Factors influencing the attainment of major motor milestones in CDKL5 Deficiency Disorder.

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Abstract

Aim

The aims of this study were to investigate the influence of factors at birth and in infancy on the likelihood of children with CDKL5 Deficiency Disorder achieving independent sitting and walking.

Methods

Data on 350 individuals with a pathogenic *CDKL5* mutation was sourced from the International CDKL5 Disorder Database. Two models were developed to accommodate explanatory variables. The first model included factors available at birth (e.g., sex, mutation group and mosaicism) and the second additionally included factors available during the first year of life (e.g., age at seizure onset, number of anti-seizure medications (ASMs) used, experience of a honeymoon period and formal therapy). Cox proportional hazard regression was used to model the time to achieve the milestones, and the probability of attaining the outcomes at specific ages was estimated by evaluating the time-to-event function at specific covariate values.

Results

Independent sitting and walking were achieved by 177/350 and 57/325 children respectively. By 7 years of age, 67.1% of females but only 37.3% of males could sit independently. About a quarter each of females and males achieved independent walking by 8 and 6 years, respectively. When observed from birth, female gender, a late truncating mutation and mosaicism impacted most positively on the likelihood of independent sitting. When observed from one year, later seizure onset and experiencing a honeymoon period, and less so, having been on three or less ASMs and receiving therapy during their first year also improved the likelihood of independent sitting. Similarly, factors that favoured sitting (except gender) improved the likelihood of achieving independent walking. Mosaicism improved but a truncation between aa178 and aa781 reduced the likelihood of achieving independent sitting and walking.

Conclusion

We have shown that it is possible to utilise factors occurring early in life to inform the likelihood of future motor development in CDD.

Keywords

CDKL5 Deficincy Disorder; milestones; factors at birth and in infancy

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