

# Molecular Basis of Unilateral Condylar Hyperplasia (UCH)

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<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruitment stopped
<b>Health condition type</b>	Congenital and hereditary disorders NEC
<b>Study type</b>	Observational non invasive

## Summary

### ID

NL-OMON45461

### Source

ToetsingOnline

### Brief title

Molecular Basis of UCH

### Condition

- Congenital and hereditary disorders NEC

### Synonym

unilateral condylar hyperplasia of the mandible, unilateral overgrowth of the lower jaw

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Academisch Medisch Centrum

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

### Intervention

**Keyword:** aetiology, condylar hyperplasia, genetics, maxillofacial surgery

## Outcome measures

### Primary outcome

detection of the gene causing UCH

### Secondary outcome

Understanding of the molecular and cellular mechanisms leading to the various manifestations of UCH

## Study description

### Background summary

Unilateral condylar hyperplasia or hyperactivity (UCH) is clinically characterized by varying degrees of mandibular overgrowth. Obwegeser et al. (1986) described a classification in 3 categories: hemimandibular elongation (HE), hemimandibular hyperplasia (HH) and a combination of these two (hybrid form). The expected concomitant skeletal and dental parameters of these categories can include a class III malocclusion on the affected side with a chin and midline deviation and cross-bite on the non-affected side, and/or a malocclusion with canting of the occlusal plane and unilateral open bite at the affected side without midline deviation respectively. The asymmetrical development in UCH patients is often attended with functional and aesthetic problems.

Treatment consists of removal of the growth centre by a partial condylectomy in case of ongoing activity. Secondary to this or in patients in whom the hyperactivity ceased, the remaining asymmetry can be corrected by orthognathic surgery and remodelling reconstruction of the lower face.

To date, there is little information about the clinical characteristics and demographic features of this group of patients worldwide. No incidence or prevalence is available. According to a recent, as yet unpublished Dutch study (Nolte, Becking et al, in preparation) approximately 30 patients with possible progressive mandibular asymmetry present each year, with confirmation of the diagnosis UCH in about 10 patients. In general it can be concluded from the available literature that UCH can occur at any age with a predilection for early twenties, and that UCH tends to occur more often in females. It seems there is no preference for left or right affected side, and no association between laterality of UCH and gender is found.

Several options have been mentioned to explain the nature of this disease such as inflammation, trauma, endocrine imbalance, hypervascularity, neoplasms and

genetic factors (Saridin et al., 2009). None of these however are confirmed by evidence based research. Basically, a congenital origin of the disease, especially a genetic defect has not been investigated so far.

We hypothesize that in at least part of the patients with UCH the cause is a somatic mutation in a gene that controls cell growth, such as PTEN or all genes acting in the AKT pathway, or PIK3CA-associated segmental overgrowth (eg. Klippel-Trenauney syndrome). Such mechanism has been proven for other entities showing localized overgrowth of tissue such as Proteus syndrome (Lindhurst et al., 2011), hemimegalencephaly (Poduri et al, 2012) and Parry-Romberg syndrome (Hennekam, Lachmeijer et al., unpublished).

The present study is aimed to evaluate this in a small number of patients, and perform next generation sequencing techniques in both fibroblasts derived from the affected condyle of the jaw and from leukocytes, and search for differences between the exomes from the two tissues.

## **Study objective**

Our aim is to find the cause of UCH. The subsequent aim is to study the pathogenesis of the entity. The final aim is to find an effective management for the disorder.

## **Study design**

Observational study with no invasive measurements

## **Study burden and risks**

no risks

blood sampling will be performed during surgery that is already scheduled, from existing access to the venous system

tissue research is performed on mandibular condyle that will be already taken out in general anesthesia for therapeutic reasons

the burden is extremely limited, both blood sampling and bone biopsy are taken at surgical intervention when the patient is under general anesthesia, we estimate this to be acceptable.

there is no individual benefit, as explained tot the participants by the investigators, however if the gene and/or mechanism is found, future individuals can benefit from the outcome. The (timing of) treatment can be adjusted and more precise predictions about the course of the condition can be made.

## Contacts

### Public

Academisch Medisch Centrum

Meibergdreef 9  
Amsterdam 1105 AZ  
NL

### Scientific

Academisch Medisch Centrum

Meibergdreef 9  
Amsterdam 1105 AZ  
NL

## Trial sites

### Listed location countries

Netherlands

## Eligibility criteria

### Age

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

### Inclusion criteria

Patients identified with Unilateral Condylar Hyperplasia, confirmed clinically, radiologically and with bonescan (SPECT)

### Exclusion criteria

None

## Study design

### Design

**Study type:** Observational non invasive

Masking: Open (masking not used)

Control: Uncontrolled

Primary purpose: Basic science

### Recruitment

NL

Recruitment status: Recruitment stopped

Start date (anticipated): 01-03-2017

Enrollment: 10

Type: Actual

## Ethics review

Approved WMO

Date: 09-02-2017

Application type: First submission

Review commission: METC Amsterdam UMC

Approved WMO

Date: 13-03-2017

Application type: Amendment

Review commission: METC Amsterdam UMC

Approved WMO

Date: 17-01-2018

Application type: Amendment

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**

<b>Register</b>	<b>ID</b>
CCMO	NL59955.018.16