

Patient's sticker

Phenotypic description

Doctor's name/Speciality

Clinical presentation

•Prepubertal child with ambiguous genitalia ➔ **Fill Section B**

•Adolescent

Ambiguous genitalia

Pubertal delay

➔ **Fill Section A and B**

Section A

•Chronological age •Bone Age on X-Ray	•Tanner's scale Gonads..... Breasts..... Hair.....
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Family history

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Section B

Assessed by clinical geneticist		Yes	<input type="checkbox"/>	No	<input type="checkbox"/>	<i>Syndromic</i>	<input type="checkbox"/>
Microarray	Yes	<input type="checkbox"/>	No	<input type="checkbox"/>		<i>Non syndromic</i>	<input type="checkbox"/>
SNP	Yes	<input type="checkbox"/>	No	<input type="checkbox"/>	Results		
CGH	Yes	<input type="checkbox"/>	No	<input type="checkbox"/>			
Combined	Yes	<input type="checkbox"/>	No	<input type="checkbox"/>			

Karyotype 46, XY 46, XX Other.....

Associated malformations and diagnosis

Face morphology Axial skeleton Oral cleft Cardiovascular system

Neurodevelopmental delay Renal morphology

Details/other.....

Internal genitalia

US MRI Laparoscopy

•Müllerian remnant: None Non-uterus like cavity

Normal uterus Didelphic or bicornate

•Gonads: Right Inguinal Intraabdominal None

Left Inguinal Intraabdominal None

•Comments (size, aspects...)

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Clinical description of external genitalia

•Gonads
Right: Vol..... Scrotal Inguinal Not palpable
Left: Vol..... Scrotal Inguinal Not palpable

•Labioscrotal swellings: Completely fused Partially fused Not fused
 (Normal scrotum) (Bifid scrotum) (Labia majora)

Pigmentation No pigmentation Striation No striation

•Labioscrotal folds: Normal foreskin Hooded foreskin Labia minora

•Size of genital tuberclemm

•Urethral meatus:
 Tip of the penis Glans Midshaft Perineum

•Vaginal opening:
 None Not visible distinctively from urethra Separated from
 (common channel, UGS) urethra

Gonadal biopsy Yes No Report attached

Results

Hormonal testing

•Basal levels Age at testing

	Level	Units	Normal range for lab
Testosterone			
DHT			
AMH			
FSH			
LH			
Oestradiol			
Others			

•hCG stimulation test

	Level before	Level after	Normal range for lab
Testosterone			
DHT			
FSH			
LH			
Others			

Additional comments

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