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Alcohol supplementation in rhizomelic chondrodysplasia punctata in the Netherlands

Submission date	Recruitment status	[ ] Prospectively registered
22/11/2006	No longer recruiting	[ ] Protocol
Registration date	**Overall study status**	[ ] Statistical analysis plan
22/11/2006	Completed	[ ] Results
Last Edited	**Condition category**	[ ] Individual participant data
23/09/2021	Musculoskeletal Diseases	[ ] Record updated in last year

Plain English summary of protocol
Not provided at time of registration

Contact information

Type(s)
Scientific

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Additional identifiers

EudraCT/CTIS number

IRAS number

ClinicalTrials.gov number

Secondary identifying numbers
NL736 (NTR746)

Study information

Scientific Title
Alcohol supplementation in rhizomelic chondrodysplasia punctata in the Netherlands

Study objectives
Plasmalogens can be synthesised out of batyl alcohol (naturally occuring alkylglycerol) in patients with the peroxisomal disorder Rhizomelic Chondro-Dypslasia Punctata (RCDP), bypassing the peroxisomal steps in the pathway.

Ethics approval required
Old ethics approval format

Ethics approval(s)
Ethics approval received from the local medical ethics committee

Study design
Cohort study

Primary study design
Observational

Secondary study design
Cohort study

Study setting(s)
Not specified

Study type(s)
Screening

Participant information sheet

Health condition(s) or problem(s) studied
Rhizomelic chondrodysplasia punctata

Interventions
Batyl alcohol supplementation 5 to 50 mg/kg/day.

The following steps will be taken:
1. Blood sampling
2. X-ray skeleton
3. Dexa scan
4. Magnetic Resonance Imaging (MRI)
5. ElectroEncephaloGram (EEG)
6. Visual Evoked Potential (VEP)
7. Brainstem Auditory Evoked Potentials (BAEP)
8. ElectroMyoGraphy (EMG)
9. SomatoSensory Evoked Potentials (SSEP)
10. Questionnaire on well-being

Intervention Type
Drug

Phase
Not Specified

Drug/device/biological/vaccine name(s)
Batyl alcohol

Primary outcome measure
Plasmalogen content in erythrocytes increases significantly in both severe and milder patients with RCDP.

Secondary outcome measures
1. Increase in plasmalogens in sputum
2. Improving quality of life scores (TNO-AZL Preschool children Quality of Life [TAPQOL])
3. Stabilisation or improvement in nerve conduction

Stabilisation in MRI/MRS will be our tertiary endpoint.

Overall study start date
01/01/2006

Completion date
01/01/2008

Eligibility

Key inclusion criteria
1. Parents or legal representatives must have given written informed consent
2. Patients must have a current diagnosis of RCDP established by biochemical analysis and/or mutation analysis
3. Parents of patients must be willing to fulfil the evaluation program

Participant type(s)
Patient

Age group
Not Specified

Sex
Not Specified

Target number of participants
10

Key exclusion criteria
1. Parents/legal representatives are unwilling to fulfil the evaluation program
2. Intolerability of the drug

3. Concomitant severe disease resulting in very short life expectancy
4. Decision by the patient and/or his/her parents or legal representatives to withdraw from the treatment

Date of first enrolment
01/01/2006

Date of final enrolment
01/01/2008

Locations

Countries of recruitment
Netherlands

Study participating centre
Academic Medical Center
Amsterdam
Netherlands
1105 AZ

Sponsor information

Organisation
Academic Medical Center (AMC) (The Netherlands)

Sponsor details
Department of Pediatrics
P.O. Box 22660
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1100 DD

Sponsor type
Hospital/treatment centre

ROR
https://ror.org/03t4gr691

Funder(s)

Funder type
Not defined

Funder Name
Not provided at time of registration

Results and Publications

Publication and dissemination plan
Not provided at time of registration

Intention to publish date

Individual participant data (IPD) sharing plan

IPD sharing plan summary
Not provided at time of registration