Phenotyping behaviour and development in Dravet Syndrome

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In this study, we aim to describe cognition and behaviour in patients with DS and analyse correlations between behavioural difficulties and various characteristics including: seizure severity; medication use; level of cognitive functioning; and...

Ethical review	Approved WMO
Status	Completed
Health condition type	Neurological disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON53592

Source
ToetsingOnline

Brief title
Phenotyping behaviour in Dravet syndrome

Condition

- Neurological disorders congenital
- Developmental disorders NEC

Synonym
mutation in the SCN1A gene, the Dravet syndrome

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht
Source(s) of monetary or material Support: Epilepsiefonds;JANIVO stichting;K.F. Hein Fonds;UMC Utrecht

Intervention

Keyword: Behaviour, Development, Dravet syndrome, Psychiatric

Outcome measures

Primary outcome

1) First, we aim to explore changes in behavioural measures (as measured by the CBCL/ABCL) over time in children with DS.

2) Second, we will study parental empowerment and quality of life (QoL) of parents caring for a child with DS. We will explore correlations between QoL and clinical measures.

3) Third, we aim to explore the psychiatric phenotype of SCN1A-related Dravet syndrome in depth and to explore correlations between behavioural measures and clinical characteristics including epilepsy severity, medication use, cognitive development, speech and language problems, sleep difficulties, nutritional problems, early development, HRQoL and effects of SCN1A gene mutation. We aim to identify clinical characteristics that are associated with more severe behavioural disturbances (risk factors) and protective factors, to facilitate early recognition and intervention. Ultimately, this will support parents in the management of their child*s health and behaviour.

Main study parameters are the average scores on behavioural and cognitive measures, including Child and adult behaviour checklist (CBCL/ABCL), Adaptive Behavior Assessment System (ABAS), Intelligence and Development Scales (IDS), Autism Diagnostic Observation Schedule (ADOS), and Family empowerment scale (FES); and main endpoints are correlations between behavioural difficulties and

clinical characteristics such as seizure severity, medication use and level of

cognitive functioning.

Secondary outcome

not applicable

Study description

Background summary

Dravet Syndrome(DS) consists of a wide phenotypic spectrum of seizures (e.g., febrile seizures, absences, status epilepticus) and is associated with behavioural difficulties. The prevalence of behavioural difficulties in DS ranges between 37% and 100% (1-4). However, the nature and extent of behavioural difficulties, i.e., the *psychiatric phenotype* is unclear. We aim to identify patient and clinical characteristics associated with more severe behavioural problems; this may facilitate early recognition and intervention.

Study objective

In this study, we aim to describe cognition and behaviour in patients with DS and analyse correlations between behavioural difficulties and various characteristics including: seizure severity; medication use; level of cognitive functioning; and quality of life (QoL). We aim to explore factors associated with more behavioural disturbances. Also, we will examine the parental empowerment and the wellbeing of parents and patients with DS

Study design

This is a prospective observational study with a cross-sectional detailed psychiatric assessment and a repeated behavioural measure.

Study burden and risks

All questionnaires will be completed online by caregivers. This means that they can do this in their own time and pace, from their own homes. We will carry out one psychiatric assessment and aim to do this during one home visit. This minimizes the burden on the patients and their families and has the added advantage of minimizing potential assessment related anxiety and stress. There are no risks associated with this study. As for group relatedness, to study behavioural aspects of DS, we will need to study patients with DS.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)

Inclusion criteria

• Subjects are diagnosed with Dravet syndrome (genetically confirmed SCN1A mutation)
• Subjects are living in the Netherlands
• Subjects speak the Dutch language
• Age 2 years and older

Exclusion criteria

• Patients with a variant of unknown significance (class III) in the SCN1A gene

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Completed
Start date (anticipated):	21-05-2024
Enrollment:	150
Type:	Actual

Ethics review

Approved WMO

Date:	28-04-2022
Application type:	First submission
Review commission:	METC NedMec

Approved WMO

Date:	08-02-2023
Application type:	Amendment
Review commission:	METC NedMec

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL80056.041.22