Cytogenetic testing protocol



Ambiguous genitalia

(enlarged clitoris / small penis)

Obvious dysmorphism

Dysmorphic facial appearance and one or more major structural congenital anomalies.

Suspected chromosomal trisomy disorder

Subtle phenotypic features (such as hypotonia or cardiac problems), no dysmorphism

suspected microdeletion disorder (including Prader Willi and 22q deletion syndromes)

Test request:

FISH for XY and karyotyping*

Test request:

FISH for trisomy 13 &/or trisomy 18 &/or trisomy 21 **and** karyotyping*

Test request:

Microarray

Sample: Lithium heparin blood

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Sample: Saliva or EDTA blood

*Notes:

VCGS will select appropriate karyotype based on FISH result:

- If FISH abnormal = conventional G-banded karyotyping
- If FISH normal = molecular karyotyping* (microarray)

*Molecular karyotype requires informed patient consent in relation to possible results. RACGP provide this useful resource about microarray.

