

Fluorescence in-situ Hybridization (FISH)

SRY (Sex Determining Region Y), Yp11.3 deletion

Specimen: :

Clinical Indication: :

Result: :

Interpretation:

Total metaphase analyzed	Normal metaphases showing 1 Orange 1 Green signals	Abnormal metaphases showing 1 Orange 1 Green signals
20		

PHOTO

Method: FISH analysis performed on 20 metaphases.

Probe: CEP X (DXZ1)(Xp11.1-Xq11.1) Spectrum Green/LSI SRY (Yp11.3) Spectrum Orange.

Remarks:

- Any male individual with SRY signal on structurally Y chromosome is considered negative for a deletion in the region tested.
- Any patient with FISH signal pattern indicating loss of critical region is reported as having deletion of the region tested by the probe
- Any patient with a FISH signal on X chromosome is considered as having a cryptic X;Y translocation involving the critical region

Comments: SRY (Sex determining region on the Y chromosome) gene is required for normal embryonic wolffian (male) genetic development. Some gene mutations block the action of SRY in development. Thus a 46XY individual with SRY deletion / mutation will develop as a female and a 46XX individual with translocation of SRY to 1 X chromosome will develop as a male. SRY negative 46XX males often have ambiguous genitalia whereas those positive for SRY usually have normal phenotype with azoospermia. SRY negative 46XY females may have other mutations also. This test is useful as an aid in the detection of SRY gene in males with Primary infertility, XY females and individuals with ambiguous genitalia.