

FISH Descriptions for Consent Form

1. FISH for SRY(Yp11.3)

FISH for SRY is indicated for individuals with genitalia/karyotype discrepancy, **Gonadal Dysgenesis, and Turner syndrome with Y chromosome material.**

Fluorescence in situ hybridization (FISH) provides clinicians with a way to visualize and map the genetic material in an individual's cells including specific genes or portions of genes.

Genetic Counseling is recommended for the interpretation of FISH testing.

2. FISH for ELN/LIMK1/D7S613 (7q11.23)

FISH for ELN/LIMK1/D7S613 (7q11.23) is used to identify individuals with **Williams Syndrome.** Fluorescence in situ hybridization (FISH) provides clinicians with a way to visualize and map the genetic material in an individual's cells including specific genes or portions of genes. Genetic Counseling is recommended for the interpretation of FISH testing.

3. FISH for SNRPN (15q11-13)

FISH for SNRPN (15q11-13) is used to identify individuals with **Prader-Willi Syndrome (PWS) and Angelman Syndrome (AS) caused by 15q11-13 deletion.**

Fluorescence in situ hybridization (FISH) provides clinicians with a way to visualize and map the genetic material in an individual's cells including specific genes or portions of genes. Genetic Counseling is recommended for the interpretation of FISH testing.

4. FISH for SMS (17p11.2)

FISH for SMS (17p11.2) is used to identify individuals with **Smith-Magenis Syndrome (SMS) and Potocki-Lupski syndrome (PLS) caused by 17p11.2 copy number changes.**

Fluorescence in situ hybridization (FISH) provides clinicians with a way to visualize and map the genetic material in an individual's cells including specific genes or portions of genes. Genetic Counseling is recommended for the interpretation of FISH testing.



5. FISH for LIS1 (17p13.3)

FISH for LIS1 (17p13.3) is used to identify individuals with **Miller-Dieker Syndrome and LIS1 deletion-caused lissencephaly**.

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6. FISH for TUPLE1 (22q11.2)

FISH for TUPLE1 (22q11.2) is used to identify individuals with **DiGeorge Syndrome and 22q11.2 Duplication syndrome**.

Fluorescence in situ hybridization (FISH) provides clinicians with a way to visualize and map the genetic material in an individual's cells including specific genes or portions of genes. Genetic Counseling is recommended for the interpretation of FISH testing.

7. Aneuvysion (FISH), "Pre-Natal FISH panel" (DXZ1,DYZ3,D18Z1)/(RB1,D21S259/D21S341/D21S342)

This is an FDA approved test used to identify gender and detect numerical abnormalities associated with chromosomes 13, 18, 21, X, and Y.

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Genetic Counseling is recommended for the interpretation of FISH testing.