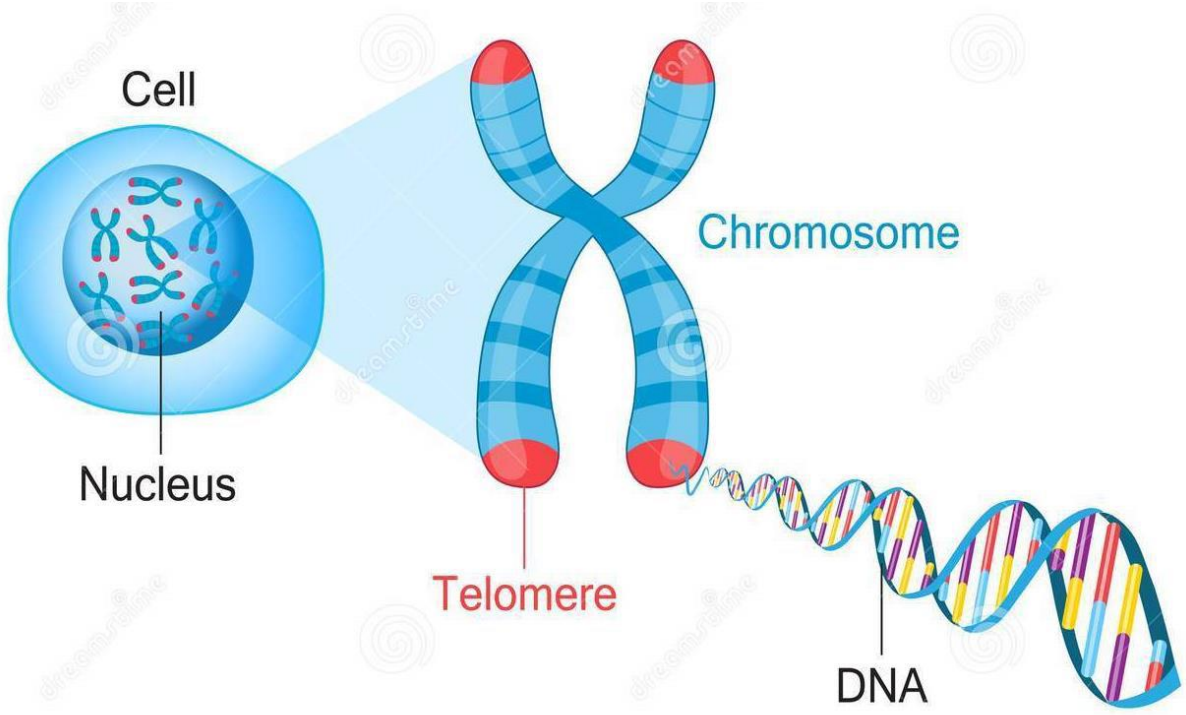
Chromosomal Analysis

What are chromosomes?

In the Nucleus of each cell, the DNA molecule is packaged into thread -like structures called **chromosomes**. Each chromosome is made up of DNA tightly coiled many times around proteins called **histones** that support its structure.

