developmental milestones were grossly delayed. The rest of the siblings as well as other family members were unaffected.

### Discussion

The siblings were diagnosed as having Lujan-Fryns syndrome based on the clinical picture. Lujan-Fryns syndrome is characterised by tall stature with asthenic habitus, a tall narrow face, maxillary hypoplasia, a high narrow palate with dental crowding, a small or receding chin, long hands with hyperextensible digits, hypernasal speech, hypotonia, mild-to-moderate mental retardation, behavioural aberrations and dysgenesis of the corpus callosum<sup>3</sup>. Behavioural and psychiatric disorders like attention deficit hyperactivity disorder, autism, schizophrenia, etc. are frequently associated with this syndrome<sup>2,4,5</sup>. 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 investigations<sup>6</sup>. Cortical malformations like complete or partial agenesis of the corpus callosum have also been seen in some cases<sup>7</sup>.

It is an X linked mental retardation syndrome attributed to a missense mutation in the MED12 gene<sup>3</sup>. The prevalence in the general population is not known. Lujan-Fryns syndrome affects predominantly males<sup>1</sup>. Two cases in females have also been described. The diagnosis is based mainly on clinical manifestations. There is no known treatment. The psychiatric problems in this syndrome are of concern.

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