

Unilateral abdominal protrusion as the main diagnostic sign of facioscapulohumeral dystrophy

Protrusão abdominal unilateral como principal sinal diagnóstico de distrofia fâscio-escápulo-umeral

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A 48-year-old man presented with slowly progressive right arm weakness and cervical pain over the last 20 years. Previous electromyography/nerve conduction studies showed a neurogenic pattern on the right cervical myotomes and he received the diagnosis of focal motor neuron disease. No relevant family history was identified. On our initial evaluation, an asymmetrical abdominal protrusion was noted as well as a mild right arm weakness and atrophy (Figure), leading to the suspicion of facioscapulohumeral dystrophy (FSHD). A DNA test detected a D4Z4 repeat contraction on chromosome 4qA, confirming the diagnosis. The frequency of an asymmetrical protuberant abdomen in these patients is not known. Although more than 80% of FSHD patients have affected abdominal muscles and 78% have some degree of asymmetry shown in MRI evaluation, less than 3% present with asymmetrical abdominal muscle involvement¹. Even though it is an uncommon sign, the association of abdominal

weakness and asymmetry is a key feature to consider FSHD diagnosis, especially in milder forms².



Figure. Right abdominal protuberance and right humeral atrophy.

References

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