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DETERMINANTS OF BALANCE DEFICIT IN CHILDREN WITH CHARCOT-MARIE-TOOTH DISEASE

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SUMMARY

Balance deficits resulting in ankle sprains, trips and falls are a considerable problem for children with Charcot-Marie-Tooth disease (CMT). This study identified a number of factors associated with balance deficit in 226 children with CMT. Multiple regression modeling identified problems with dorsiflexion strength, long jump power and functional gait tasks as the most significant independent predictors of poor balance in children with CMT, contributing to a model that predicted 48% of balance deficit.

INTRODUCTION

Charcot-Marie-Tooth disease (CMT), the most common neuromuscular disorder, affects millions of people worldwide. The peripheral nerve demyelination and axonal loss associated with the disease causes progressive weakness and deformity of the hands and feet, sensory loss and difficulty walking.

Poor balance resulting in frequent and debilitating ankle sprains, trips and falls are also common clinical sequelae in children with CMT. The aim of this study was to characterize the degree of balance problems in children with CMT and identify factors related to balance deficit.

METHODS

The sample comprised 226 patients (116 girls and 110 boys) with CMT aged 3-20 years (mean 11, SD 4 years) who attended one of the Inherited Neuropathies Consortium centers (88 The Children's Hospital at Westmead, Sydney Australia; 46 Wayne State University, Detroit, MI; 28 Children's Hospital of Philadelphia; 28 UCL Institute of Child Health & Great Ormond Street Hospital, London, UK; 22 Carlo Besta Neurological Institute, Milan, Italy; 9 University of Iowa, Iowa City IA; 5 University of Rochester, Rochester, NY). CMT subtypes were: 49% Type

1A; 10% Type 1B-E; 6% Type 2A-L; 4% Type 4A-J; 3% X1-5; 28% Unidentified gene.

As part of the validated CMT Pediatric Scale [1], balance was measured using subtests of the Bruininks-Oseretsky Test of Motor Proficiency, 2nd Ed. (BOT-2, Pearson Education, Upper Saddle River, New Jersey) in all children. Timed and performance-based BOT-2 balance test items include: standing and walking balance included: standing with feet apart on a line (eyes open/closed); walking forward on a line and walking forward heel-to-toe on a line; standing on one leg on a line (eyes open/closed); standing on one leg on a balance beam (eyes open/closed); and standing heel-to-toe on a balance beam.

Other lower limb measures included in the CMT Pediatric Scale were also collected: foot alignment (Foot Posture Index), ankle flexibility (lunge test), dorsiflexion and plantarflexion foot strength (hand-held dynamometry), sensation (pinprick and vibration), functional gait tasks (difficulty heel walking, difficulty toe walking and presence of foot drop), power (long jump) and endurance (6-minute walk test).

For data analysis, items dependent on growth and development (balance, strength, power and endurance) were converted to z-scores based on age and gender-matched normative reference values using the CMT Pediatric Scale calculator (<http://cmtped.org/>). Descriptive statistics were calculated to characterize the study sample in SPSS (IBM SPSS Statistics for Windows, Version 21.0. Armonk, NY: IBM Corp). Data were subsequently analyzed from one limb only (dominant limb) to satisfy the independence requirements for statistical analysis. Normality of data distribution was assessed using the Kolmogorov-Smirnov

test with Lilliefors significance correction, and the appropriate parametric or nonparametric tests subsequently employed. A hierarchical multivariate regression model was constructed to determine whether deficits in balance could be independently explained by other items of the CMT Pediatric Scale.

RESULTS AND DISCUSSION

Balance was markedly affected for most children with CMT (mean z-score -3.4, SD 3.1, range -13.7 to 1.1). Those with CMT Type 2A-L (-4.5, SD 3.6) and CMT Type 4A-J (-6.6, SD 3.8) were significantly worse than CMT Type 1A (-2.1, SD 2.2).

Factors related to balance in univariate analyses were age ($r=-0.287$, $p=0.001$), dorsiflexion foot strength ($r=0.427$, $p<0.0001$), plantarflexion foot strength ($r=0.244$, $p<0.0001$), sensation (pinprick $r=-0.233$, $p<0.0001$; vibration $r=-0.317$, $p<0.0001$), functional gait tasks ($r=-0.613$, $p<0.0001$), power ($r=0.600$, $p<0.0001$) and endurance ($r=0.582$, $p<0.0001$). Foot alignment, ankle flexibility and gender were not related to balance ($p>0.05$).

Multivariate regression analysis revealed three measures that were significant independent determinants of balance deficit in CMT: dorsiflexion strength (β weight 0.120, $p=0.031$), long jump power (β weight 0.337, $p<0.0001$) and functional gait tasks (β weight 0.372, $p<0.0001$). The model explained a total 48.2% of the variance in balance deficit in children with CMT.

CONCLUSIONS

Balance deficit was found to be an almost universal manifestation in this large sample of children with CMT. Multiple regression modeling identified problems with dorsiflexion strength, long jump power and functional gait tasks as the most significant independent predictors of poor balance in children with CMT, contributing to a model that predicted 48% of balance deficit. Therapeutic interventions that aim to improve balance in children with CMT could specifically target foot and ankle weakness to prevent long-term morbidity associated with ankle sprains, trips and falls.

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