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PSYCHOSOCIAL ASPECTS OF DRAVET SYNDROME

Akademisk avhandling

Som för avläggande av medicine doktorsexamen vid Sahlgrenska akademin, Göteborgs universitet kommer att offentligen försvaras i aulan Tallen, Behandlingsvägen 7, Drottning Silvias Barnsjukhus, SU, Göteborg, torsdagen den 8 februari 2024, 13.00
av Björn Bjurulf

Fakultetsopponent:

Professor Andreas Brunklaus

School of Health and Wellbeing, College of Medical, Veterinary and Life Sciences, University of Glasgow, Great Britain

Avhandlingen baseras på följande delarbeten:

I. Bjurulf, B., Reilly, C., Sigurdsson, G.V., Thunstrom, S., Kolbjer, S., Hallböök, T. 2022. Dravet syndrome in children-A population-based study. *Epilepsy Res* 182: 106922.

II. Bjurulf, B., Reilly, C., Hallböök, T. 2022. Caregiver reported seizure precipitants and measures to prevent seizures in children with Dravet syndrome. *Seizure* 103: 3-10.

III. Reilly, C., Bjurulf, B., Hallböök, T. 2022. Intellectual functioning and adaptive behaviour in children with Dravet syndrome: A population-based study. *Dev Med Child Neurol.* 65(6): 831-837.

IV. Bjurulf B., Reilly, C., Hallböök, T. 2023. Caregiver reported Behavior, Sleep and Quality of Life, in Children with Dravet Syndrome: A population-based study. *Epilepsy Behav*. Accepted manuscript.

SAHLGRENSKA AKADEMIN
INSTITUTIONEN FÖR KLINISKA VETENSKAPER

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PSYCHOSOCIAL ASPECTS OF DRAVET SYNDROME

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Abstract

Dravet syndrome (DS) is a developmental and epileptic encephalopathy, associated with significant neurodevelopmental comorbidity. The aims of this population-based study were to describe the epidemiology, genetics, mortality, seizure burden, and treatments in Swedish children with DS (Paper I), to describe seizure-provoking factors and strategies to avoid seizures (Paper II), cognitive function and adaptive behaviour (Paper III), and behaviour, sleep- problems and quality of life (Paper IV).

We identified 55 children born 2000–2018. A semi-structured interview regarding seizure burden, treatments, seizure-provoking factors, and strategies to avoid seizures was used (Papers I and II). The Vineland Adaptive Behaviour scales and Wechsler or Griffiths scales were used to assess adaptive behaviour and intellectual function (Paper III). In paper IV we used the Developmental Behaviour Checklist, the Insomnia Severity Index, and a global question regarding the child´s quality of life.

Of 53 children, 51 (96%) had a variant in the *SCN1A*-gene. Seven (13%) had died. When comparing `younger´ children (born 2010-2018) with `older´ (born 2000-2009), median age at diagnosis was lower, the use of contra-indicated sodium channel inhibitors was lower, and the cumulative incidence was higher in `younger´ children. Status epilepticus was seen in 90%, tonic seizures in 60%, focal to bilateral tonic clonic seizures in 98% and myoclonic seizures in 83%. (Paper I). Seizure precipitants and strategies to prevent seizures were reported in all children. Most common were febrile and afebrile infections (100%, 93%). Most avoided were warm weather (83%) and physical activity (64%) (Paper II). Intellectual Disability was seen in 86% of the children and 93% had difficulties with adaptive behaviour (Paper III). Problems with behaviour were seen in 72%, `moderate' or 'severe' insomnia in 43% and `poor´ or `very poor´ quality of life in 17% of children (Paper IV).

This is one of the first population-based studies that describes psychosocial aspects of DS. We confirm the high mortality and high seizure burden. Results suggest increased knowledge of DS in Sweden, leading to earlier diagnosis and improved treatment. The high number of seizure provoking factors and measures taken to avoid seizures, the high proportion of ID, behavioural and sleep problems can significantly affect the child and the whole family.

Keywords: Anticonvulsants, behaviour, children, Dravet syndrome, demography, intellectual disability, mortality, prevention, precipitants, quality of life, sleep.

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