

PRENATAL FAST FISH

Rapid prenatal chromosome diagnosis is very useful in selected circumstances. Unlike conventional chromosome analysis on an amniocentesis sample, which can take up to 14 days because the procedure entails culturing cells. This technique enables identification of Trisomy 21 (Down Syndrome), Trisomy 13, Trisomy 18, and sex chromosome abnormalities in **24 to 72 hours**.

What is prenatal FAST FISH ?

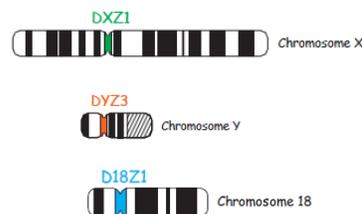
Fluorescent DNA probes are used to analyze the chromosome make up of cells obtained from amniotic fluid. While not all chromosome abnormalities can be identified by this technique, the majority of common abnormalities can be identified. These include Down syndrome (trisomy 21), trisomy 18, trisomy 13, Klinefelter syndrome (47,XXY) Triple-X syndrome (47XXX), Turner Syndrome (45,X) and 47XYY. The technique is known as interphase FISH, which stands for fluorescent in situ hybridization, or simply as FISH. Our laboratory uses the Cytocell Aquarius FAST FISH prenatal probes.

FAST FISH X, Y, 13, 18 and 21

The complete FAST FISH Prenatal screen for the detection and quantification of chromosomes 13, 18, 21, X and Y. Each kit contains two probe sets,

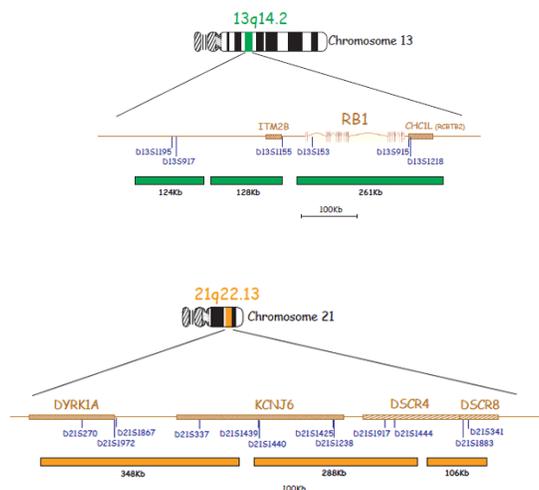
Probe set 1:

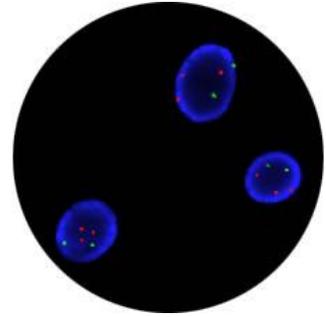
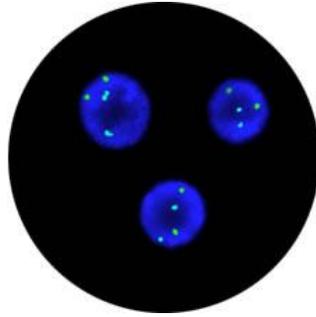
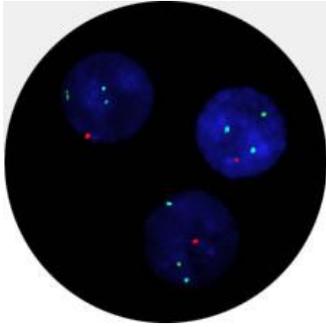
X centromere Xp11.1-q11.1 (DXZ1) Green
Y centromere Yp11.1-q11.1 (DYZ3) Orange
18 centromere 18p11.1-q11.1 (D18Z1) Blue



Probe set 2:

13 unique sequence (13q14.2) Green
21 unique sequence (21q22.13) Orange





FAST FISH Prenatal X, Y and 18 (male sample) FAST FISH Prenatal X, Y and 18 (female sample) FAST FISH Prenatal 13 and 21 (trisomy 21)

When should this be considered?

Sometimes important pregnancy management decision need to be made quickly, while waiting for a complete cytogenetic result. The following are some of the situations in which a rapid assay may be helpful.

- Maternal age, serum screening or ultrasound findings indicated a high risk for Down Syndrome or trisomy 18
- Prenatal chromosome diagnosis is indicated, but the patient is more than 20 weeks gestation.
- An ultrasound examination in the second or third trimester suggests the fetus has Down syndrome, trisomy 18 or trisomy 13.
- A patient has concern about whether the fetus has a chromosome abnormality; for example. A chromosome problem was discovered in a previous pregnancy.

What are the limitations of Prenatal FAST FISH?

- The analysis does not screen for all chromosome abnormalities, only missing or extra copies of chromosome 21,13,18 X and Y.
- Prenatal FISH is not designed to detect chromosome mosaicism, partial chromosome duplications and deletions, or structural rearrangements.
- Although prenatal FISH analysis is highly accurate, irreversible therapeutic decisions should not be made on FISH results alone.
- An additional 5-10 ml of amniotic fluid is required for Prenatal FAST FISH assay, in addition to the 15-20ml required for conventional chromosome studies. Sample with visible blood may not be suitable for FISH analysis.

Genetic consultation for patients undergoing prenatal diagnosis, including Prenatal FAST FISH is often very helpful. This can help patients understand limitations of prenatal diagnosis and FISH analysis and enables physician to order the optimal combination of prenatal diagnostic analyses.