electromyography showed positive sharp waves, fibrillations, and absence of functioning motor units in muscles innervated by the left radial nerve, with the exception of the triceps.

**DISCUSSION**

Appleton¹, in 1911, reported the first description of this anatomical variation based on a forearm dissection. He found only the posterior radial nerve interosseous branch with absent SRN, below the elbow. On the dorsum of the hand, the LACN extended out beyond its usual distribution to supply the SRN territory (Figure). The dorsal branch of the ulnar nerve had greater ramifications than those usually observed by completing the innervation of the dorsal hand. Other authors²⁵ have studied the distribution of LACN and SRN in the dorsal of the hand and confirmed the existence of this anatomical variation.

In our assessment, the occurrence of an anatomical variation involving LACN and SRN, in which the cutaneous territory of the SRN is wholly or partially supplied by the LACN, can explain the data obtained in the presence of total lesion of the radial nerve in the arm. In cases of proximal radial nerve injury, the occurrence of this variation may lead to diagnostic errors in the EDX study, in which a total axonal damage may erroneously be considered as partial lesion with a conduction block component.

References


**Marfanoid features and X-linked mental retardation associated with craniofacial abnormalities: the Lujan-Fryns syndrome**

*Aspectos marfanoides e retardo mental ligado ao X associados a anormalidades craniofaciais: síndrome de Lujan-Fryns*

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Marfanoid features associated with mental retardation are often a diagnostic challenge for general neurologists, since several genetic and inborn metabolic diseases present this clinical spectrum. Some of these diseases have typical clinical markers that may aid diagnosis. For instance, Lujan-Fryns syndrome is a recessive X-linked condition characterized by mental retardation, marfanoid habitus, and facial dysmorphisms

Herein, we described a male patient with marfanoid habitus and mental retardation, in whom the presence of facial dysmorphisms was the clue for diagnosing the Lujan-Fryns syndrome.

**CASE REPORT**

A 25-year-old man presented to the Division of General Neurology, at Universidade Federal de São Paulo, with developmental delay and learning disabilities since childhood. His family history was unremarkable and there was no parental consanguinity. He had another brother with possible marfanoid features and learning disabilities, and other three ones and a sister that had not been evaluated. A possible X-linked inherited disorder was suspected. On examination, the patient
presented high stature, prominent forehead, large nose, deep and short filters, long narrow face, low-set ears, small mandible, long arms and fingers, and hypotonia (Figure). He also showed severe mental retardation. Cardiologic and ophthalmologic evaluations were normal. Brain magnetic resonance resulted normal. His karyotype was 46 XY, and fragile X genetic test was negative. Serum homocysteine was 9.95 mmol/L (normal range from 5 to 15 mmol/L). With this clinical presentation and after exclusion of other diseases, the diagnosis of Lujan-Fryns syndrome was suggested.

**DISCUSSION**

In 1984, a new form of X-linked mental retardation was described. The so-called Lujan-Fryns syndrome or X-linked mental retardation with marfanoid habitus syndrome is a rare syndromal X-linked form of mental retardation (mild to moderate mental retardation), associated with tall marfanoid stature, distinct facial dysmorphism, and behavioral problems. Despite known genetic origin, there is no genetic test available to acknowledge the diagnosis of Lujan-Fryns syndrome, though a mutation in MED12 gene has already been described.

Craniofacial features in Lujan-Fryns syndrome include prominent forehead, long narrow face, maxillary hypoplasia, small mandible, long nose with high and narrow nasal bridge, short and deep philtra, thin upper lip, highly arched palate, receding chin, and low-set retroverted normal shaped ears. In some cases, brain imaging shows corpus callosum agenesis.

The diagnosis of Lujan-Fryns syndrome is based on clinical manifestations and on exclusion of other conditions, such as homocystinuria, Fragile X syndrome, 22q11 deletion syndrome, Klinefelter and Marfan syndromes. Opitz-Kaveggia syndrome is an allelic disorder with an overlapping phenotype. It should also be remembered as a differential diagnosis of schizophrenia, considering the important behavioral problems associated with that condition (hyperactivity, aggressiveness, and autistic-like behavior). There is no available treatment for this syndrome. In general, patients need special education and psychological support. Genetic counseling is recommended, considering transmission pattern of the syndrome.

In summary, our report reinforces craniofacial features as a clue for the diagnosis of Lujan-Fryns syndrome in the context of marfanoid habitus and possible X-linked mental retardation.

**Figure.** Patient has prominent forehead, large nose, deep and short filters, long narrow face, low-set ears, small mandible, and marfanoid features with long arms and fingers.

**References**