

Molecular Genetics Laboratory of NYU School of Medicine

Genetic Studies in Patients with Disorders of Sexual Development

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NFD Database:

Karyotype – please indicate

FISH (if performed)

Phenotypic Diagnosis

- 46,XX DSD (testes with male somatic phenotype)
- 46,XX DSD (streak gonads with female somatic phenotype)
- 46,XX ovotesticular DSD
- 46,XY pure gonadal dysgenesis (streak gonads)
- 46,XY DSD (ambiguous genitalia and testicular elements on gonadal biopsy)

Additional Clinical Features

Hypospadias [] Yes [] No
Micropenis [] Yes [] No
Ovotestes [] Yes [] No
Cryptorchidism [] Yes [] No
Bifid scrotum [] Yes [] No
Clitoromegaly [] Yes [] No
Blind vaginal pouch [] Yes [] No
Turner phenotype [] Yes [] No

(please specify)

Other (please specify)

Other Diagnostic Tests Baseline Testosterone DHT

LH FSH Inhibin Anti-Mullerian Hormone

ACTH stimulation test

	ACTH	17Δ5 ng/dl	17OHP ng/dl	Δ4 ng/dl	DHEA ng/dl	T ng/dl	DHT ng/dl
Reference							
0 min							
60 min							

HCG stimulation test

	17OHP ng/dl	Δ4 ng/dl	DHEA ng/dl	E1	E2 ng/dl	T ng/dl	DHT ng/dl
Reference							
0 min							
24 hours after HCG x 3							

Gonadal pathology

Treatments (Please describe below and on additional pages)

Date Sample Collected ___ / ___ / ___

Specimen Type

- Blood
- Tissue (describe) _____

PATIENT

NAME _____

DOB: ___ / ___ / ___

SEX: M F UNKNOWN

ETHNICITY

COUNTRY

- 1) African _____
- 2) African-American _____
- 3) Asian _____
- 4) Ashkenazi Jewish _____
- 5) Caucasian _____
- 6) Latino _____
- 7) Other Jewish _____
- 8) Other _____

RELATIONSHIP TO PROBAND _____

FAMILY HISTORY (Be sure to enter relationship of proband to patient and mutation, if known)

SERVICE REQUESTED

- ✓ ArrayCGH
- ✓ DNA Sequencing

REQUESTING PHYSICIAN

NAME: _____

(Please print)

ADDRESS: _____

CITY, STATE, ZIP: _____

TEL # _____

For additional information

http://www.med.nyu.edu/genetics/research/sex_det.html