Endo- and extranasal sequelae of Treacher-Collins syndrome

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Primary Objective: To determine the prevalence of endo- and extranasal deformities, problems with nasal functioning and satisfaction with nasal appearance in patients with TCS.Secondary Objective: To adjust and improve the treatment of patients with...

Ethical review	Approved WMO
Status	Pending
Health condition type	Musculoskeletal and connective tissue disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON37845

Source
ToetsingOnline

Brief title
Nasal sequelae of Treacher-Collins syndrome

Condition

- Musculoskeletal and connective tissue disorders congenital
- Congenital respiratory tract disorders

Synonym
Treacher-Collins syndrome

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Fonds Nuts Ohra en Carolien Bijl stichting

Intervention

Keyword: endonasal deformities, extranasal deformities, nasal functioning, treacher-collins syndrome

Outcome measures

Primary outcome

Study parameters/endpoints

Main study parameter/endpoint

Main study parameters/endpoints will be a detailed description of all endo- and extranasal deformities (description of affected anatomical structures), nasal functioning and satisfaction with nasal appearance (as addressed in NAFEQ score).All are described in more detail in the -following- study procedures

Secondary outcome

Secondary endpoint is a change in the current treatment of TCS as will be described in (as part of) a treatment protocol.

Study description

Background summary

Treacher-Collins syndrome (TCS) is clinically associated with endonasal and extranasal deformities and problems with nasal functioning. Patient reports learn us that these functional problems of the nose are considered the most significant impairment. However, the anatomical explanation for these findings is lacking.

Study objective

Primary Objective: To determine the prevalence of endo- and extranasal deformities, problems with nasal functioning and satisfaction with nasal appearance in patients with TCS.
Secondary Objective: To adjust and improve the treatment of patients with TCS according to our outcomes and summarize that together with the results of

MEC-2008-402 in a treatment protocol.

Study design

We will conduct a cross-sectional cohort study with a single cohort: a TCS group. Patients in the TCS group are included if they are diagnosed with TCS and have been treated at the multidisciplinary craniofacial team of the Erasmus MC, University Medical Center since 1974 (i.e. a 37 year period). All adult patients are eligible for inclusion.

Study burden and risks

History: A history anamnesis will be obtained concerning medical nasal issues (five minutes).
Questionnaire: The NAFEQ score (Nasal Appearance and Function Evaluation Questionnaire) will be filled out by the patients (five minutes).1
Physical examination: Anterior rhinoscopy and the Cottle test will be performed (approximately five minutes).
Additional examination: Endonasal deformities will be determined with a standardized flexible laryngoscope (approximately 5-10 minutes). No adverse events are expected. Standardized digital photography will determine extranasal deformities (10 minutes).
Outcomes of our (planned) study can determine the cause of reported endonasal complaints and anatomic origin of extranasal deformities. Outcomes of this study could lead to an additional treatment focus and thus to an improved treatment of TCS based on a sufficient level of evidence. Multidisciplinary treatment will be further optimised. This study will give some burden to the patient with regard to the aspects that are not part of the standard clinical practice. No part of this study is associated with (serious) adverse events and the risks are negligible. Patients will receive a result of the endoscopy and this will be saved in the medical chart and if necessary patients will be treated.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

diagnosis Treacher Collins syndrome
informed consent
ability to speak Dutch
18 years or above

Exclusion criteria

no ability to speak Dutch
age < 18 years

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-07-2012
Enrollment:	22
Type:	Anticipated

Ethics review

Approved WMO

Date:	11-07-2012
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	NL39809.078.12