DNA onderzoek om symptomen bij epilepsie of koortsstuipen te kunnen voorspellen

Gepubliceerd: 06-01-2015 Laatst bijgewerkt: 15-05-2024

Ethische beoordeling	Positief advies
Status	Werving nog niet gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON27611

Bron
NTR

Aandoening

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

Ondersteuning

Primaire sponsor: University Medical Center Utrecht
Overige ondersteuning: Vrienden van ht WKZ

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

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

Toelichting onderzoek

Achtergrond van het onderzoek

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.

Onderzoeksopzet

Date of first enrollment: 1-3-2015

Onderzoeksproduct en/of interventie

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Contactpersonen

Publiek

R.S. Boerma
Utrecht
The Netherlands

Wetenschappelijk

R.S. Boerma
Utrecht
The Netherlands

Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

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

-living in the Netherlands

-informed consent form signed

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

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

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Anders

Controle: N.v.t. / onbekend

Deelname

Nederland

Status:	Werving nog niet gestart
(Verwachte) startdatum:	01-02-2015
Aantal proefpersonen:	200
Type:	Verwachte startdatum

Ethische beoordeling

Positief advies

Datum:	06-01-2015
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

ID: 42281
Bron: ToetsingOnline
Titel:

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

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

Resultaten