

# Alcohol supplementation in rhizomelic chondrodysplasia punctata in the Netherlands

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		<input type="checkbox"/> Protocol
<b>Registration date</b> 22/11/2006	<b>Overall study status</b> Completed	<input type="checkbox"/> Statistical analysis plan
		<input type="checkbox"/> Results
<b>Last Edited</b> 23/09/2021	<b>Condition category</b> Musculoskeletal Diseases	<input type="checkbox"/> Individual participant data
		<input type="checkbox"/> 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 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

## 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

Amsterdam

Netherlands

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