DNA onderzoek om symptomen bij epilepsie of koortsstuipen te kunnen voorspellen

No registrations found.

Ethical review	Positive opinion
Status	Pending
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON27611

Source
Nationaal Trial Register

Health condition

Dravet syndrome; GEFS+; epileptic encephalopathy; Dravet syndroom; seizures; koortsstuipen

Sponsors and support

Primary sponsor: University Medical Center Utrecht
Source(s) of monetary or material Support: Vrienden van ht WKZ

Intervention

Outcome measures

Primary outcome

Classification of developmental outcome, rated independently by a child neurologist, neuropsychologist, and clinical geneticist

Secondary outcome

-Intelligence quotient

-Epilepsy syndrome classification

-Mobility

-Quality of life

-Behavioural difficulties

Study description

Background summary

Mutations in the SCN1A gene have been shown to cause a wide spectrum of neurological symptoms, ranging from isolated febrile seizures, to severe myoclonic epilepsy in infancy (SMEI), also known as Dravet syndrome. Dravet syndrome is a severe neurological disorder of childhood, usually presenting in the first year of life with generalized or unilateral clonic seizures. Psychomotor development is initially normal, but slows down in the second year of life. Outcome is usually poor and patients develop intractable epilepsy and mental retardation. In around 75% of the cases, a mutation in the SCN1A gene is found, which occurs de novo in most patients. However, a clear genotype-phenotype relation has not been established yet, and patients with the same mutations may show very different phenotypes ranging from mild to profound disability in the patient.
The overall goal of this study is to establish if early genetic screening on SCN1A mutations in infants would be feasible. Prerequisites of such a screening test would be that the clinical outcome of a child with a mutation can be predicted accurately, and that early diagnosis benefits the patient and improves the course of disease.
The specific aim of this study is to assess if clinical outcomes of a patient with a pathogenic SCN1A mutation can be predicted based on advanced genotyping. Therefore, we will subsequently investigate the association between somatic mosaicism, variants in regions in and around the SCN1A gene, and mutations in modifier genes on the one hand, and clinical outcomes of patients with SCN1A related febrile seizures/epilepsy on the other hand. Furthermore, we will evaluate if patients who were diagnosed with SCN1A related febrile seizures/epilepsy at an early age have better clinical outcomes than children who were diagnosed at a later age.
If we can predict clinical course of the disease based on an early genetic diagnostics, we can give physicians and parents more accurate information about the prognosis, which is of importance for medical treatment and coping. In addition, if we could demonstrate that children with an early diagnosis have better clinical outcomes than children with a later diagnosis, early genetic testing might be considered in children with febrile seizures before the age of 12 months or even as part of a neonatal screening program.

Study design

Date of first enrollment: 1-3-2015

Intervention

-

Contacts

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Eligibility criteria

Inclusion criteria

-patients with SCN1A related epilepsy/febrile seizures and their parents

-living in the Netherlands

-informed consent form signed

Exclusion criteria

-patients with a variant of unknown significance (class III) in the SCN1A gene

Study design

Design

Study type: Observational non invasive

Intervention model:	Other

Control: N/A , unknown

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-02-2015
Enrollment:	200
Type:	Anticipated

Ethics review

Positive opinion

Date:	06-01-2015
Application type:	First submission

Study registrations

Followed up by the following (possibly more current) registration

ID: 42281
Bron: ToetsingOnline
Titel:

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

No registrations found.

In other registers

Register	ID
NTR-new	NL4927
NTR-old	NTR5029
CCMO	NL50984.041.14
OMON	NL-OMON42281

Study results