Phenylalanine supplementation in Tyrosinemia type 1

No registrations found.

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

Summary

ID

NL-OMON23208

Source
NTR

Health condition

Tyrosinemia type 1
phenylalanine
supplementation
tyrosine

Sponsors and support

Primary sponsor: University Medical Center Groningen
Source(s) of monetary or material Support: Metakids stichting

Intervention

Outcome measures

Primary outcome

Blood phenylalanine concentrations

Blood tyrosine concentrations

Secondary outcome

Blood NTBC concentrations

Blood succinylacetone concentrations

Study description

Background summary

Rationale:Hereditary Tyrosinemia Type 1 (HT1) patients usually present with liver dysfunction and or renal tubular dysfunction with rickets early in life. After the introduction of 2-(2-nitro-4-trifluoro-methylbenzoyl)-1,3-cyclohexanedione (NTBC), problems resolved and life expectancy greatly increased. However, due to NTBC, which blocks the conversion of tyrosine at an earlier step, tyrosine concentrations increase, making dietary restrictment of tyrosine and its precursor phenylalanine necessary. Recently, some articles report low phenylalanine concentrations in HT1, making phenylalanine supplementation necessary despite possible conversion to tyrosine and resulting increase in tyrosine. Next to that, previous research of us showed a strong diurnal variation of phenylalanine, with extremely low phenylalanine concentrations early in the afternoon. These strong diurnal variation and increase in tyrosine concentrations make more studies on the optimal dose of phenylalanine supplementation necessary.

Objective: The main objective of this study is to investigate the effect of phenylalanine supplementation on phenylalanine and tyrosine concentrations.

Study population: We estimate that 15 HT1 patients will be included in this study all treated with NTBC, a tyrosine and phenylalanine restricted diet and phenylalanine suppletion.

Main study parameters: phenylalanine, tyrosine, NTBC and succinylacetone concentrations during the day at different doses of phenylalanine supplementation in HT1 patients.

Study design: Patients will be treated with 2 different amounts of phenylalanine supplementation. The participating HT1 patients will perform 2 rounds of 13 bloodspots (taken by finger prick). Next to this 8 bloodspots are done to set a baseline without supplementation. The total duration of the study is 24 days.

Study objective

Phenylalanine supplementation can result in higher phenylalanine concentrations without causes tyrosine concentrations to rise too much

Study design

Patients from the University Medical Center Groningen, the Netherlands will be included first. Afterwards, patients from the Birmingham Children's Hospital, UK will be included in this study.

Intervention

Different doses of phenylalanine supplementation will be given during some days. The effect of the supplementation on metabolic control will be studied while receiving the different dosages.

Contacts

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Eligibility criteria

Inclusion criteria

- Diagnosed with Tyrosinemia type 1

- Treated with NTBC

- Adequate dietary control (tyrosine concentrations: 200-600 µmol/L)

Exclusion criteria

- Tyrosinemia type 1 patients who received liver transplantation

Study design

Design

Study type:	Interventional
Intervention model:	Crossover
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-02-2017
Enrollment:	15
Type:	Anticipated

Ethics review

Positive opinion

Date:	08-06-2018
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	NL7205

Register	ID
NTR-old	NTR7404
Other	: METc 2016/296

Study results

Summary results
not applicable yet