The IBIS study: Infant motor development as an early Biomarker in children with SCN1A gene mutation - a pilot project

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Objective: The primary objective of this pilot project is to investigate the early course of motor development in children with DS. Secondary objectives are to investigate the relation between type of SCN1A mutation and developmental profile and to...

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

Summary

ID

NL-OMON51547

Source
ToetsingOnline

Brief title
The IBIS study

Condition

- Neurological disorders congenital
- Seizures (incl subtypes)

Synonym
developmental delay, Dravet syndrome, Epilepsy

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Subsidie van de Christian society en van de Stichting Dravetsyndroom NL / VL

Intervention

Keyword: Dravet syndrome, Early biomarker, Infant Motor Profile (IMP), SCN1A gene mutattion

Outcome measures

Primary outcome

Main study parameters/endpoints: Primary assessment tool is the Infant Motor

Profile (IMP), a qualitative method to assess motor development in infancy.

Secondary outcome

Secondary parameters are the scores on the Bayley Scales of Infant and Toddler

Development, third edition (Bayley-III-NL) and the genetic data on the specific

SCN1A mutation type.

Study description

Background summary

Rationale: Dravet syndrome (DS) is a severe, disabling epileptic encephalopathy with core symptoms intractable epilepsy, profound developmental delay and behavioural difficulties. In at least 80% of the cases DS is caused by a mutation in the SCN1A gene. A substantial number of children with an SCN1A mutation, however, do not develop DS, but a less severe phenotype such as genetic epilepsy with febrile seizures plus (GEFS+) or only febrile seizures (FS). As the phenotypic diagnosis of DS relies on the presence of developmental delay and this takes time to become apparent, there may be a long diagnostic gap which causes severe burden and uncertainty for parents. To allow adequate parents* counselling it is therefore crucial to find early prognostic biomarkers for developing DS. Also, early diagnosis is crucial in the light of the newly emerging gene-specific therapy approaches such as antisense oligonucleotide modulation. Little is known about early motor development in DS

syndrome and whether qualitative assessment of motor behaviour can serve as an early biomarker for DS.

Study objective

Objective: The primary objective of this pilot project is to investigate the early course of motor development in children with DS. Secondary objectives are to investigate the relation between type of SCN1A mutation and developmental profile and to assess which domains of motor development are affected.

Study design

Study design: Prospective longitudinal study with repeated longitudinal assessments, inclusion of infants between ages 6 months and 2 years, follow-up for one year with assessments every 3 months.

Study burden and risks

Nature and extent of the burden and risks associated with participation, benefit and group relatedness: The study is not associated with risks for the child or the parents. If the child is tired, crying or hungry, the assessment will be stopped. Costs of the study are an investment of time. As the assessments are for the large part performed by means of home visits there is no burden of travel time and effort for the parents and the child involved.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Children (2-11 years)
Babies and toddlers (28 days-23 months)

Inclusion criteria

- Age 2 years of less at time of inclusion
- Presented with seizures in the first two years of life and have an (likely) pathogenic SCN1A gene variant
- Caregivers have sufficient understanding of the Dutch or English language to give informed consent

Exclusion criteria

- Caregivers having insufficient understanding of the Dutch or English language
- Diagnosis of cerebral palsy (CP) or other neuromotor condition affecting motor development in addition to the SCN1A mutation

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-05-2023

Enrollment:	24
Type:	Actual

Ethics review

Approved WMO Date:	05-08-2022
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

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	NL81376.042.22
Other	UMCG research register: 202200315