

Department of Cytogenetics

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# CYTOGENETIC TESTs



# Department of Cytogenetics

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Cytogenetics is the study of chromosomes in any species. Chromosomes are structures of DNA strands and protein that contain most of the genetic information in a cell. We can visualize chromosomes in metaphase during the cell cycle. Cytogenetics refers to the study of tissue, blood, blood marrow, or culture cells in a laboratory, using banding or manipulating techniques to look for changes in the chromosomes, including broken, missing, rearranged, or extra chromosomes. Changes in the chromosomes may be a sign of a genetic disease or condition. Cytogenetics may be used to help diagnose, plan a treatment, or find out how well a treatment is working.

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## Karyotype

Blood/ Bone marrow

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## Karyotype

Amnion and Chorionic villus sampling (CVS)

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## Fanconi Anemia

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## Fluorescence in situ hybridization (FISH)

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## Sperm DNA Fragmentation Assay (SDFA)

# Test Information

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Test Name	Specimen	Indications
Karyotype	<ul style="list-style-type: none"><li>• <b>Peripheral Blood</b></li><li>• <b>Bone-Marrow</b></li></ul>	<ul style="list-style-type: none"><li>• Checkup</li><li>• Infertility</li><li>• Abortion</li><li>• Cancer</li></ul>
Amniocentesis		<ul style="list-style-type: none"><li>• Down Syndrome</li><li>• Edward Syndrome</li><li>• Pataue Syndrome</li><li>• Chromosomal disorders</li></ul>
Fanconi Anemia	<ul style="list-style-type: none"><li>• Peripheral Blood</li><li>• Bone-Marrow</li></ul>	<ul style="list-style-type: none"><li>• Fanconi Anemia Types</li></ul>
FISH	<ul style="list-style-type: none"><li>• Peripheral Blood</li><li>• Bone-Marrow</li><li>• Tissue Biopsy</li></ul>	<ul style="list-style-type: none"><li>• Translocations on chromosomes in Cancer study</li><li>• Del/Dup</li></ul>
SDFA	Semen	<ul style="list-style-type: none"><li>• Varicocele management</li><li>• Infertility</li><li>• Abortion</li><li>• Cancer</li></ul>

# Karyotype

Karyotype is a technique in which, scientists take a picture of the chromosomes from one cell, cut them out, and arrange them using size, banding pattern, and centromere positions as guides. Karyotype describes the amount of chromosome count and morphology of an organism under the light microscope. The derivation and study of karyotypes is part of cytogenetic studies. In normal diploid organisms, autosomal chromosomes are present in two copies.

Karyotypes can be used for many purposes, such as studies of chromosomal iterations in prenatal diagnostics or tumor studies. Also, to understand cellular function, taxonomic relationships, and providing information about past evolutionary events.

The typical human karyotype contains 22 pairs of autosomal chromosomes and one pair of sex chromosomes.

The most common karyotypes for a female contain two X chromosomes and are denoted for the sex XX.

Males usually have both an X and a Y chromosome, denoted for the sex XY.

A chromosome analysis on bone marrow may be clinically significant to identify and diagnose acquired neoplastic conditions associated with indications such as, cytopenia's, T-cell lymphoma, B-cell lymphoma, multiple myeloma, acute myeloid leukemia, acute lymphocytic lymphoma, chronic myeloid leukemia, chronic lymphocytic leukemia, myeloproliferative disorder and myelodysplastic syndrome. It may also be relevant in cases to confirm or exclude the diagnosis of hematological malignancies based on morphology and track progress of treatment.

Test	Chromosome analysis	Result time
Karyotype Blood	46 chromosomes	10 Day
karyotype Bone -Marrow	46 chromosomes	10 Day

# Amniotic Fluid

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Amniotic Fluid Karyotype test is a laboratory test analysis . that Examining chromosomes through this test allows your doctor to determine whether there are any abnormalities or structural problems within the chromosomes.

When a cell divides, it needs to pass on a complete set of genetic instructions to each new cell it forms.

This test examines these dividing cells. The arrangement of pairs of chromosomes are one the basis of size and appearance. This helps your doctor easily determine if any chromosomes are missing or damaged.

Test Name	Chromosome analysis	Result time
AF Karyotype test Amniocentesis	46 chromosomes	3-4week

# Fanconi Anemia

Fanconi anemia (FA) is a rare inherited condition that affects your bone marrow and many other parts of your body. People with FA have an increased risk of developing blood disorders and some kinds of cancer.

FA also causes physical abnormalities that can affect people's organs and appearances.

Test Name	Chromosome analysis	Result time
Fanconi anemia	46 chromosome	10-14 DAY
FANCA	16q24.3	10-14 DAY
FANCB	Xp22.31	10-14 DAY
FANCC	9q22.3	10-14 DAY
FANCD1	13q12.3	10-14 DAY
FANCD2	3q25.3	10-14 DAY
FANCD3	6p21.3	10-14 DAY
FANCF	11p15	10-14 DAY
FANCG	9p13	10-14 DAY
FANCI	15q25-26	10-14 DAY
FANCL	17q22.3	10-14 DAY
FANCM	2p16.1	10-14 DAY
FANCN	14q21.3	10-14 DAY
FANCO	16p12.1	10-14 DAY
FANCP	17q25.1	10-14 DAY
FANCQ	16p13.3	10-14 DAY
FANCR	16p13.12	10-14 DAY

# Fluorescence in situ hybridization (FISH)

FISH is a molecular cytogenetic technique that uses fluorescent probes that bind to only particular parts of a nucleic acid sequence with a high degree of sequence complementarity, to detect and localize the presence or absence of specific DNA sequences on chromosomes.

Fluorescence microscopy can be used to find out where the fluorescent probe is bound to the chromosomes

Test Name	Chromosome analysis	Result time
Fish for abnormal chromosome	46 chromosomes	10 DAY
Down syndrome	Ch. 21	10 DAY
Edwards Syndrome	Ch. 18	10 DAY
Patau Syndrome	Ch. 13	10 DAY
BCR/ABL1-t(9;22)	9q34.1;22q11.2	10 DAY
Her2	17q12	10 DAY
t(9;11)	9p21;11q23.3	10 DAY
t (12;21)	12p13.2; 21q22.1	10 DAY

# SDFA

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Sperm DNA fragmentation testing involves looking at the ability of a sperm's DNA to withstand fragmentation when placed under stress. Through several decades of research, elevated levels of sperm DNA fragmentation have been associated with lower rates of fertility success (including with IVF) as well as increased chances of pregnancy loss.

Test Name	Result time
SDFA	4 DAY