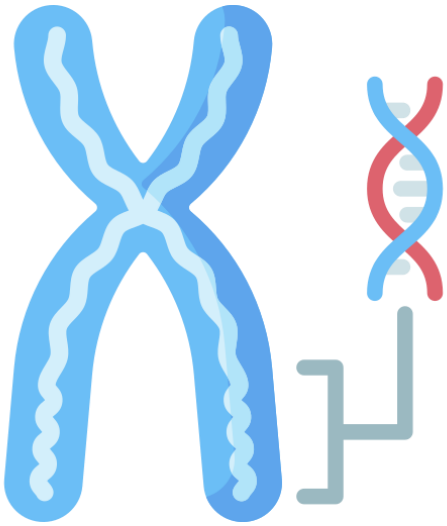
مدينة الملك فهد الطبية
King Fahad Medical City



Cytogenetics Laboratory

Cytogenetics Laboratory

Cytogenetics laboratory aims to test samples of peripheral blood, bone marrow, tissue or amniotic fluid in a laboratory to look for numerical or structural abnormalities of the chromosomes. Changes in certain chromosomes may be a sign of a genetic disease or some types of cancer. Cytogenetics help to diagnose the disease, evaluate prognosis and monitor the effects of treatment and remission in patients.

The Cytogenetics Laboratory testing include:

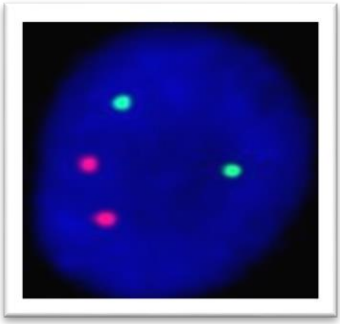
Chromosomal analysis (karyotyping):

It is a test that evaluates the number and structure of chromosome, to diagnose genetic diseases, some birth defects, and certain disorders of the blood and lymphatic system.

Chromosomal analysis
(karyotyping)

Fluorescence in situ hybridization (FISH)

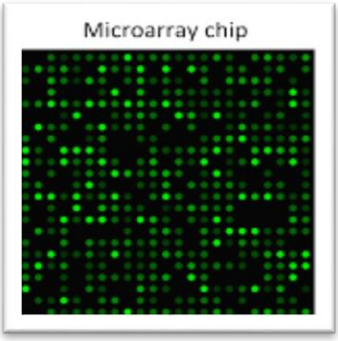
It is a test that enables detecting and locating a specific DNA sequence on a chromosome, using a probe that has a fluorescent molecule attached to it.



Fluorescence in situ hybridization (FISH)

Chromosomal Microarray

A technique that can be used to detect and analyze clinically-significant microdeletions or duplications. Microarray uses a chip with tiny probes consisting of small pieces of DNA from known locations on each of the 46 chromosomes, looking for imbalances between the DNA from a control and the patient's DNA.



Chromosomal Microarray

Cytogenetics testing indications:

Prenatal

- To provide a definitive diagnosis of fetuses who are at risk of chromosomal abnormality.
- Performed in second trimester (15-20 weeks).

Referral reasons:

- Previous pregnancy with chromosomal abnormalities.
- Family history of certain birth defects.
- Advanced maternal age.
- Multiple abnormalities under ultrasound.

Cytogenetics testing indications:

Postnatal

- To study the chromosomes of babies, children or adults with symptoms that may indicate a chromosomal abnormality.

Referral reasons:

- Down syndrome, Edward syndrome, Patau syndrome.
- Klinefelter syndrome.
- Microdeletions syndromes.
- Recurrent miscarriage.
- Ambiguous genitalia.
- Amenorrhea.
- Infertility.

Cytogenetics testing indications:

Hematologic Malignancy

- To identify acquired chromosomal abnormalities that leads to uncontrolled cells division in tissues or organs causing tumors.

