|  |
| --- |
| 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**: Klik en vul aan.

**PATIENT SURNAME**: Klik en vul aan.

**DATE OF BIRTH**: Choose date.

**REASON FOR ENCOUNTER**: Klik en vul aan. **REFERRING PHYSICIAN**: Klik en vul aan.

**REFERRING CENTER**: Klik en vul aan.

**AGE AT EXAMINATION**: Klik en vul aan.

1. Genetic investigations

Karyotype: Klik en vul aan.

FISH for SRY: present  absent not performed

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

Klik en vul aan.

1. Clinical information

Birth weight: Klik en vul aan.

External genitalia score (EGS): Klik en vul aan. /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: Klik en vul aan.

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: Klik en vul aan.

**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: Klik en vul aan.

1. 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…)

1. 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:

Klik en vul aan.