Cognition in classic galactosemia: What happens to the developing brain?

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Inborn errors of metabolism
Study type	Observational non invasive

Summary

ID

NL-OMON32979

Source
ToetsingOnline

Brief title
Cognition and the developing brain in galactosemia

Condition

- Inborn errors of metabolism

Synonym
Classic Galactosemia; inherited metabolic disorder

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Ziekenhuis
Source(s) of monetary or material Support: Galactosemie Onderzoek Fonds (GOF)

Intervention

Keyword: Classic galactosemia, Cognition, ERP, Inherited metabolic disorders

Outcome measures

Primary outcome

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

Secondary outcome

The secundary parameter is the performance on the neuropsychological testbattery (i.e. a composition of tests focusing on the major cognitive domains: memory, attention, speech and language, visuo-motor skills).

Study description

Background summary

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 (i.e. grammar and construction of sentences) 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.

Study objective

In this study we aim 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.

Study design

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

Study burden and risks

The participants will be asked to visit the Maastricht University Medical Centre twice (i.e. each visit lasting about two hours). They are expected to perform several neuropsychological tasks as well as to participate in an EEG recording. The EEG recording might be experienced as slightly unpleasant at the most, although there are no adverse reactions expected.

This study will be conducted with minors being the subjects, instead of adults. The goal of this study is namely to gain more knowledge about the cognitive abilities of a group of young patients with classic galactosemia. The examination of the development of these abilities is therein an important and indispensable part. An adequate description of the development of cognitive functions is necessary for multiple reasons [see also D8]. First, there is an ongoing debate as to whether intellectual capacities regress with age. By taking a broader approach and studying besides intellectual capacities other cognitive functions as well, we can give a significant contribution solving this question. Second, virtually all children and adolescents with galactosemia receive therapy and/or special education because of their cognitive impairments. A characterization of these impairments and furthermore of the development of these impairments is therefore indispensable and will contribute to a (more) optimal treatment. Finally, the fact that children and adolescents still have the possibility to be treated for their specific problems, because many functions are still in development, is essential to report as well.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

A diagnosis of classic galactosemia as assessed by GALT enzyme activity assay or GALT-gene mutation analysis (information obtained from treating physician)
Age between 10 and 18 years old

Exclusion criteria

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

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	13-01-2010
Enrollment:	52
Type:	Actual

Ethics review

Approved WMO

Date:	17-07-2009
Application type:	First submission
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

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

ID: 22209
Source: Nationaal Trial Register
Title:

In other registers

Register	ID
CCMO	NL27398.068.09
OMON	NL-OMON22209