A Multicenter, Multinational, Longitudinal Clinical Assessment Study of Subjects with Mucopolysaccharidosis IVA (Morquio Syndrome).

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The objective of the study is to quantify endurance and respiratory function in subjects with MPS IV A and to better characterize the spectrum of symptoms and biochemical abnormalities in MPS IV A disease over time.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Cytoplasmic disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON36408

Source
ToetsingOnline

Brief title
A Cross-sectional Clinical Assessment of Subjects with MPS IV A.

Condition

- Cytoplasmic disorders congenital
- Inborn errors of metabolism

Synonym
metabolic disease, syndrome of Morquio

Research involving
Human

Sponsors and support

Primary sponsor: BioMarin
Source(s) of monetary or material Support: BioMarin Pharmaceutical Inc.

Intervention

Keyword: Clinical Assessment, MOR-001, Morquio Syndrome, Mucopolysaccharidosis IV A

Outcome measures

Primary outcome

To quantify the endurance and respiratory function.

Secondary outcome

To characterize the spectrum of symptoms and biochemical abnormalities.

Study description

Background summary

Mucopolysaccharidosis IV type A (MPS IV A, also known as Morquio syndrome) is a disease charachterized by a deficient activity of N-acetylgalactosamine 6-sulfatase (GALNS) causing excessive lysosomal storage of keratan sulfate (KS). This excessive storage causes systemic skeletal dysplasia, short stature, and joint abnormalities, all of which limit mobility and endurance. Malformation of the thorax and obstructive disease impair respiratory function. Odontoid process hypoplasia and ligamentous laxity cause instability of the cervical spine that may lead to cord compression. Other symptoms may include hearing loss, corneal clouding, and heart valvular disease, among others. In a questionnaire-based survey study, most subjects reported progressive skeletal dysplasia, frequent surgical procedures, and limitation in walking distance. However, no clinical assessment studies have been conducted that were based on observation and testing of a substantial number of subjects affected with MPS IV A. Characterization of clinical impairments across a large subject population is expected to improve understanding of the disease and facilitate selection of appropriate clinical and biomarker endpoints for future therapeutic clinical studies.

Study objective

The objective of the study is to quantify endurance and respiratory function in

subjects with MPS IV A and to better characterize the spectrum of symptoms and biochemical abnormalities in MPS IV A disease over time.

Study design

A multicenter, multinational, longitudinal, cross-sectional study in subjects diagnosed with MPS IV A.

Study burden and risks

Patients are asked about their medical history, a physical examination is done, a visual acuity test, a neurological test, a 6-minute walking test and a 3-minute stair climb test, a respiratory examination, an electrocardiogram, a cardiac ultrasound, a questionnaire about the health status and the use of health facilities. Besides that a urine sample and a blood sample are taken and examined.

Contacts

Public
BioMarin

105 Digital Drive
Novato, CA 94949
US
Scientific
BioMarin

105 Digital Drive
Novato, CA 94949
US

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

• Willing and able to provide written, signed informed consent, or in the case of subjects age < 18 years, provide written assent (if required) and written informed consent by a legally authorized representative after the nature of the study has been explained, and prior to any research-related procedures.
• Documented history of reduced GALNS activity relative to the normal range of the laboratory
performing the assay, or documented result of molecular genetic testing confirming diagnosis of
MPS IVA.
• Willing and able to comply with all study procedures.

Exclusion criteria

• Use of any investigational product or investigational medical device within 30 days prior to screening.
• Previous hematopoietic stem cell transplant (HSCT).
• Concurrent disease or condition that would interfere with study participation or pose a safety
concern.

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	01-07-2009
Enrollment:	25
Type:	Actual

Ethics review

Approved WMO

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

No registrations found.

In other registers

Register	ID
CCMO	NL27978.018.09