

Femoral head pathology in Sanfilippo disease.

Published: 10-05-2012

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To evaluate the prevalence of femoral head pathology in patients with MPS III.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Metabolic and nutritional disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON37709

Source

ToetsingOnline

Brief title

Sanfilippo hip study

Condition

- Metabolic and nutritional disorders congenital
- Joint disorders

Synonym

MPS III, Sanfilippo disease

Research involving

Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum

Source(s) of monetary or material Support: stichting Kinderen en Kansen

Intervention

Keyword: Femoral head pathology, MPS III, Sanfilippo disease

Outcome measures

Primary outcome

What is the prevalence of femoral pathology in (Dutch) MPS III patients?

Secondary outcome

Can the abnormalities be classified using the avascular necrosis classification (Herring).

Do patients with femoral head pathology have clinical signs and symptoms which may predict the presence of these complications?

Study description

Background summary

Sanfilippo syndrome (MPS III) is a lysosomal storage disorder consisting of four different subtypes, depending on the deficient enzyme in the degradation of the glycosaminoglycan (GAG) heparan sulfate (HS). The disease is characterized by progressive and severe neurodegeneration. The clinical course does not differ between the four subtypes.

The somatic signs and symptoms of the disease are mild compared to the severe central nervous system damage. However, in the recent years we have diagnosed moderate to severe femoral head abnormalities, resembling avascular femoral head necrosis (AVN), in several young patients with Sanfilippo disease, all presenting with signs of hip pain. AVN was previously reported in MPS III patients (REF!) but the prevalence of this symptom in MPS III patients is not known.

As cognition and communication are almost invariably severely affected in patients with MPS III it can be very difficult to assess if *discomfort* in a patient with MPS III is due to hip pain or to other somatic or non-somatic causes. As a consequence, the prevalence of femoral head disease is probably underestimated. This may result in inadequate care and treatment.

Study objective

To evaluate the prevalence of femoral head pathology in patients with MPS III.

Study design

Approximately 40 patients with confirmed MPS III will participate in the study. The study will take 6 months. From each patient two X-rays of the hip will be taken. Also length, weight and a history of pain and discomfort or problems in relation of hipfunction will be reported by a questionnaire. These X-rays will be evaluated on femoral head pathology by a radiologist.

Study burden and risks

Radiation exposure, and incidences occurring while performing the X-rays, because of unexpected movements of the patient.

Contacts

Public

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)

Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

The patient should have a biochemically diagnosed MPS III, type A, B or C and living in the Netherlands.

Exclusion criteria

The parent or legal representative is unwilling to participate.

Study design

Design

Study type: Observational invasive

Masking: Open (masking not used)

Control: Uncontrolled

Primary purpose: Other

Recruitment

NL

Recruitment status: Recruitment stopped

Start date (anticipated): 20-05-2012

Enrollment: 40

Type: Actual

Ethics review

Approved WMO

Date: 10-05-2012

Application type: First submission

Review commission: METC Amsterdam UMC

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

ID: 25923

Source: Nationaal Trial Register

Title:

In other registers

Register	ID
CCMO	NL39185.018.12
OMON	NL-OMON25923