ISRCTN44820021 https://doi.org/10.1186/ISRCTN44820021

# 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

**Contact name**
Dr A M Bams-Mengerink

**Contact details**
Academic Medical Center
Department of Pediatry
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 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

## 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