Part of the curriculum: Genetics and evolution [1A1 – Chromosomal abnormalities]

FISH Test to Diagnose Down Syndrome

A New Technique Used to Diagnose Down Syndrome

Adapted from www.verywell.com
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FISH stands for fluorescent *in situ* hybridization. FISH testing, or FISH analysis as it is sometimes referred to, is a relatively new cytogenetic technique that allows a cytogeneticist to determine how many copies of a particular chromosome are present without having to go through all of the steps involved in producing a karyotype.

A FISH test is done using a fluorescent probe that binds to certain specific chromosomes. These fluorescent probes are made of DNA specific to certain chromosomes and are tagged with a fluorescent dye. The cells used in FISH analysis don't have to be grown or cultured (which can take 7 to 10 days), so the results of a FISH analysis are available much faster than the results of a karyotype.

After hybridization, the slide is examined under a special microscope that can see fluorescent images. By counting the number of fluorescent signals, a cytogeneticist can determine how many of a specific chromosome are present. For example, a person without Down syndrome will have two fluorescent signals corresponding to their two number 21 chromosomes. Typically, cytogeneticists will use probes for the 13, 18, 21, X and Y chromosomes. These are the chromosomes that can result in trisomies for humans.

The main advantage of FISH analysis is that it can provide information about certain chromosomes quickly. For example, in three to four days, it can tell how many copies of a number 21 chromosome a particular person may have. In contrast, a traditional karyotype can take up to two weeks. The main disadvantage of FISH analysis compared to karyotyping is that FISH analysis gives you less information about all of the chromosomes being studied. For example, a typical prenatal FISH test will tell you how many number 13, 18, 21, X and Y chromosomes are present (i.e., whether there are two copies or three) but will not give you any information about any of the other chromosomes or any information about the actual structure of chromosomes.

After presenting the document, you will use it as well as your scientific knowledge to:

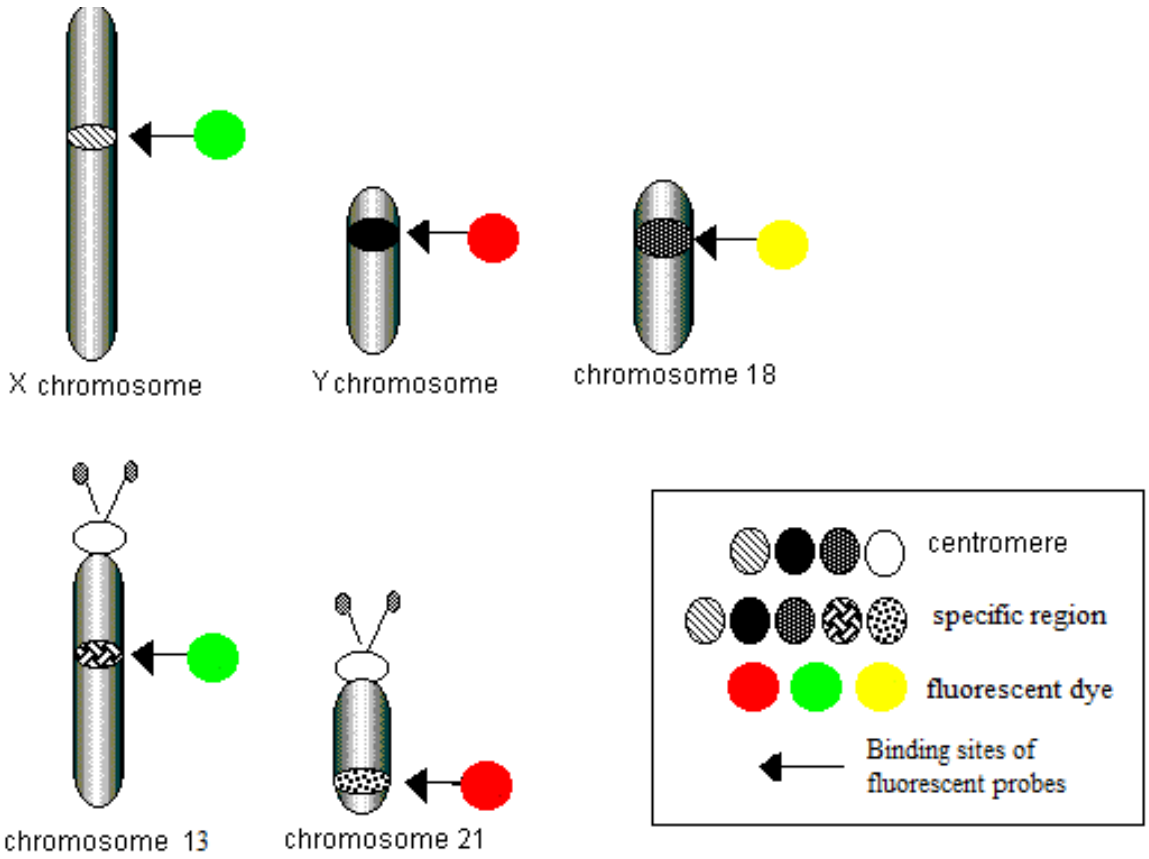
1. Compare this new technique with karyotyping.

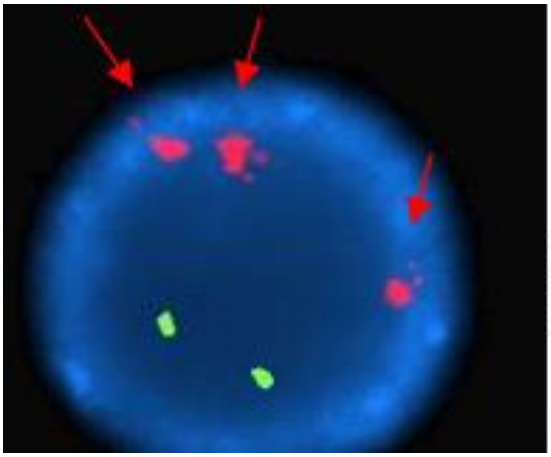
2. Explain the origin of trisomy 21, using sketches and the additional document.

You may use the following key words:

Meiosis – chromosomal abnormality – gametes – nondisjunction

Principles of FISH (fluorescence in situ hybridization)





A cell from amniotic fluid that is positive for trisomy 21 by FISH