Fluorescence In Situ Hybridization (FISH)

TThe FISH (fluorescence in situ hybridization) test provides results within 24-72 hours, depending on lab and shipping times. It is particularly useful if a blood test (cell-free DNA Screen or Multiple Marker Screen) shows positive for Down syndrome or another chromosomal condition. The FISH test assesses specific chromosomes:

- Chromosome 21
- Chromosome 13
- Chromosome 18
- Chromosome X and Y (sex chromosomes)

However, it is not recommended to rely solely on the FISH test due to the rare possibility of false positives or false negatives. This can occur because the test relies on probes that bind to unique sequences on the chromosomes. If a probe fails to bind or binds to the wrong sequence, the results may be inaccurate. Nevertheless, the FISH test is more accurate than available blood tests.

Who We Are

Modern Reproduction's mission is to facilitate the discussion on the current technologies and genetic testing options available prior and during pregnancy by educating, empowering, and embracing modern reproduction. This handout is not medical advice. Please refer to Modern Reproduction's Disclaimer and Terms and Service on their website.

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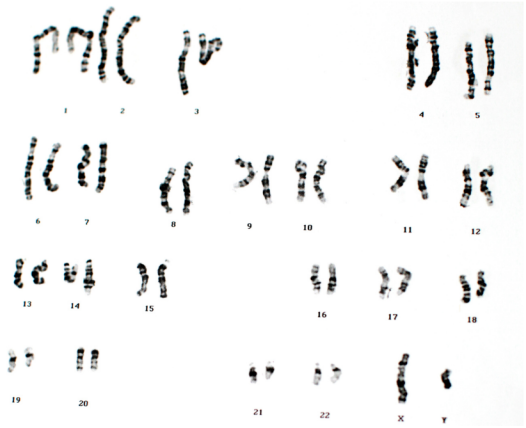
Diagnostic Testing in Pregnancy

What are the genetic testing options?

Karyotype



A karyotype provides a snapshot of our chromosomes to detect any extra or missing copies, as shown in the above picture. It helps determine if these changes occurred by chance or were inherited. Parental karyotypes may be ordered if inheritance is suspected. Results take around 2-3 weeks as cells need to culture and reach a specific stage.

Karyotypes are useful when the FISH test is positive or for common chromosomal conditions. However, karyotypes cannot detect all chromosomal conditions, such as small deletions or duplications which are missing or extra parts of chromosomes.

Microarray Analysis

Microarray analysis can identify small deletions, duplications, and whole chromosomal abnormalities. However, it may detect a variation with unclear health implications called a "variant of uncertain significance." In genetics, sometimes results are uncertain due to limited data on whether it causes a condition or is a normal variation for an individual.

Parental studies are usually offered when a variant is found. If a parent with the variant is healthy, it may suggest no health impact. However, testing parents may not always provide a conclusive answer if other family members have health concerns.

Microarray analysis can also be performed directly on procedure samples, providing results in less than 2 weeks. However, if there are insufficient cells, cell culture may be required, increasing turnaround time.

Gene Testing

Gene Panels
A panel test allows for the testing of multiple genes simultaneously. It is typically ordered when ultrasound identifies a birth defect or there is a suspected diagnosis like Noonan syndrome or a skeletal dysplasia. The genes included in the panel have been carefully selected by the lab and are known to be associated with symptoms that begin during pregnancy.

Known Familial Variants
If a couple or individual is known to be carriers of a genetic condition or has a personal diagnosis with a confirmed genetic cause, it is possible to assess that specific condition.

Whole Exome Sequencing
Whole exome sequencing is now available in certain cases where a pregnancy is found to have multiple birth defects and previous testing has been inconclusive. This test focuses on a specific part of numerous genes, although it does not cover every possible gene. It may uncover variants of uncertain significance and identify conditions unrelated to the initial reason for testing. Before choosing this option, detailed consent forms provided by the labs should be carefully reviewed.