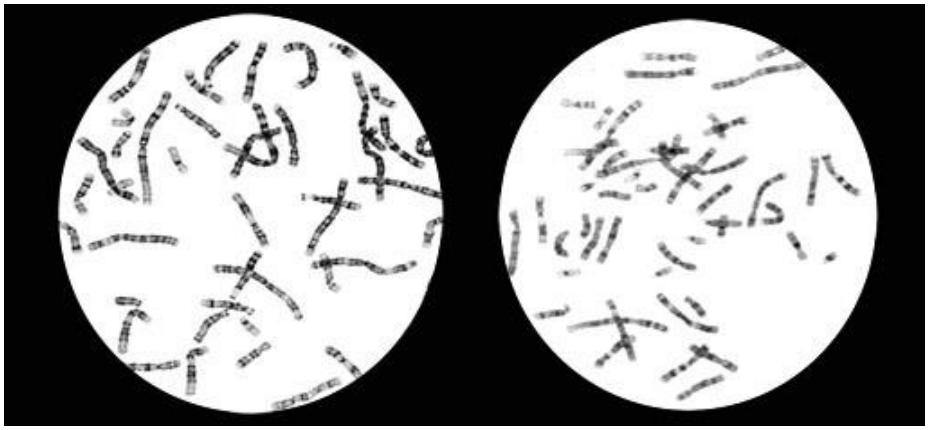
The Royal College of Pathologists
Pathology: the science behind the cure

Object 10: A karyotype



What is it?
A karyotype is a picture of a complete set of chromosomes in a cell. Chromosomes are the structures that contain our DNA, the genetic blueprint that makes us who we are. DNA determines characteristics such as eye colour but can also determine what diseases we develop.

Analysis of a karyotype allows scientists to assess the number, size and structure of chromosomes. Humans usually have 23 pairs of chromosomes, including a pair of sex chromosomes that determine whether we're male or female.

History
The first chromosomes identified were in plant cells, described by Swiss botanist Karl Wilhelm von Nageli in 1842. The human karyotype wasn't completely understood until the 1950s.

Pathology
Geneticists are the pathologists and scientists involved in the preparation and analysis of karyotypes. They interpret the characteristics of the chromosomes to diagnose genetic disorders such as Down's syndrome, in which there is an extra copy of chromosome 21.

Find out more
You can have a go at matching up chromosomes in a karyotype on the Genetics Science Center Website.

To see a sample karyotype result, with an explanation about how it is interpreted, visit The International Mosaic Down Syndrome Association's website.