FISH, or fluorescence in situ hybridization, is a test that can be done on the cells from a chorionic villus sampling (CVS) or amniocentesis procedure. FISH can provide rapid results (usually within 48 to 72 hours) for limited chromosome conditions. Most labs offer FISH testing on CVS or amniocentesis cells for chromosomes 21, 18, 13, and the sex chromosomes (X and Y). To learn more about chromosomes, visit our Genetics 101 page.

Each of our chromosomes have sequences of letters, our DNA, that are unique to them. Our DNA is made up four different letters, called nucleotides: A, C, G, and T. These nucleotides work in pairs – A always pairs with T, and G always pairs with C.

Every chromosome has a pattern of these letters that are different than all of the other chromosomes, which is how we can identify them. Once we know what that pattern is, the lab can make a probe (which is a small piece of DNA that is made by the lab) that can find and attach to the matching sequence.

For example, if we know chromosome 18 has the sequence ACG TAT CGC, then the lab would make a probe that has the sequence TGC ATA GCG (remember T matches with A, G matches with C, etc). Once this probe is made, the lab can put it in with the cells from the CVS or amniocentesis procedure. When the probe finds it's matching chromosome, it attaches and then lights up (like a fluorescent light!).

FISH results on CVS and amniocentesis will generally tell you how many of chromosomes 21, 18, 13, X, and Y the lab saw. We generally have two of each chromosome, so if the FISH test shows that three chromosome 21s lit up, that means that there is a high chance that the pregnancy has three number 21 chromosomes, which is what causes Down syndrome.

Because the FISH testing also looks at the X and Y chromosomes, it will also be able to tell whether the pregnancy is genetically male or female. So, if you don't want any spoilers, make sure your doctor or genetic counselor know ahead of time!

Although FISH is thought to be a reliable preliminary test, it is most often recommended to wait until the full genetic testing results have come back before making any permanent decisions about the pregnancy.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

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