Referral reasons:

- Acute myeloid leukemia.
- Chronic myeloid leukemia.
- Acute lymphoblastic leukemia.
- Chronic lymphoblastic leukemia.
- Myelodysplastic syndromes.
- Multiple myeloma.

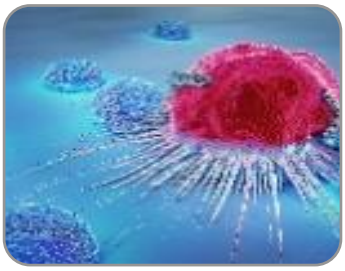
Cytogenetics testing indications:

Oncology

- To identify acquired chromosomal abnormalities that leads to uncontrolled cells division in tissues or organs causing tumor.

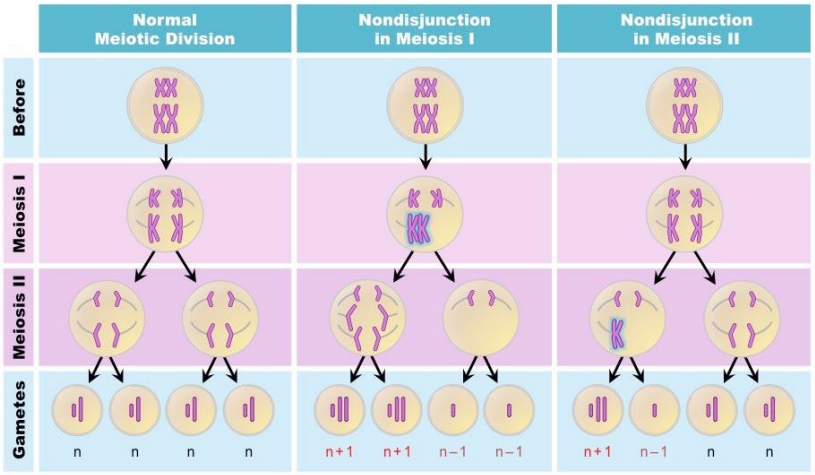
Referral reasons:

- Breast cancer.
- Glioma.
- Sarcoma



Sex Chromosome Abnormalities

Humans are born with 46 chromosomes in 23 pairs. The X and Y chromosomes determine a person's sex. Normal female is 46, XX and normal male is 46, XY. Sex chromosome abnormalities are those caused by numerical or structural alteration in X and Y chromosomes due to an error at some point of the meiosis process during gametogenesis (egg and sperm formation) in the parents.



When to suspect sex chromosome abnormality:

- Recurrent abortions.
- Abnormalities detected through ultrasound.
- Clinical findings in new born (Ambiguous Genitalia).
- Failure of puberty and 2° sexual development (Amenorrhea).
- Infertility.

Most common abnormalities:

- Klinefelter syndrome 47, XXY
- XYY syndrome
- Triple X syndrome 47, XXX
- Turner syndrome 45, X

The main tests used to diagnose these syndromes are **Chromosome analysis (Karyotyping)** and **FISH analysis.**

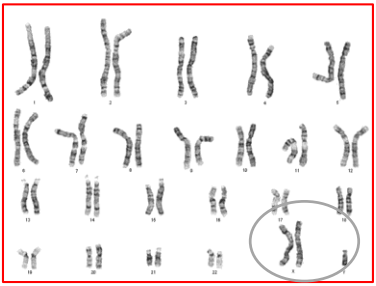
Klinefelter syndrome 47, XXY

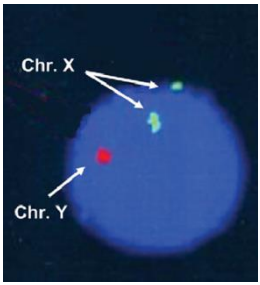
It is a genetic condition that affect males only, caused by a random error in cell division that causes an extra copy of the X chromosome. The additional copy of the X chromosome can occur as a result of either the mother's egg or the father's sperm having an extra X chromosome.

