A patient that changed my practice

Excuse me, is that your own hair?

Figure 1 The patient with and without her wig. Note the mild ptosis.

LESSONS LEARNED

- Always ask the patient to remove their wig! Well, perhaps not every patient, but had the patient been asked specifically about baldness at her initial presentation and her alopecia uncovered, the correct diagnosis would have been considered at that stage.
- Perhaps a more practical lesson would be 'Never assume the hair on the head is real. Don't forget wigs!'
- Myotonia may only be demonstrable electrically and not apparent clinically, despite appropriate cooling of the extremities.

Gillian Hall

Department of Clinical Neurosciences, Western General Hospital, Edinburgh EH4 2XU; Email: ghall@skull.dcn.ed.ac.uk *Practical Neurology*, 2002, 242–243

THE HISTORY

A 54-year-old lady, Mrs T, presented with deteriorating mobility. This had come on gradually over several years. Two years prior to presentation, while on holiday she could walk her dog 2–3 miles, twice a day. The following year she was aware she could not walk as far, and at presentation she could only walk short distances secondary to fatigue and a fear of falling, although there had been no falls. She could manage one flight of stairs with difficulty and complained of problems opening jars. In retrospect, a friend had commented on thinning of her calves some 4 or 5 years previously. There had been no rash or myalgia, nor any history suggestive of myoglobinuria. Her only sensory symptom was of numbness on the anterolateral aspect of the right thigh.

She had been born at full term following a normal labour and delivery. There were no reported respiratory or feeding problems. Although not interested in sport at school, she recalls being able to run around with her peers and do all that she wanted. There was no known family history of muscle disease. She had had a cholecystectomy and excision of a left axillary lipoma.

THE EXAMINATION

On examination she was in sinus rhythm. There was thinning of the forearm, calf and tibial muscles. There was no *pes cavus*. No myotonia was demonstrated. She had mild bilateral ptosis with weakness of eye closure, neck flexion and exten-

sion. In the upper limbs there was mild weakness of triceps but more significant weakness of wrist flexors and extensors and of the small muscles of the hand. In the lower limbs there was distal weakness with good proximal power. Reflexes were all diminished. Sensory disturbance was limited to the distribution of the right lateral cutaneous nerve of the thigh.

THE TESTS

She was felt to have an acquired muscle disorder and an incidental right meralgia paraesthetica. Nerve conduction studies showed reduced amplitude compound motor action potentials, but were otherwise normal. However, electromyography revealed myopathic motor units and marked myotonic discharges in all muscle groups studied. This raised the question of myotonic dystrophy.

THE DIAGNOSTIC PROCEDURE

On subsequent questioning, the patient revealed that both her parents, now diseased, had had cataracts and that her father had had significant frontal balding for as long as she could recall. Her father had no siblings. When the question of baldness was raised, the patient removed her hitherto unsuspected wig, revealing her own frontal baldness (see Fig. 1). Genetic analysis identified a pathogenic expansion confirming the diagnosis of myotonic dystrophy. The patient was informed of the diagnosis and counselled appropriately. She was an only child and has no children of her own.