Alcohol supplementation in Rhizomelic chondrodysplasia punctata in the Netherlands.

No registrations found.

Ethical review	Positive opinion
Status	Recruiting
Health condition type	-
Study type	Interventional

Summary

ID

NL-OMON27819

Source
Nationaal Trial Register

Brief title
N/A

Health condition

patients with rhizomelic chondroysplasia punctata, severe and milder phenotypes.

Sponsors and support

Source(s) of monetary or material Support: European Leukodystrophy Association

Intervention

Outcome measures

Primary outcome

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

Secondary outcome

Secondary endpoints include

1. increase in plasmalogens in sputum
2. improving quality of life scores (TAPQOL)

3. stabilization or improvement in nerve conduction. Stabilization in MRI/MRS will be our tertiary endpoint.

Study description

Background summary

Rationale: Plasmalogen deficiency is the main biochemical defect in patients with Rhizomelic chondrodysplasia punctata (RCDP); a peroxisomal disorder characterized by skeletal abnormalities and severe neurological (both mental and motor) impairments. RCDP patients can have either a mild or a severe phenotype depending on the rest-capacity to synthesize plasmalogens. Most patients with the severe phenotype of RCDP die in their first decade of life. Currently there is no therapy available. Batyl alcohol is a naturally occurring mono ether glycerol. In vitro studies in human fibroblast cell lines and in vivo studies in the mice model for RCDP and Zellweger syndrome patients showed that plasmalogen levels can be restored by batyl alcohol.
Objective: The central aims of this study are to evaluate whether the plasmalogens in RCDP patients can be increased by batyl alcohol supplementation and to find clinical effects of oral batyl alcohol supplementation.
Study design: Cohort study
Study population: All patients with biochemically proven RCDP can enter this study. The aim is to include 10 patients in a study period of 2 years.
Intervention: Patients will be taking 5 to 50 mg/kg batyl alcohol daily. The batyl alcohol will be administered orally.
Main study parameters/endpoints: the primary endpoints in this study will be an increase in plasmalogen content of erythrocytes to at least 50% of normal C18:0 DMA/C18:0 fatty acid values in patients with the severe phenotype and to at least 90% of normal C18:0 DMA/C18:0 fatty acid values in patients with the milder phenotype of RCDP and an increase in growth and bone development. Secondary endpoints include 1) increased plasmalogen content in sputum 2) improving quality of life scores (TAPQOL) and 3) stabilization or improvement in nerve conduction. Stabilization in MRI/MRS will be our tertiary endpoint.
Nature and extent of the burden and risks associated with participation, benefit and group relatedness: Patients will take batyl alcohol capsules every day. No side-effects of alkylglycerols including batyl alcohol have been described in literature. An extended work-up / follow-up will be at baseline and after 12 months, including a physical examination, blood sampling, urine sampling, electroneurophysiological examinations, skeletal X-rays, joint function tests and MRI/MRS. The MRI/MRS, sputum collection and the blood sampling will be performed during narcosis. For these investigations the child will be admitted to the paediatric ward of the AMC. At 2, 4, 8, 12, 26, 39 weeks the patient will come to the outdoor

clinic for a physical examination and blood and urine sampling. The investigators will try to minimize the burden for patients and their parents by minimizing the hospitalisation time and by performing the invasive studies during the narcosis. Medical X-ray burden is minor.

Study objective

Plasmalogens can be synthesized out of batyl alcohol (naturally occuring alkylglycerol) in patients with the peroxisomal disorder rhizomelic chondrodypslasia punctata (RCDP), bypassing the peroxisomal steps in the pathway.

Intervention

1. batyl alcohol supplementation 5 - 50 mg/kg/day

2. blood sampling

3. X-ray skeleton

4. Dexa scan

5. MRI

6. EEG

7. VEP

8. BAEP

9. EMG

10. SSEP

11. questionnaire well being

Contacts

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Eligibility criteria

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

Exclusion criteria

Parents / legal representatives are unwilling to fulfil the evaluation program. Patients will be excluded from treatment if the following occurs:

1. Intolerability of the drug.

2. Concomitant severe disease resulting in very short life expectancy.

3. Decision by the patient and/or his/her parents or legal representatives to withdraw from the treatment.

Study design

Design

Study type:	Interventional
Intervention model:	Other
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-01-2006
Enrollment:	10
Type:	Anticipated

Ethics review

Positive opinion

Date:	04-08-2006
Application type:	First submission

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
NTR-new	NL736
NTR-old	NTR746

Register	ID
Other	: N/A
ISRCTN	ISRCTN44820021

Study results

Summary results
N/A