Symptoms:

- Enlarged breast.
- Absence of facial hair.
- Longer legs.
- Poor muscles development.

- Small testicles



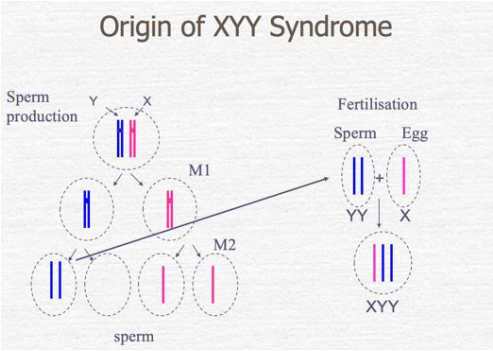


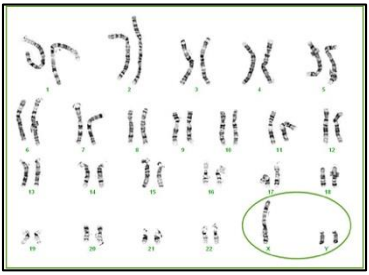
47, XYY syndrome

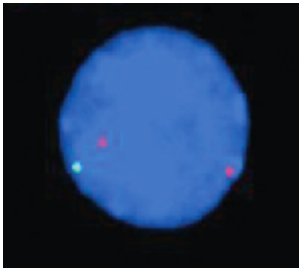
It is a rare chromosomal disorder that affects males only and caused by an extra copy of the Y chromosome due to a cell division error in the sperm.

Symptoms:

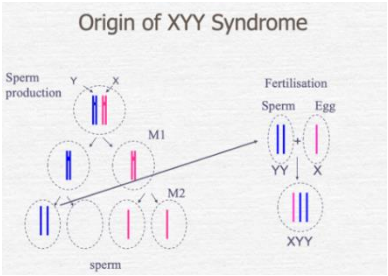
- Most affected males typically have a normal physical appearance.
- Taller than average height.
- Learning disabilities.
- Behavioral problems.







Triple X syndrome 47, XXX

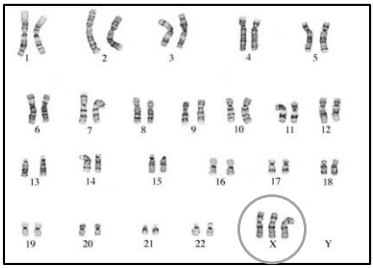


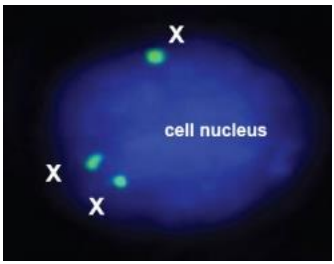
It is a genetic disorder that affects females only. Normally, a female has two X chromosomes,

one from her father and one from her mother. A female with triple X syndrome has three copies of X chromosome due to a random error in cell division.

Symptoms:

- Most affected females typically have a normal physical appearance.
- Taller than average height.
- Learning disabilities.
- Behavioral problems.



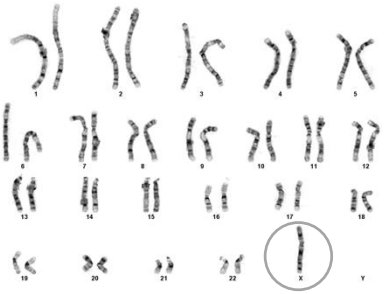


Turner syndrome 45, X

It is a genetic condition that affects only females, caused by the absence of all or part of the second X chromosome in some or all of the cells of the body due to a random error in cell division.

Symptoms:

- Most affected females typically have a normal physical appearance.
- Short stature.
- Shield like chest.
- Webbed neck.



لأن الوعي وقاية ..

إدارة التثقيف الصحي

Clinical Pathology Department

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