

Karyotyping

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Karyotyping is a test using a microscope to identify chromosome abnormalities from malformation or disease. It examines the size, shape, and number of chromosomes in a certain sample of cells. Extra, missing, or abnormal positions of chromosome pieces can cause problems with a person's growth, development, and body functions.

A few of the abnormalities that can be found are Down syndrome, Turner syndrome, Klinefelter's syndrome and the Philadelphia chromosome.

Down syndrome is caused by an extra number 21 chromosome.

Turner syndrome is a defect that results in abnormal female sexual development where only one X chromosome is present.

Klinefelter's syndrome is a disorder that affects male fertility, when at least one extra X chromosome is present.

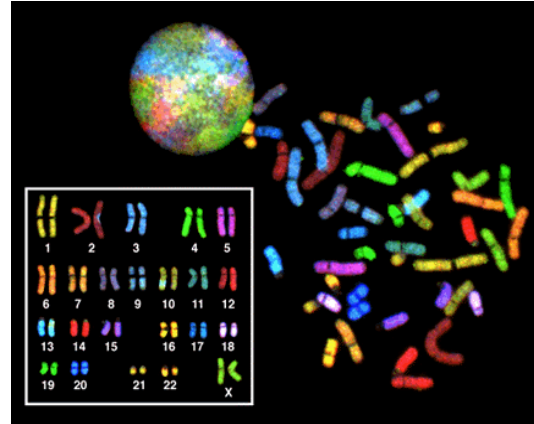
The Philadelphia chromosome is an abnormality in chromosome 22 in which part of chromosome 9 is transferred to it. Bone marrow cells containing this are often found in chronic myelogenous leukemia.

The types of karyotyping are:

- 1) Multicolor Karyotyping (Spectral and Color Changing Karyotyping)
- 2) Fluorescence in Situ Hybridization (FISH)

- 3) Comparative Genomic Hybridization (CGH).

Multicolor karyotyping identifies all chromosomes in a metaphase preparation based on their color differences.



FISH uses fluorescent molecules to vividly paint genes or chromosomes. It is particularly useful for gene mapping and for identifying chromosomal abnormalities and it is one of the few types of karyotyping that can perform on nondividing cells.

Comparative Genomic Hybridization (CGH) is a molecular-cytogenetic method for the analysis of regional changes in the DNA content of tumor cells.

Using epifluorescence microscopy and quantitative image analysis, differences in the fluorescence ratio of the tumor versus that of the control DNA can be detected.

The whole procedure is estimated to be about \$200.

