Neurocognitive functioning in school-aged cystinosis patients: general effects of chronic disease or specific deficits?

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The aim of the study is to improve the quality of life of cystinosis patients. Part of it forms neuropsychological screening in order to identify specific defects in the neurocognitive development and learning abilities. The results will contribute...

Ethical review	Approved WMO
Status	Pending
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational non invasive

Summary

ID

NL-OMON30443

Source
ToetsingOnline

Brief title
Neurocognitive functioning in school-aged cystinosis patients

Condition

- Congenital and hereditary disorders NEC
- Electrolyte and fluid balance conditions
- Nephropathies

Synonym
cystine accumulation disease

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Sint Radboud

Source(s) of monetary or material Support: Nierstichting Nederland

Intervention

Keyword: cystinosis, learning problems, neurocognitive functioning, school-age

Outcome measures

Primary outcome

-total intelligence, verbal and performal intelligence

-neurocognitive functioning

-perceptual (visual)-motor functioning

-behavioural- and schoolproblems

Secondary outcome

-quality of life

-subjective parental stress

Study description

Background summary

According to a limited number of studies patients with cystinosis are assumed to be normal intelligent (Wolff et al.1982; Williams et al.1994, Colah et al.1997; Ballantyne et al.1997). However neuropsychological research performed by the latter investigators, all participants in the group of Doris Trauner, San Diego, showed problems in visual- and tactile information processing (Colah et al., visual-spatial functioning (Williams et al.) resulting into impaired school performance e.g. spelling and arithmatic (Ballantyne et al.) Our pilot study showed disturbances in fine-motor functioning in 6 out of 9 children. Williams et al. hypothesized a relation between these "minor symptoms" and a neurotoxic effect of cystine accumulation in the brain. Moreover it has to be referred to global effects of chronic renal failure, side effects of medication an the psychosocial impact of chronic ilness in general. Recent literature mentionede late adverse effects of renal disease and treatment on cognitive functioning in general and a-specific neurocognitive defects: attention, memory, visual-motor functioning, but also verbal abilities (Groothoff et al. 2002; Gipson et al. 2004). A rather high percentage of children from our

patient group has been referred because of school problems. Neuropsychological investigation showed a specific neurocognitive profile. Learning problems resulting from specific defects have not or insufficiently been recognized by parents and teachers, because the child is assessed as normal intelligent or even "smart". Identification of "minor"neurological symptoms,visual and perceptual disorders and schoolproblems by neuropsychological screening in all school-aged cystinosis patients is strongly recommended.

Study objective

The aim of the study is to improve the quality of life of cystinosis patients. Part of it forms neuropsychological screening in order to identify specific defects in the neurocognitive development and learning abilities. The results will contribute to an early recognition of specific problems, information and advices to parents and teachers.

Study design

Neuropsychological screening of all school-aged cystinosis patients treated in the Netherlands (age 6-16 yrs., n=20)
-cognitive functioning WISC-lll, verbal and performal intelligence
-neurocognitive functioning (ANT,Stroop,Bourdon)
-perceptual (visual)- motor functioning (VMI and experimental drawing- and writing tasks by an electronic digitizer identifying an analyzing pressure, tremors, fluency, speed and pauses
-behavioural- and school problems (CBCL (parents)and TRF (teachers)

The tests , as well as reports are executed by a psychological assistant, supervised by a clinical psychologist. The junior investicator/psychologist will analyze the data writing the manuscript. The results will be discussed personally and reported by written with the patient and the parents and if indicated (after having obtained permission) information and advice will be provided to the school.
See enclosed appendix

Study burden and risks

see under section E2

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

-treated for cystinosis in a Dutch UMC
-school-aged (6-18 years)

Exclusion criteria

-mental retardation (not being able to perform tests)
-medical or psychiatric co-morbidity
-insufficient knowledge of the Dutch language

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-04-2007
Enrollment:	24
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL14646.091.07