Muscle MRI in Bethlem myopathy

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DESCRIPTION

A 44-year-old man presented with progressive limb girdle weakness from 10 years of age. The inheritance pattern was autosomal dominant; two siblings, his mother and maternal grandfather were similarly affected. On examination there was an exaggerated lumbar lordosis, wasting of proximal limb musculature, and contractures at elbows, wrists and finger flexors. There was moderate symmetrical proximal upper and lower limb weakness without facial weakness or cardiorespiratory involvement. Previous investigations included electromyography which was myopathic, creatine kinase 2–4× normal and dystrophic muscle biopsy but no definitive diagnosis had been made.

The differential diagnosis is of a collagen VI myopathy (Bethlem), laminopathy or limb girdle muscular dystrophy. This differential diagnosis can be narrowed with lower limb muscle MRI¹ which was performed. The pattern (figure 1) was typical of Bethlem myopathy. The muscle biopsy was sent for further analysis which showed marked reduction of collagen VI at the basal lamina of most fibres. Subsequent genetic studies identified a heterozygous missense mutation in *COL6A2*.

Bethlem myopathy is the mild end of the collagen-VI-related myopathy spectrum. Although symptoms start in childhood, ambulation is typically maintained into adulthood. Contractures and mildly elevated creatine kinase are also typical.² Bethlem myopathy has a highly specific pattern of muscle involvement on imaging with 90% sensitivity.^{1–3} The peripheral involvement of the vasti muscles and antero-central involvement of rectus femoris^{1–3} is particularly striking and once seen easily recognised subsequently, facilitating definitive genetic diagnosis.

Figure 1 T1-weighted MRI, transverse slice at mid left thigh. Normal muscle has low-signal intensity, fat has high-signal intensity. The quadriceps muscle shows the typical pattern for Bethlem myopathy: peripheral fat infiltration with central sparing of the vasti muscles (VM, vastus medialis; VI, vastus intermedius; VL, vastus lateralis) and both peripheral and antero-central infiltration of rectus femoris (RF) leaving a 'U' shape of less-affected muscle. Central sparing of the hamstring muscles (BF, biceps femoris; ST, semitendinosus; SM, semimembranosus) is also apparent.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

Learning points

- ▶ Consider Bethlem myopathy in adults with progressive myopathy and contractures.
- ▶ A highly specific pattern of muscle involvement may be seen on lower limb MRI in Bethlem myopathy and guide definitive genetic testing.

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To cite: Morrow JM, Pitceathly RDS, Quinlivan RM, *et al*. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-008596

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