# Description of Mutations

In general, the recommendations of HGVS are followed.

## CHD7 c

The mutation or deletion in the coding DNA for CHD7 is indicated here.
Please leave out the "c." notation and describe the variant in as much detail as possible.
If unknown, fill in "unknown" and describe the variant in "CHD7_p" or in "Mutation_type".

## CHD7 p

The predicted effect on protein level is indicated here. Please leave out the "p." notation.
Use 3-letter amino acid code.
Use short description for frameshift effects. For example: Ser261fs
Use "X" for nonsense mutations. For example: Gln456X
For effects that are not predictable, please choose from the following categories:

- unknown
- deletion/duplication exon *no.* For example: deletion exon 1
- deletion whole gene
- translocation
- For intronic mutation: IVS*no.+locus of mutation*. For example: IVS2+34G>A (in intron 2)

## Exon

The exon or intron of *CHD7* in which the mutation or deletion is located.
For whole gene deletions use "2-38".
For variants located before exon 1 use "5'UTR".

## Pathogenicity

The pathogenicity of a mutation or deletion is indicated here. Please choose from:

- Benign
- Likely benign
- Unclassified variant
- Likely pathogenic
- Pathogenic

## Mutation type

The following mutation types are indicated here:

- Deletion
- Deletion whole gene
- Duplication
- Frameshift
- Missense
- Nonsense
- Silent
- Splice site

- Translocation
- Unknown

## Other information

Other information on the mutation or deletion can be mentioned here, including information on allele frequencies, how often the mutation or deletion is detected in healthy controls, and the rs-numbers in the dbSNP Database.

# Description for Patient's information

## Pubmed ID

If the patient is described in a published paper, report a link to the paper in Pubmed here.
For example: http://www.pubmed.com/20624498

## Original ID reference

If the patient is described in a published paper, report the ID number used in the paper here.

## Positive family history

"yes" if the patient has a positive family history, which means family members with CHARGE syndrome and/or a pathogenic CHD7 variant.
"no", if the patient has no family members with CHARGE syndrome and/or a pathogenic CHD7 variant.
"unknown" if information on family history is not available

## Familial information

Additional information on family members is reported here, including both positive and negative findings. For example: parent with CHD7 variant has no coloboma, heart defect or deafness on examination.

## Phenotype

The clinical diagnosis is mentioned here. Choose from:

- CHARGE syndrome
- Kallman syndrome
- Control (in research settings)
- Other
- Unknown

**For the following clinical features, unless stated otherwise report:**

- **"yes" if the clinical feature complied with the definition**
- **"no" if the feature did not comply with the definition**
- **"unknown" if not tested**

### Coloboma

Coloboma of the iris, retina, choroid and/or disc, including microphthalmia. Unilateral or bilateral.

### Congenital heart defect

Includes congenital cardiovascular malformations of all types, such as conotruncal defects (e.g. tetralogy of Fallot), AV canal defects, and aortic arch anomalies.

### Choanal anomaly

Choanal atresia or stenosis, bony or membranous. Unilateral or bilateral.

### C(L)P
Cleft lip and/or palate.

### Growth retardation
If height is below the third percentile or < 2.5 SD, with or without growth hormone deficiency.

### Developmental delay
If IQ < 70 or when developmental delay is reported, including delayed motor milestones.

### Genital hypoplasia
Includes micropenis and/or cryptorchidism or hypoplastic labia. Delayed, incomplete pubertal development with or without gonadotrophin deficiencies is also included here.

### Sense of smell
Choose

- Normosmia (normal smell)
- Hyposmia (diminished smell)
- Anosmia (absent smell)

### External ear anomaly
“yes” if any external ear anomalies are present. For example absent or hypoplastic lobes, prominent antihelix, decreased cartilaginous folds, and triangular concha.

### Hearing loss
Includes both sensorineural as well as conductive hearing loss.

### Semicircular canal anomaly
Includes absent or hypoplastic semicircular canals as seen on CT or MRI.

### Facial palsy
Defect of facial nerve. Unilateral or bilateral.

### TE anomaly
Tracheo-esophageal defects of all types, including fistula.

### Feeding difficulties
“yes” if tube feeding is necessary or when swallowing problems or gastro-esophageal reflux are present.

### Other
Other phenotypic features can be mentioned here, for example skeletal, renal, and immunological abnormalities.

### Deceased
If patient was deceased please choose at which age period:

- pregnancy termination
- neonatal
- < 10 years
- $\geq$ 10 years

- age at death unknown

“no” if patient is alive.