Phenylalanine supplementation in Tyrosinemia type 1

Gepubliceerd: 08-06-2018 Laatst bijgewerkt: 18-08-2022

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

Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Interventie onderzoek

Samenvatting

ID

NL-OMON23208

Bron
NTR

Aandoening

Tyrosinemia type 1
phenylalanine
supplementation
tyrosine

Ondersteuning

Primaire sponsor: University Medical Center Groningen
Overige ondersteuning: Metakids stichting

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

Blood phenylalanine concentrations
Blood tyrosine concentrations

Toelichting onderzoek

Achtergrond van het onderzoek

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.

Doel van het onderzoek

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

Onderzoeksopzet

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.

Onderzoeksproduct en/of interventie

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.

Contactpersonen

Publiek

F.J. van Spronsen
Groningen
The Netherlands

Wetenschappelijk

F.J. van Spronsen
Groningen
The Netherlands

Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

- Diagnosed with Tyrosinemia type 1

- Treated with NTBC

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

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

- Tyrosinemia type 1 patients who received liver transplantation

Onderzoeksopzet

Opzet

Type:	Interventie onderzoek
Onderzoeksmodel:	Cross-over
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	01-02-2017
Aantal proefpersonen:	15
Type:	Verwachte startdatum

Ethische beoordeling

Positief advies

Datum:	08-06-2018
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

Geen registraties gevonden.

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL7205

Register	ID
NTR-old	NTR7404
Ander register	: METc 2016/296

Resultaten

Samenvatting resultaten
not applicable yet