# VKS/Erasmus MC Mucopolysaccharidosis Survey

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1. To include the patients* point of view in the characterization and description of the mucopolysaccharidoses; and to compare between the different types of mucopolysaccharidoses.2. To assess *health damage* at the moment of diagnosis, in order to…

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| **Ethical review** | Approved WMO |
| **Status** | Recruiting |
| **Health condition type** | Metabolic and nutritional disorders congenital |
| **Study type** | Observational non invasive |

## Summary

### ID

NL-OMON35162

**Source**
ToetsingOnline

**Brief title**
VKS/Erasmus MC MPS Survey

### Condition

- Metabolic and nutritional disorders congenital
- Inborn errors of metabolism
- Musculoskeletal and connective tissue disorders congenital

**Synonym**
Hunter's syndrome, Hurler's syndrome, Maroteaux-Lamy's syndrome, Morquio's syndrome, mucopolysaccharidosis, Sanfilippo's syndrome, Sly's syndrome

**Research involving**
Human

### Sponsors and support

**Primary sponsor:** Erasmus MC, Universitair Medisch Centrum Rotterdam

**Source(s) of monetary or material Support:** Ministerie van OC&W

## Intervention

**Keyword:** clinical course, mucopolysaccharidosis, quality of life, questionnaire

## Outcome measures

### Primary outcome

The results of the Mucopolysaccharidosis Questionnaire will be used to describe the natural course of MPS, the current clinical outcome, the health damage at diagnosis and the effect of treatment. The scores obtained from the standardized questionnaires will be used to measure the consequences of MPS disease on daily activities, quality of life, development and pain and to evaluate. the effects of treatment options.

### Secondary outcome

not applicable

# Study description

### Background summary

The mucopolysaccharidoses (MPS) are a group of lysosomal storage disorders, all caused by the deficiency of a specific enzyme required for the stepwise degradation of glycosaminoglycans (GAGs), also known as mucopolysaccharides. The clinical features of these mucopolysaccharidoses are to a large extent comparable, although there are differences in the level of involvement of the different organ systems. In all MPS there is a chronic and progressive clinical course leading ultimately to premature death. Different treatment options (enzyme replacement therapy and haemopoietic stem cell therapy) are available for several MPS. The current protocol describes a patient oriented Mucopolysaccharidosis Survey which gathers information on clinical course and quality of life directly from patients themselves by means of questionnaires.. The project is a joint initiative of the Erasmus MC Center for Lysosomal and Metabolic Diseases and the patient organization for adults and children with

metabolic disorders (VKS).

### Study objective

1. To include the patients* point of view in the characterization and description of the mucopolysaccharidoses; and to compare between the different types of mucopolysaccharidoses.
2. To assess *health damage* at the moment of diagnosis, in order to estimate to what extent patients could benefit from early diagnosis via newborn screening and early treatment;
3. To include the patients* point of view in the evaluation of the long-term effects of available treatment options and support measures including enzyme replacement therapy and haemopietic stem cell therapy.

### Study design

Longitudinal observational study

### Study burden and risks

There are no risks involved in this study. The patient and/or the parents of the patients fill in a maximum of five questionnaires once a year. This will take approximately 1* hour of their time.

## 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

All patients (all ages) who have signed the VKS / Mucopolysaccharidosis Survey patient information and authorization form and have a confirmed diagnosis of mucopolysaccharidosis are eligible for inclusion.

### Exclusion criteria

none

## Study design

### Design

**Study type:** Observational non invasive

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|---|---|
| Masking: | Open (masking not used) |
| Control: | Uncontrolled |
| Primary purpose: | Other |

### Recruitment

NL

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|---|---|
| Recruitment status: | Recruiting |
| Start date (anticipated): | 02-08-2010 |
| Enrollment: | 100 |

| | |
|---|---|
| Type: | Actual |

## Ethics review

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| Approved WMO Date: | 28-05-2010 |
| Application type: | First submission |
| Review commission: | METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam) |

## 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 | NL29695.078.10 |