Title: Cytogenetics

Abstract:

Cytogenetics is the field of study devoted to chromosomes, and involves the direct observation of a cell’s chromosomal number and structure, together known as its karyotype. Many chromosomal abnormalities are associated with disease. Each chromosome in a karyotype can be stained with a variety of dyes to give unique banding patterns. More recent techniques, including comparative genomic hybridization and fluorescence *in situ* hybridization (FISH), allow for detecting specific chromosomal features or abnormalities.

This video will begin by examining the principles of these classical and modern cytogenetics techniques. This is followed by an examination of a general protocol for performing FISH. Finally, several examples of how karyotyping can be applied to various medical applications are presented.

Application videos:

1. FISH for Pre-implantation Genetic Diagnosis **(2570 Thumbnail @ 5:54 – FISH result images)**

Description: During assisted reproduction, genetic screening can be performed on embryos before their implantation to detect any chromosomal abnormalities. In this article, researchers demonstrate the techniques for performing FISH on single cells, or blastomeres, biopsied from pre-implantation embryos.

2. Chromosomics: Detection of Numerical and Structural Alterations in All 24 Human Chromosomes Simultaneously Using a Novel OctoChrome FISH Assay **(3619 Thumbnail @ 5:13 Karyotyping result)**

Description: This video presents the basic steps for performing FISH-based karyotyping on all 24 unique chromosomes in humans (the 22 autosomes, and the sex-determining X and Y chromosomes), on a single slide in one hybridization step. An 8-chambered OctoChrome device is used, with separate “whole-chromosome paint” probe sets against three chromosomes in each chamber. This device is then combined with a template slide with eight separate sets of metaphase spreads, or condensed chromosomes generated from cell samples, generating eight images with three chromosomes labeled in each.

3. Spectral Karyotyping to Study Chromosome Abnormalities in Humans and Mice with Polycystic Kidney Disease **(3887 Thumbnail @ 12:10 – Spectral Karyotype)**

Description: Here, researchers demonstrate another technique for simultaneously visualizing all chromosomes, called spectral karyotyping or SKY. Each chromosome is labeled with a set of probes with a different combination of fluorescent dyes, which produces a unique spectral color for each chromosome. The stained chromosomes are then analyzed with the SKY View software to generate a multicolor karyotype for analysis. This technique allows for quick determination of chromosomal abnormalities such as translocations.

4. Chromosome Preparation From Cultured Cells **(50203 Thumbnail @ 6:50 – Chromosome spread)**

Description: Giemsa staining, or “G-banding”, highlights AT-rich DNA, typically found in condensed or heterochromatin, which will show up as dark bands in the karyotype. In this video, researchers demonstrate the basic steps for preparing chromosome spreads from cells and performing G-banding.

5. High-throughput Physical Mapping of Chromosomes using Automated in situ Hybridization **(4007 Thumbnail @ 0:48 – Stained polytene chromosome)**

Description: This article presents a protocol for the preparation, automatic staining, and detection of polytene chromosomes --- oversized chromosomes formed when multiple rounds of DNA replication occur without cell division, typically in dipteran insects such as flies and mosquitoes. High pressure is used to generate the chromosome spread from insect tissues, followed by automated FISH staining and imaging with the help of motorized systems and computer software. The benefits of this automation include increased reproducibility of results as well as reduced handling time.

Related Videos

5040 – Introduction to Fluorescence Microscopy

5041 – Introduction to Light Microscopy

5052 – Passaging Cells

5543 – Genetics and Disease