Cognitie en het ontwikkelende brein in galactosemie.

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

Samenvatting

ID

NL-OMON22209

Bron
NTR

Aandoening

Classic galactosemia (Klassieke galactosemie)

Ondersteuning

Primaire sponsor: Dr. M.E. Rubio-Gozalbo, academisch Ziekenhuis Maastricht
Overige ondersteuning: Galactosemie Onderzoek Fonds (GOF)

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

1. The performance on the language-related behavioural paradigms (four paradigms, each focusing on one aspect of speech processing or production);
2. The event-related potentials (ERPs) extracted from the EEG recorded during the behavioural tasks and.

Toelichting onderzoek

Achtergrond van het onderzoek

Rationale:

Classic galactosemia is a hereditary disorder caused by the body's inability to breakdown galactose, a sugar mainly found in milk. A galactose-restricted diet (soy diet) is the basis of therapy. This, however, does not prevent the emergence of long-term complications. One of these long-term complications is related to cognition. Reduced intelligence quotient (IQ) scores as well as impairments in speech and language have been reported. In fact, ninety percent of affected children have speech problems, usually diagnosed as Childhood Apraxia of Speech (CAS) (i.e. an impairment of motor programming of speech musculature). However, at present, CAS is over-diagnosed. This means that there are patients that receive a treatment for CAS, while other speech disabilities are being overlooked and not addressed properly resulting in suboptimal treatment. Our preliminary research concerning speech shows that syntax is a major problem in these children. The innovative approach in this study, examining the different levels of speech (e.g. semantic and syntactical speech) in combination with behavioral data and simultaneous brain activity recordings, aims to elucidate which levels of speech are specifically impaired in these children. Pinpointing at which level speech and other cognitive functions are affected is imperative to design successful treatment approaches with a better outcome.

Objectives:

To elucidate which areas of speech are specifically affected in children and adolescents with classic galactosemia; and to sketch a profile of the galactosemic patients' general cognitive functions and of the development of these cognitive functions.

Study design:

Observational case control design consisting of a neuropsychological assessment and a behavioural paradigm with simultaneous electroencephalographic brain activity recording.

Doel van het onderzoek

Specific areas of language production are affected in children and adolescents with classic galactosemia, in particular syntactic production. Further, it is hypothesized that the galactosmic patients will show a profile of general cognitive functions deviating from typically developing children and adolescents.

Onderzoeksopzet

Two sessions will be scheduled: One for the neuropsychological test and one for the EEG study.

Onderzoeksproduct en/of interventie

N/A

Contactpersonen

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

For patients, a diagnosis of classic galactosemia as diagnosed by GALT enzyme activity assay or GALT-gene mutation analysis (information obtained from treating physician); Age between 10 and 18 years old.

Belangrijkste redenen om niet deel te kunnen nemen

(Exclusiecriteria)

Any other disorder or disease that could affect cognitive functioning independently of classic galactosemia (an exception is made for the diagnosis of ADHD, because this disorder is common in this group).

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Factorieel
Toewijzing:	Niet-gerandomiseerd
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestopt
(Verwachte) startdatum:	01-08-2009
Aantal proefpersonen:	52
Type:	Werkelijke startdatum

Ethische beoordeling

Positief advies

Datum:	26-04-2011
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

ID: 32979

Bron: ToetsingOnline
Titel:

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL2731
NTR-old	NTR2869
CCMO	NL27398.068.09
ISRCTN	ISRCTN wordt niet meer aangevraagd.
OMON	NL-OMON32979

Resultaten

Samenvatting resultaten
N/A