Diagnostics and diagnosis oriented research of mental retardation of unknown cause

Published: 12-01-2009
Last updated: 08-05-2024

The main objective of the study is to reach an etiological diagnosis in as many cases of a selected group of MR patients. The aim is to do so in at least 15-20% of hitherto unexplained MR.Also an objective is the establishment of a genetic-...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON30801

Source
ToetsingOnline

Brief title
Diagnostics of mental retardation of unknown cause

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Mental impairment disorders

Synonym
mental handicap, mental impairment

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Sint Radboud
Source(s) of monetary or material Support: Ministerie van OC&W,de 3 betrokken

instellingen voor VG-zorg

Intervention

Keyword: diagnostics, mental retardation, mental retardation research, MR genes

Outcome measures

Primary outcome

The primary outcome of the study is having a etiological diagnosis and

establishing a genetic-diagnostic protocol to be used in the MR sector by the

AVG.

Secondary outcome

Finding new genes and new MR syndromes are secundary outcome measures as are

the genotype/phenotype correlation studies

Study description

Background summary

Mental retardation is definied as an IQ < 70, significant limitation in adaptive functioning and onset before the age of 18 years. The prevalence is estimated to be 2-3%. Severe MR has a prevalence of about 0,5%, while mild MR (IQ 50-70) is more common. There is a male excess of about 40%, partially due to monogeneic X-linked MR. The societal and psychosocial buden of MR is high. The percentage health budget spent on MR is the highest among the different disease categories. Causes of MR are multiple and very often genetic; well known examples are Down syndrome and fragile X syndrome. However, in the majority of cases the etiology is unknown. The importance of having an etiological diagnosis is that it shed light on the prognosis and the related comorbidity and is therefore contributing to the management plan. Also it is a prerequisite for a reliable genetic counseling to healthy relatives of MR patients.

Study objective

The main objective of the study is to reach an etiological diagnosis in as many cases of a selected group of MR patients. The aim is to do so in at least

15-20% of hitherto unexplained MR.
Also an objective is the establishment of a genetic-diagnostic protocol, to be used in the MR sector by the physicians in the care of the mentally handicapped (AVG).
Besides it is expected to find:
- one or more new MR syndromes
- one or more new MR genes
- genotype-phenotype correlations

Study design

In the 3 involved institutions about 8000 MR patients are known. Based on the inclusion and exclusion criteria about 400 cases will be selected by the AVG in consultation with the research team. These 400 cases will be physically examined and will have their blood sampled. Also their parents will be sampled in order to differentiate between polymorphisms and causal defects. The physical examination is primarily meant to delineate carefully the dysmorphological phenotype. The blood sample will be used for the identification of genetic defects (chromosomal defects, monogenetic defects and genomic defects).
A MR clinic will be held twice a month in the UMC St Radboud, in which the PhD student, clinical geneticists and on a consultative basis also a child neurologist and paediatrician for metabolic diseases are participating.

Study burden and risks

The burden of the study is having good preparation and selection, low. The same holds true for risks.

Contacts

Public
Universitair Medisch Centrum Sint Radboud

Postbus 9101
6500 HB Nijmegen
NL
Scientific
Universitair Medisch Centrum Sint Radboud

Postbus 9101
6500 HB Nijmegen
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

mental retardation of unknown origin
availability of both parents
additionnal features (e.g congenital malformations, dysmorphology, consanguinity, micro/macrocephaly, abnormal growth)

Exclusion criteria

mental retardation of known cause

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	14-07-2009
Enrollment:	400
Type:	Actual

Ethics review

Approved WMO

Date:	12-01-2009
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	NL13636.091.07