

## Toe-walking as the first presenting symptom of Emery-Dreifuss muscular dystrophy type 1

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Up to 10% of the children show persistent toe walking patterns. Toe walking usually starts with or shortly after beginning to walk. Clinical and differential diagnosis of the common idiopathic toe walking and secondary toe walking caused by neuromuscular diseases is often difficult.

**Case report:** A 4-year-old boy developed progressive toe walking after initially normal history. There was no muscular weakness at this time. At the age of 5 the neuropaediatric examination was as follows: primary toe walking with secondary short calcaneus, weak deep tendon reflexes, slight distal muscle weakness. Creatinkinase was 10-fold elevated to 25.3 ukat/l (range: <2,90). Electromyography: increased polyphasic potentials. Muscle biopsy: a few myopathological changes with necrosis of single fibers, immunohistochemistry of emerin was negative. Genetic: mutation (g.618\_619delAG) in the STA gene which causes an early stop of translation and leads therefore most likely to a non functional emerin. The results are consistent with the x-chromosomal Emery-Dreifuss muscular dystrophy type I.

At the age of 7 the achilles tendon was prolonged, progressive contractures of the hip abductors, scoliosis and contractures of the neck muscles occurred. Muscular strength was measured distally 3/5, proximally 4/5, the tendon reflexes were absent.

**Conclusion:** Toe walking first seen after the age of 3 should always encourage the search for neuromuscular disorders. Rarely toe walking and contractures without any muscular weakness are the first feature of an Emery-Dreifuss muscular dystrophy as in our case. Confusion with the common idiopathic toe walking inhibits therapy and prophylaxis of progressive contractures and skeleton deformities. Serum creatinkinase concentration might be therefore used as a screening for muscle dystrophies in toe walkers.