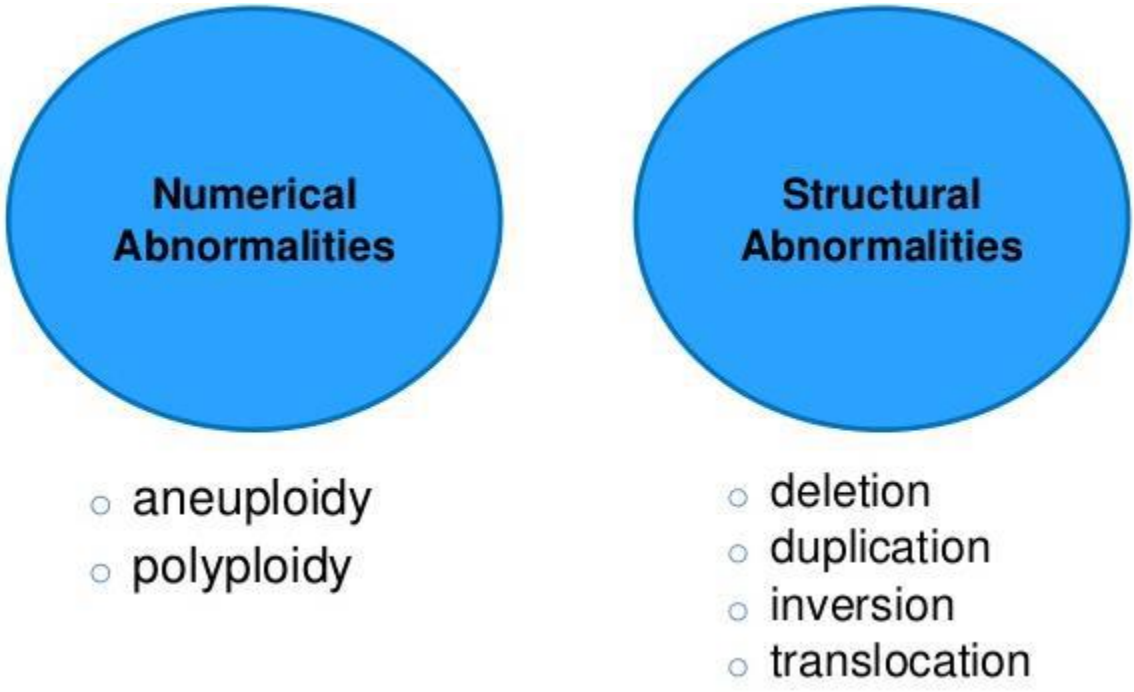


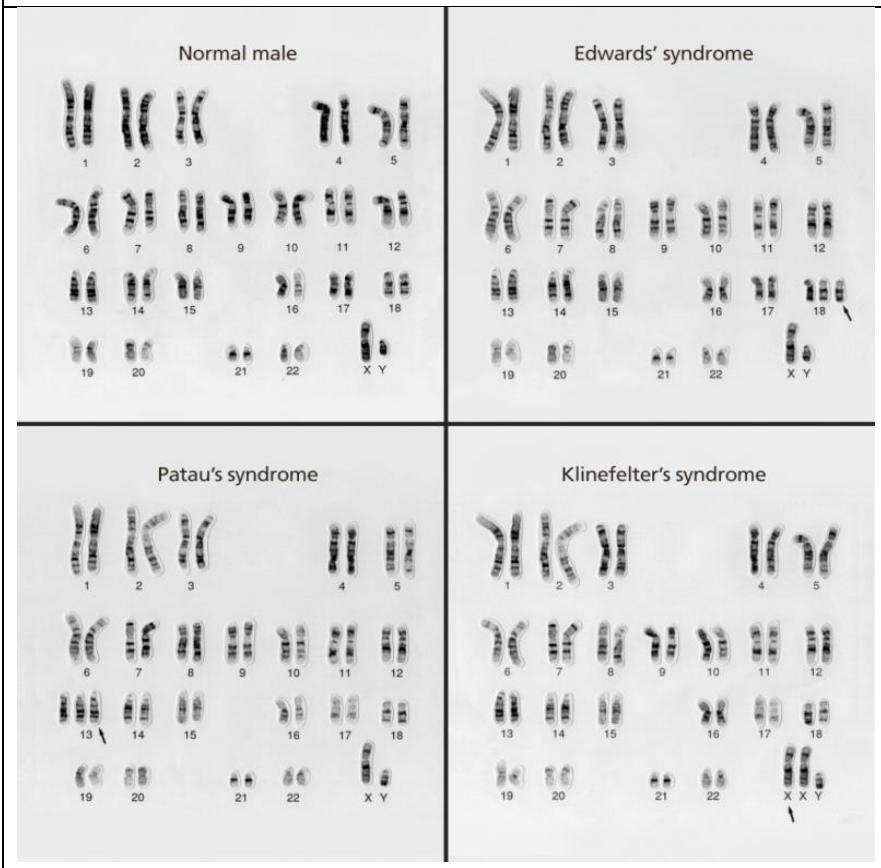
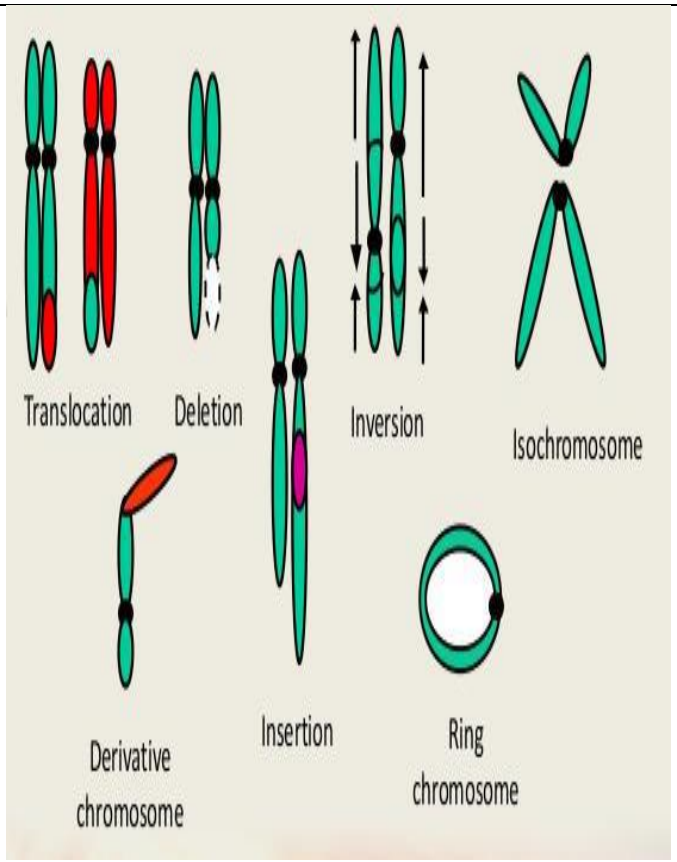
What is Chromosomal Analysis?

