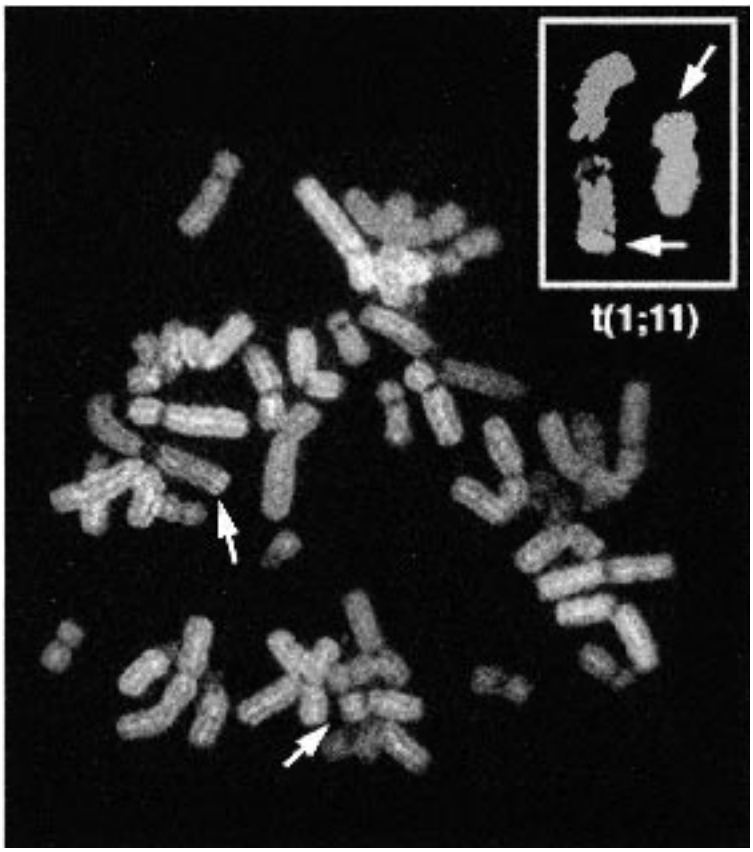
Molecular Biology, VII



Spectral Karyotyping Analysis in Diagnostic Cytogenetics

The technique of spectral karyotyping (SKY) allows the simultaneous visualization of the 24 human chromosomes, each in a different color (see Images in Neuroscience, April 1997 issue of the *Journal*). The combination of conventional fluorescence microscopy, charge-coupled device imaging, and Fourier spectroscopy produces the spectral images. Because the genetic material from each chromosome hybridizes distinctively with the paint probes used in this technique, SKY can be used to study constitutional chromosomal aberrations in the cytogenetics laboratory. The smallest discernible aberration analyzed to date is the t(1;11) (q44; p15.3) translocation shown above; this translocation was unrecognizable by conventional banding analysis. This SKY figure was prepared from the peripheral blood lymphocytes of the father of a mentally retarded child. Seen in the right upper inset, a small amount of genetic material from chromosome 1 (yellow) has been exchanged with material from chromosome 11 (blue). Although balanced in the father, the translocation resulted in transmission of inadequate genetic material to the offspring and caused mental retardation. The size of this chromosomal alteration was estimated to be approximately 1500 kilobase pairs (Kbp). At present, this is the sensitivity limit of the SKY technique using chromosome painting probes.

The SKY technique has obvious diagnostic applications in clinical and cancer cytogenetics. In addition, it should be available for the study of interspecies evolutionary divergence and for the analysis of chromosomal aberrations in animal models of human diseases.

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