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Evolution of foot manifestations in children with Charcot-Marie-Tooth disease

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Introduction

Charcot-Marie-Tooth disease (CMT) is the most common genetic nerve disorder. The most prevalent form, CMT1A, is characterised by demyelinating neuropathy with progressive foot and ankle weakness, contractures and deformity. The wide range of foot/ankle manifestations in CMT1A complicates the assessment, diagnosis and therapy. We aimed to characterise foot and ankle strength, flexibility, morphology and symptoms in children with CMT1A.

Methods

81 children aged 2–16 y with CMT1A were objectively assessed for strength (dorsiflexion, plantarflexion, inversion, eversion) with hand-held dynamometry [1], ankle dorsiflexion flexibility using the weight bearing lunge [2] and foot morphology with the Foot Posture Index (FPI) [3]. We also looked for difficulties in heel or tip-toe walking, foot drop during gait, and questioned about foot/ankle pain, cramps, ankle instability, trips and falls during walking.

Results

Mean strength was: dorsiflexion 55N (SD, 20), plantarflexion 175N (SD, 49), inversion 76N (SD, 29), eversion 63N (SD, 24). While age (r = 0.65–0.80, $p < 0.001$) correlated with foot strength, compared to age-equivalent norms, mean strength was lower in all ages of childhood CMT1A. This disparity was most evident for dorsiflexion (Figure 1).

Ankle dorsiflexion ranged from 7–41° (mean 25°, SD 7). Compared to normal, ankle flexibility was lower in children at all ages with CMT1A. Interestingly, there were no significant correlations between ankle flexibility and age, height or body weight.

Foot morphology ranged from a FPI of -12 to +11 (mean 2, SD 5). 35% of children exhibited pes cavus, 39% had normal feet and 26% had pes planus. There was a significant correlation between FPI and age (r = -0.47, $p < 0.001$), indicating the evolution of pes cavus during growth (Figure 2).

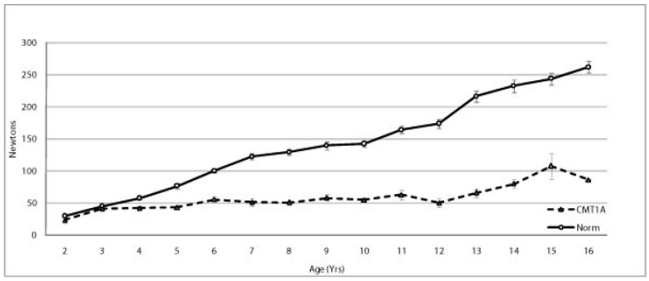

Figure 1
Foot dorsiflexion weakness in CMT.

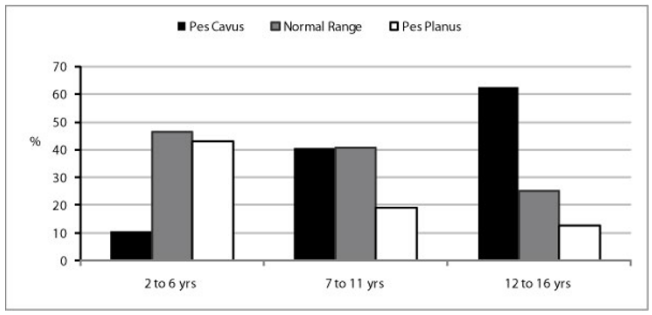

Figure 2
Age-related foot morphology in CMT.

Compared to norms, foot morphology in preschool children with CMT1A did not differ, but from the age of 5 years, deviated from normal towards a more cavoid appearance.

Difficulty heel walking occurred in 82% of children, 4% had difficulty tip-toe walking and 4% exhibited foot drop. 27% reported foot pain, 36% reported cramps, 72% described ankle instability, 63% reported frequent trips and 47% falls. Foot pain, cramps, toe-walking and foot drop worsened with age ($p < 0.05$).

Conclusion

Children with CMT1A experience foot weakness, contracture and deformity from an early age. These manifestations are expected to impact negatively on daily function. Early intervention targeting the foot and ankle may prevent long-term disability in CMT1A.

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