Steinert's disease

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DESCRIPTION

On a cold winter's day, a 52-year-old man presented with a 3-year history of fatigue and gait disturbance. He had previously been diagnosed with Steinert's disease (type 1 myotonic dystrophy) through genetic testing showing cytosine–thymine–guanine repeats in the dystrophia myotonica–protein kinase gene. He sometimes found it difficult to sit up in bed because of weakness of his neck muscles. On examination, marked atrophy of the temporal, masseter and sternocleidomastoid muscles with frontal alopecia was observed (figure 1). Muscle strength was intact but was grade 4/5 in the hip extensors and flexors. Grip myotonia

Figure 1 A marked atrophy of the temporal, masseter and sternocleidomastoid muscles with frontal alopecia.

Video 1 Grip myotonia.

Video 2 Percussion myotonia.

and percussion myotonia were observed (videos 1 and 2). In Steinert's disease, myotonia is observed most consistently in small muscles including the hand intrinsic muscles, typically elicited as grip myotonia and percussion myotonia.¹ Muscular symptoms are seen especially in the early stages of the disease, typically aggravated by cold and stress.² The day the patient visited was a cold winter's day, which may have aggravated his symptoms.

Learning points

- ▶ Steinert's disease (myotonic dystrophy type 1) is a multisystem disorder mainly characterised by skeletal muscle weakness and myotonia.
- ▶ Myotonia is prominent especially in the early stages of the illness, which is aggravated by cold and stress. It is seen most consistently in the facial and hand intrinsic muscles.

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REFERENCES

- 1 Avaria M, Patterson V. Myotonic dystrophy: relative sensitivity of symptoms signs and abnormal investigations. *Ulster Med J* 1994;63:151.
- 2 Machuca-Tzili L, Brook D, Hilton-Jones D. Clinical and molecular aspects of the myotonic dystrophies: a review. *Muscle Nerve* 2005;32:1.

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