Efficacy and safety of enzyme replacement therapy for MPS I with 100 I.U./Kg recombinant human a-L-iduronidase (ALDURAZYME™).

Gepubliceerd: 12-09-2005 Laatst bijgewerkt: 13-12-2022

MPS I patients can be treated with Aldurazyme safely and effectively.

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

Samenvatting

ID

NL-OMON21663

Bron
NTR

Verkorte titel
N/A

Aandoening

Mucopolysaccharidose type I.

Ondersteuning

Primaire sponsor: CVZ
Overige ondersteuning: N/A

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

1. Improvement of joint mobility;
2. Improvement of quality of life.

Toelichting onderzoek

Achtergrond van het onderzoek

Mucopolysaccharidose type I (MPS I) is caused by the deficiency of the lysosomal enzyme a-L-Iduronidase.

Due to this deficiency heparan sulphate and dermatan sulphate (glycosaminoglycans, GAGs) accumulate in the lysosomes of all cells, but predominantly in the connective tissue. Clinical features encompass a spectrum of disease manifestations.

Three phenotypes are recognized;

1. the neuronopathic (Hurler) type at one end of the spectrum;

2. an intermediate (Hurler-Scheie) phenotype;

3. and a non-neuronopathic (Scheie) phenotype at the far end of the spectrum.

In both the non-neuronopathic and neuronopathic forms, visceral complications occur, such as joint abnormalities, hepatomegaly, cardiac valve abnormalities, skeletal abnormalities and corneal clouding.

The most severe expression of the disease is found in the neuronopathic form; here visceral symptoms occur very early in life, with concomitant devastating, irreversible central nervous system involvement, giving rise to considerable morbidity from a very early age onwards and death on average around the 5th year of age.

In the Scheie phenotype psychomotor development is normal. Skeletal and joint manifestations form the important disease burden in these patients.

Recently, trials with weekly a-L-Iduronidase (Aldurazyme™, Genzyme/Biomarin) infusions showed improvement in joint mobility, lung function and exercise tolerance, as determined by the 6 minute walk test. Aldurazyme™ received marketing approval as an orphan drug from the EMEA in April 2003.

However, there are still many open issues regarding the efficacy of treatment, making uniform evaluation of treatment in selected groups of MPS I patients mandatory.

Doel van het onderzoek

MPS I patients can be treated with Aldurazyme safely and effectively.

Onderzoeksopzet

N/A

Onderzoeksproduct en/of interventie

Enzyme replacement therapy with Aldurazyme.

Contactpersonen

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

1. The patient must give written informed consent;

2. If the patients is younger than 12 years, informed consent from his/her parents or his/her

legal representative is necessary;

3. If the patient is below 18 years, but older than 12 years, informed consent from the child is necessary if the patient is mentally and physically able to do so;

4. The patients can be included in this protocol, and not in any of the two other MPS I treatment protocols;

5. The patient must have a current diagnosis of MPS I, as documented by a decreased á-L-Iduronidase activity in leukocytes or fibroblasts;

6. Patients must be willing and able to comply with the study protocol;

7. Female patients must have a negative pregnancy test, and must use a medically accepted method of contraception during the study.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

1. Patient is unable or unwilling to comply with the study protocol;

2. Parent(s) or legal representatives are unable or unwilling to comply with the evaluation program;

3. Patient is pregnant or lactating;

4. Life expectancy < 6 months;

5. Very severe neurological involvement as evidenced by:

a. Total or subtotal absence of cortical activity (vegetative state);

b. Untreatable seizures;

c. Loss of (almost) all abilities to communicate.

Onderzoeksopzet

Opzet

Type: Interventie onderzoek

Onderzoeksmodel:	Parallel
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestopt
(Verwachte) startdatum:	01-01-2004
Aantal proefpersonen:	35
Type:	Werkelijke startdatum

Ethische beoordeling

Positief advies

Datum:	12-09-2005
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	NL331
NTR-old	NTR369
Ander register	: N/A
ISRCTN	ISRCTN22324060

Resultaten

Samenvatting resultaten
N/A