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Lab.5
Karyotyping

What is Karyotyping?

Karyotyping is a laboratory procedure that allows your doctor to examine your set of chromosomes. "Karyotype" also refers to the actual collection of chromosomes being examined. Examining chromosomes through karyotyping allows your doctorto determine whether there are any abnormalities or structural problems within the chromosomes.

A karyotype test examines the dividing cells. The pairs of chromosomes are arranged by their size and appearance. This helps doctor easily determine if any chromosomesare missing or damaged.

Why Get Tested?

1. Detect chromosome abnormalities.
2. Help diagnose genetic diseases.
3. Birth defects.

When Get Tested?

1- When pregnancy screening tests are abnormal**.**

2- Indicated to detect chromosomal abnormalities in a person whose detect aspecific.

3- Abnormality in family members.

Sample Required?

1. Blood sample drawn from a vein.
2. Sample of amniotic fluid or chorionic villus from pregnant woman.
3. Bone marrow or tissue sample.

After the sample has been taken, it's placed in a laboratory dish that allows the cells to grow. A lab technician will take cells from the sample and stain them. This makes it

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possible for your doctor to view the chromosomes under a microscope.
These stained cells are examined under a microscope for potential abnormalities. Abnormalities can include:

- Extra chromosomes
- Missing chromosomes
- Missing portions of a chromosome
- Extra portions of a chromosome
- Portions that have broken off of one chromosome and reattached to another

The lab technician can see the chromosomes' shape, size, and number. This information is important in determining if there are any genetic abnormalities.

What is being tested?

Chromosome analysis or karyotyping is a test that evaluates the number and structure of a person's chromosomes in order to detect abnormalities.

Chromosomal abnormalities include both numerical and structural changes.

For numerical changes in complete set of 46 chromosomes represents a change in the amount of genetic material present and can cause health and development problems.

For structural changes, the significance of the problems a problem may vary fromperson to person, even when the same chromosome abnormality is present.

The test is performed by:

- ❖ Taking a sample of a person's cells, culturing them in nutrient-enriched media to promote cell division in vitro. This is done in order to select a specific time during the cells' growth phase when the chromosomes are easiest to distinguish.
- ❖ Isolating the chromosomes from the nucleus of the cells, placing them on a slide, and treating them with a special stain.
- ❖ Taking microphotographs of the chromosomes.
- ❖ The chromosomes arrangeg by size, from largest to smallest, numbers 1 to 22, followed by the sex chromosomes as the 23rd pair.

- The pictures also allow the chromosomes to be vertically oriented. Each chromosome looks like a striped straw. It has two arms that differ in length (a shortarm (p) and a long arm (q)), a pinched-in area between the arms called a centromere,and a series of light and dark horizontal bands. The length of the arms and the location of the bands help determine top from bottom.

Once the chromosome photo arrangement is completed, a laboratory specialist evaluates the chromosome pairs and identifies any abnormalities that may be present.

Some chromosomal disorders that may be detected include:

- Down syndrome (Trisomy 21), caused by an extra chromosome 21; this may occurin all or most cells of the body.
- Edwards syndrome (Trisomy 18), a condition associated with severe mental retardation; caused by an extra chromosome 18.
- Patau syndrome (Trisomy 13), caused by an extra chromosome .13
- Klinefelter syndrome, the most common sex chromosome abnormality in males; caused by an extra X chromosome.
- Turner syndrome, caused by missing one X chromosome in females.
- Chronic myelogenous leukemia, a classic 9;22 translocation that is diagnostic of the disease.

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