Chromosomal analysis is the test that evaluates the number and structure of a person's chromosome to detect abnormalities.

Types of Chromosomal Abnormalities:

A chromosomal abnormality may be numerical or structural and examples are described below.



Numerical Abnormalities	Structural Abnormalities
	

Clinical Utilities:

Chromosomal Breakage Syndromes:

Evaluating patients for chromosome instability syndromes, including ataxia telangiectasia and Nijmegen breakage syndrome.

Genetics Test Information:

- This test is useful for detecting increased spontaneous chromosomal rearrangement in patients with ataxia telangiectasia (AT) or Nijmegen breakage syndrome (NBS), both are chromosomal instability syndromes.
- Chromosomal instability syndromes are characterized by defects in DNA repair mechanisms or genetic instability.
- Patients with these syndromes have an increased risk of developing malignant disorders.

Chromosomal Studies (Fanconi's Anemia):

- Diagnose Fanconi anemia (FA) which is characterized by bone marrow failure, increased risk for cancer, and physical abnormalities.
- Progressive bone marrow failure is responsible for the most significant morbidity and mortality.
- Clinically heterogeneous, FA individuals are at increased risk for acute myelogenous leukemia, myelodysplastic syndrome, and solid tumors of the neck, head, oral cavities, and genitourinary system.

Chromosome Studies (Genetic Disorders):

This test may be done:

- On a couple with Bad Obstetric History (BOH) with history of spontaneous abortions, couples with previous history of malformed child or mental retardation, Couples with Total Infertility (male factor mediated retardation).
- To rule out known syndromes by detecting common aneuploidies in newborn children. Common syndrome includes, Down syndrome (Trisomy 21), Edward syndrome (Trisomy 18), Patau syndrome (Trisomy 13), Turner syndrome (Monosomy X) and Klinefelter syndrome (XXY).
- Children with congenital malformations, ambiguous genitalia and testicular feminization.

Chromosome Analysis (Hematological Malignancy):

- It defines and interpret aberrations that occur in neoplastic cells associated with leukemia, lymphoma and other hematologic malignancies.
- It includes acute and chronic myeloid and lymphoid leukemias, myelodysplastic and myeloproliferative disorders, lymphomas and unexplained anemias may correlate with the diagnosis, prognosis, treatment and etiology of disease.

Chromosome Analysis (POC):

- This test has been done when a more comprehensive assessment for chromosome abnormalities is desired.
- Identification of a chromosome anomaly can provide an explanation for the loss and help to determine risks of recurrence.

Prenatal Chromosomal Microarray Analysis (CMA):

- It can detect genetic abnormalities in a foetus.
- CMA offers additional diagnostic benefits by revealing sub-microscopic imbalances or copy number changes (CNVs) that are too small to be seen on a standard G-banded chromosome preparation.
- Detection of major chromosomal imbalances such as aneuploidy and unbalanced rearrangements.

Y Chromosome Microdeletion:

- Y chromosome infertility is caused by deletions of genes in the AZF (Azoospermia factor) regions. It may remove several genes or in rare case single gene.
- YCM is known to be present in a significant number of men with reduced fertility.
- Diagnosed by extracting DNA from leukocytes in a man's blood sample.

Test Details:

Test Name	Test Code	MRP	TAT / Reported on
Chromosomal microarray test, by blood	SMO10456	21600	28th working day by 7:00 p.m.
Chromosomal Studies (Fanconi's Anaemia)	SCG10008	7900	25 Working Days if received before 1300 hrs.
Chromosome Analysis (Genetic Disorders)	SCG10002	3200	12th Working Day if received before 1300 hrs.
Chromosome Analysis (Haematological Malignancy)	SCG10001	3600	12th Working Day if received before 1300 hrs.
Chromosome Analysis (Married Couple)	SCG10003	6200	12th Working Day if received before 1300 hrs.
Chromosome Analysis (POC)	SCG10004	6000	21 Working Day if received before 1300 hrs.
Prenatal Chromosomal Microarray Analysis (CMA)	SMO10249	36000	21st working day by 7:00 p.m.
Y Chromosome Microdeletion	SCG10012	8500	5th working day if sample received by 2 pm on Day 1