

Duchenne muscular dystrophy

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DESCRIPTION

A 15-year-old boy presented with progressive proximal weakness of the lower limbs starting at 4 years of age followed by involvement of the upper limbs. He is the product of a consanguineous marriage; he had a family history of similar disease in a second-degree cousin and also had a history of delayed motor developmental milestones since birth. Clinically, he had flaccid quadriplegia with wasting and contractures without any sensory or neurological involvement. His weakness worsened leading to an inability to walk without support by the age of 9 and total wheelchair dependence by the age of 12. He was frequently admitted to hospital with chest infections.

The patient's creatine kinase was 2600 IU/L (normal 50–150 IU/L) and muscle biopsy from left quadriceps showed rounded small muscle fibres with evidence of degeneration and an absence of dystrophin protein. He was diagnosed as a case of duchenne muscular dystrophy. He is presently bed bound with weakness and contractures of all limbs and spinal deformities as shown in figure 1. He developed scoliosis at the age of 12 which has gradually worsened to the present state as shown in figures 2 and 3.



Figure 1 Duchenne muscular dystrophy.



Figure 2 Lateral view.

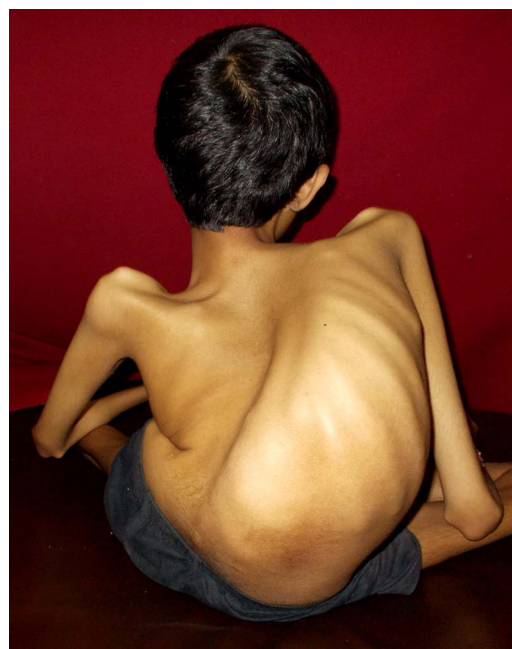


Figure 3 Back view.

He was treated with regular physiotherapy, limb-strengthening exercises, 0.9 mg/kg/day deflazacort, aggressive management of respiratory infections, periodical cardiac and respiratory follow-up, genetic counselling and other supportive therapies.

Learning points

- ▶ Duchenne muscle dystrophy is a progressive inherited myopathy with an early onset in childhood.¹
- ▶ It progresses to the bed-bound state in the second decade of life and patients usually succumb to respiratory or cardiac complications.
- ▶ Conservative management, active physiotherapy, genetic counselling and other supportive therapies hold the key to successful management of these cases.²

Contributors All authors have contributed to the manuscript.

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