

## Clinical checklist

### 'Disorder of sex development-Primary Ovarian Insufficiency-Hypogonadotropic Hypogonadism (DSD-POI-HH) gene panel'

CENTRUM MEDISCHE GENETICA UZ GENT

Providing relevant clinical information is required for DSD/HH/POI panel analysis. We kindly ask you to be as precise and specific as possible. We welcome clinical letters and laboratory reports as they contribute to a correct data interpretation.

This checklist can be found online at <https://www.cmgg.be/en/professionals/forms/test-specific-questionnaires>

PATIENT FIRST NAME: .....

REFERRING PHYSICIAN: .....

PATIENT SURNAME: .....

REFERRING CENTER: .....

DATE OF BIRTH: Choose date.

AGE AT EXAMINATION: .....

REASON FOR ENCOUNTER: .....

## 1. Genetic investigations

Karyotype: .....

FISH for SRY:      present    absent    not performed

Other WES panels / targeted testing (please specify genes or panel and genetic center):

.....

## 2. Clinical information

Birth weight: .....

External genitalia score (EGS): ... /12 (see table).

Please complete the table below by encircling the appropriate items  
(GT=genital tubercle)

EGS	Labio-scrotal fusion	Genital tubercle (mm)	Urethral meatus	Left gonad	Right gonad
3	Fused	>31	Top of the GT		
2.5		26-30	Coronal Glandular		
2			Along the GT		
1.5	Posterior fusion	21-25	At the GT base	Labioscrotal	Labioscrotal
1		10-20	Labio-scrotal	Inguino- Labioscrotal	Inguino- Labioscrotal
0.5				Inguinal	Inguinal
0	Unfused	<10	Perineal	Impalpable	Impalpable

**Internal genitalia**

Uterus:  present  absent  Mullerian remnants  uncertain

Gonads:

- Left:  testis  ovary  (likely) dysgenetic  absent  ovotestis
- Right:  testis  ovary  (likely) dysgenetic  absent  ovotestis

Other particularities: .....

Please specify how the information was obtained (several options possible):

- Physical examination  MRI
- Ultrasound  Laparoscopy

Gonadal biopsy performed:  no  yes

History of malignancy:  no  yes

- Gonadoblastoma / germ cell neoplasia in situ
- Invasive germ cell cancer: please specify tumor type
- Wilms' tumor

Gonadectomy:  no  yes: please specify the center

Histological assessment of gonads:  yes  not performed

*If yes, please add a copy of the results*

**Secondary sexual characteristics**

Pubertal development:  normal  absent  delayed  unexpected virilization

- Primary amenorrhea
- Gynecomastia
- Other particularities: .....

**Other**

History of inguinal hernia:  no  yes

Adrenal insufficiency:  no  yes  not known

Renal abnormalities:  no  yes  not known

*If yes (proteinuria, chronic renal failure, other): Please specify*

Congenital heart defects:  no  yes  not known

*If yes: Please specify*

Other clinical features:

.....  
.....

**3. Hormonal assessment** *(prior to gonadectomy)*

Please add a copy of hormonal results and please specify circumstances if applicable (e.g. mini-puberty, hCG/Pregnyl test, LHRH test...)

**4. Familial history for DSD**

Consanguineous parents:  no  yes  unknown

Affected family members:  no  yes  unknown

*If yes, please indicate which family members are affected and their phenotype:*

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