

Alcohol supplementation in rhizomelic chondrodysplasia punctata in the Netherlands

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

Plain English summary of protocol
Not provided at time of registration

Contact information

Type(s)
Scientific

Contact name
Dr A M Bams-Mengerink

Contact details
Academic Medical Center
Department of Pediatrics
H8-141
Meibergdreef 9
Amsterdam
Netherlands
1105 AZ
+31 (0)20 5667508
a.m.mengerink@amc.uva.nl

Additional identifiers

Protocol serial number
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 occurring alkylglycerol) in patients with the peroxisomal disorder Rhizomelic Chondro-Dysplasia 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

Study type(s)

Screening

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(s)

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

Key secondary outcome(s)

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.

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

Healthy volunteers allowed

No

Age group

Not Specified

Sex

Not Specified

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)

ROR
<https://ror.org/03t4gr691>

Funder(s)

Funder type
Not defined

Funder Name
Not provided at time of registration

Results and Publications

Individual participant data (IPD) sharing plan

IPD sharing plan summary
Not provided at time of registration