Prevalence Assessment of Unrecognized Mucopolysaccharidosis I, II, IVA, and VI in Juvenile Idiopathic Arthritis Patients with low inflammatory markers

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Primary objective:To determine the prevalence of unrecognized mucopolysaccharidosis (MPS) I (Hurler, Hurler-Scheie, or Scheie syndromes), II (Hunter syndrome), IVA (Morquio syndrome), and VI (Maroteaux-Lamy syndrome) patients among a population of...

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

Summary

ID

NL-OMON45958

Source
ToetsingOnline

Brief title
ASY13969

Condition

- Metabolic and nutritional disorders congenital
- Inborn errors of metabolism
- Joint disorders

Synonym
Juvenile Arthritis, Juvenile Idiopathic Arthritis

Research involving
Human

Sponsors and support

Primary sponsor: Sanofi-aventis
Source(s) of monetary or material Support: sanofi-aventis

Intervention

Keyword: International Multicenter, Juvenile Idiopathic Arthritis, Mucopolysaccharidosis, Non-interventional

Outcome measures

Primary outcome

Prevalence rate (number of new diagnosed cases among the total enrolled

patients) of MPS I, II, IVA, and VI in pediatric population treated in

pediatric rheumatology clinics.

Secondary outcome

- JADAS-27 score: Active Joint Count + Patient Global Evaluation +

Physician Global Assessment + Erythrocyte Sedimentation Rate

- Duration of morning stiffness

Study description

Background summary

Because of the overlap of symptoms, MPS disorders are often misdiagnosed as juvenile idiopathic arthritis (JIA). Patients with MPS disorders lack the enzymes necessary to metabolize glycosaminoglycans (GAGs). GAGs then accumulate in organs, which can lead to extensive damage to multiple organ systems and potentially life-threatening conditions. When a MPS disorder is undiagnosed for years, irreparable damage has often occurred. Prompt recognition is the key to early initiation of therapy, which is closely linked to the prognosis and outcome. Enzyme replacement therapies are available for MPS I, II, and VI (and in development for MPS IV).
The goal of this investigation is to determine how often children with unrecognized MPS I, II,
IVA, and VI are presenting in experienced pediatric rheumatology practices.

Appropriate screening to determine the true prevalence of such presentation is an important first step in developing an educational program for physicians in such clinics to facilitate the recognition and appropriate referral of patients with MPS disorders.

Study objective

Primary objective:
To determine the prevalence of unrecognized mucopolysaccharidosis (MPS) I (Hurler, Hurler-Scheie, or Scheie syndromes), II (Hunter syndrome), IVA (Morquio syndrome), and VI (Maroteaux-Lamy syndrome) patients among a population of pediatric rheumatology patients with low inflammatory markers (ESR and or CRP) using the dried blood spot (DBS) testing to screen for MPS.

Secondary objective:
To study the pattern of Joint involvement in Juvenile Idiopathic Arthritis (JIA) patients.

Study design

International Multicenter study, Non-interventional

Study burden and risks

Risks and burdens related to blood collection

Contacts

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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

- Male or female juvenile idiopathic arthritis patients, between 6 months and 18 years of age, inclusive.
- History of JIA documented at an experienced pediatric rheumatology clinic. All JIA subtypes can be included if patients have at least 1 low ESR (*20 mm/Hr) and/or CRP (*10 mg/L) value measured at a preceding visit (timelines of the precedent visit is defined as per the patient standard of care) or at the study visit, and assessed as being independent from concomitant anti-inflammatory/anti-infective treatments at the discretion of the investigator.
- Signed informed consent/assent obtained from patient and patient*s legal representative (parents or guardians) according to local regulations.

Exclusion criteria

- Patients for whom MPS enzyme activity tests (ie, enzyme levels tested in fibroblasts, leukocytes, serum, or blood spots) have already been performed and for which the result was normal. (Patients who have been screened for MPS through urinary GAG and tested normal may be included in the study).
- Patients with at least 1 high ESR (>20 mm/Hr) and/or CRP (>10 mg/L) value measured at a preceding visit or at the study visit, not related to an identified concomitant infection or intercurrent illness at the discretion of the investigator.
- Patient has any medical condition or extenuating circumstance which, in the opinion of the Investigator, could interfere with the patient*s ability to complete the study procedure, or with the interpretation of study results.

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	18-02-2016
Enrollment:	120
Type:	Actual

Ethics review

Approved WMO Date:	09-12-2015
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)
Approved WMO Date:	06-06-2016
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)
Approved WMO Date:	15-03-2017
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)

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
CCMO	NL54603.041.15