Orofacial abnormalities of patients with mucopolysaccharidoses or mucolipidosis II or III in the Netherlands, a singlecenter study

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To investigate the prevalence and existence of orofacial abnormalities, in patients with Mucopolysaccharidoses and Mucolipidosis type II and III and how these abnormalities impair the quality of life.

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

Summary

ID

NL-OMON49498

Source
ToetsingOnline

Brief title
Orofacial abnormalities of patients with MPS or ML II/III

Condition

- Metabolic and nutritional disorders congenital
- Lipid metabolism disorders

Synonym
Hurler-Scheie Syndrome, metabolic disorder

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: mucolipidosis, mucopolysaccharidosis, Orofacial

Outcome measures

Primary outcome

- The incidence of orofacial abnormalities, determined with light photos, CBCT-scans, 3D stereophotographs, Temporomandibular Joint Examination (Diagnostic Criteria for Temporomandibular Disorders, DC-TMD) and a general dental/oral examination.

- Outcomes of questionnaires filled out by the patient/caregiver (OHIP-14 / COHIP (Child Oral Health Impact Profile) and Darriford appearance scale (for non-neuronopathic adult patients)).

Secondary outcome

n.a.

Study description

Background summary

Orofacial abnormalities in MPS and ML patients have not yet been investigated in a standardized manner in a large cohort of patients. The frequency and severity of these abnormalities are unclear. Both are important for developing protocols for early detection of orofacial problems and prevention of complications, to improve quality of life.

Study objective

To investigate the prevalence and existence of orofacial abnormalities, in

patients with Mucopolysaccharidoses and Mucolipidosis type II and III and how these abnormalities impair the quality of life.

Study design

The study design is a cross-sectional cohort study with all MPS and ML II and III patients (both children and adults) in the Erasmus MC. The patients will be asked to come visit the department of oral and maxillofacial surgery of the Erasmus MC once to investigate orofacial abnormalities by making light photos, a Cone-Beam CT (CBCT) (three-dimensional imaging of the jaws and maxillofacial structures) and 3D stereophotographs, by performing a Temporomandibular Joint examination (Diagnostic Criteria for Temporomandibular Disorders, DC-TMD), a general dental/oral examination and filling out questionnaires regarding their subjective wellbeing, considering their oral health (using the (Child) Oral Health Impact Profile (OHIP-14 / COHIP) questionnaire) and (for non-neuronopathic adult patients, using the Darriford appearance scale) their appearance.

Study burden and risks

The risk of side-effects is very limited. The patients will visit the department of oral and maxillofacial surgery of the Erasmus MC one day(part). This visit can be combined with the standardised annual follow-up of these patients. The patients receive an extensive examination of orofacial features, which is important for early detection of orofacial problems and preventing complications.

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

Patients diagnosed with MPS or ML II or ML III (enzyme and/or DNA diagnostics) and known in the Erasmus MC.

Exclusion criteria

- Patients who don't want to participate in the study;
- Patients with an end-stage disease or other medical conditions making the examinations impossible.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-01-2021
Enrollment:	60
Type:	Anticipated

Ethics review

Approved WMO

Date:	30-03-2021
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	NL74342.078.20