

NHGRI *fact sheet*

Spectral Karyotyping (SKY)

What is SKY?

Spectral karyotyping (SKY) is a laboratory technique that allows scientists to visualize all 23 pairs of human chromosomes at one time, with each pair of chromosomes painted in a different fluorescent color.

What is SKY used for?

Many diseases are associated with particular chromosomal abnormalities. For example, chromosomes in cancerous cells frequently exhibit aberrations called translocations, where a piece of one chromosome breaks off and attaches to the end of another chromosome. Identifying such chromosome abnormalities and determining their role in disease is an important step in developing new methods for diagnosing many genetic disorders.

Traditional karyotyping allows scientists to view the full set of human chromosomes in black and white, a technique that is useful for observing the number and size of the chromosomes. Scientists, however, cannot accurately identify many translocations or other abnormalities using only a black and white karyotype. But by using SKY, they can easily see instances where a chromosome, painted in one color, has a small piece of a different chromosome, painted in another color, attached to it.

How does SKY work?

SKY involves the preparation of a large collection of short sequences of single-stranded DNA called probes. Each of the individual probes in this DNA library is complementary to a unique region of one chromosome; together, all of the probes make up a network of DNA that is complementary to all of the chromosomes within the human genome.

Each probe is labeled with a fluorescent molecule that corresponds to the chromosome to which it is complementary. For example, probes that are complementary to chromosome 1 are labeled with yellow molecules, while those that are complementary to chromosome 2 are labeled with red molecules, and so on.

When these probes are mixed with the chromosomes from a human cell, the probes hybridize, or bind, to the DNA in the chromosomes. As they hybridize, the fluorescent probes essentially paint the set of chromosomes in a rainbow of colors. Scientists can then use computers to analyze the painted chromosomes to determine whether any of them exhibit translocations or other structural abnormalities.