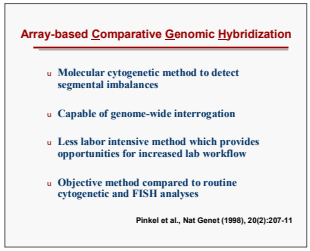
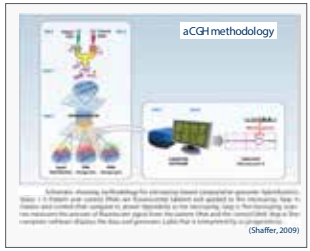
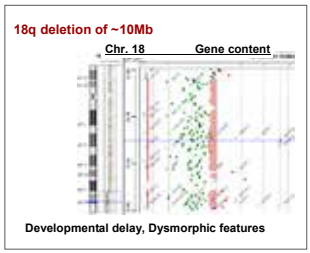
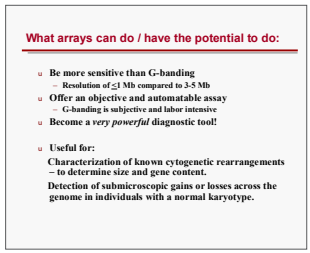
Department of Laboratory, Medicine & Pathology

Cytogenetics: Traditional karyotype to molecular karyotype



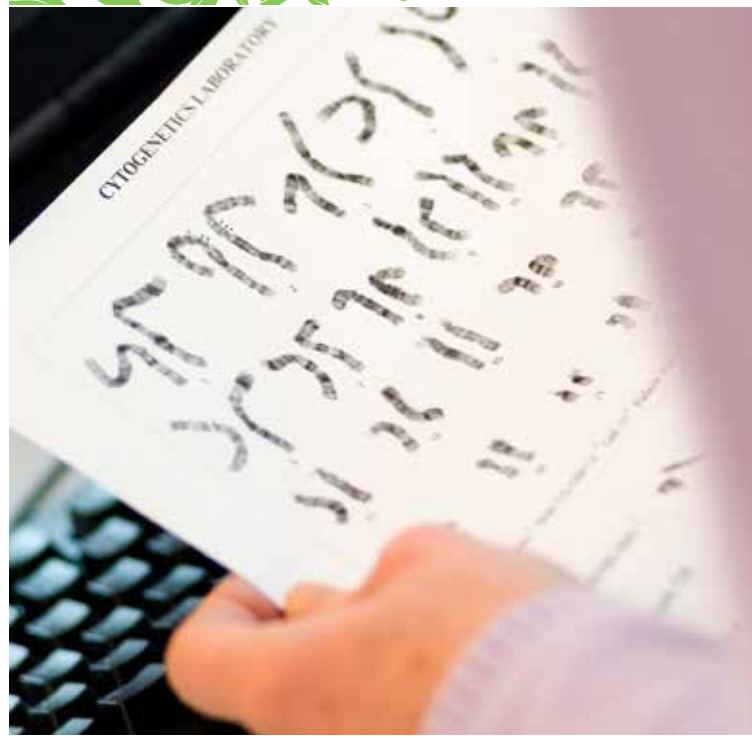






Conclusion:

The aCGH has completely changed the field of cytogenetic diagnostics and allowed for the detection of chromosomal abnormalities at an unprecedented rate. High resolution analysis provided by aCGH may lead to this technology being chosen as a first-line test for chromosomal analysis in the future.



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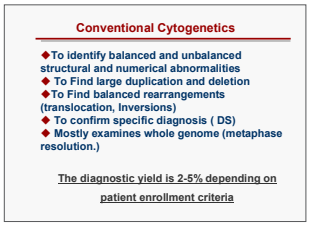
The Cytopathology laboratory

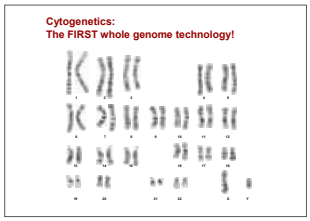
Background:

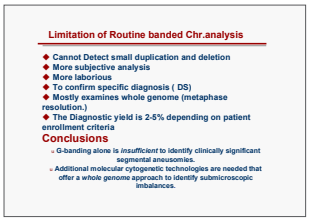
Many genetic conditions have been shown to arise from the dosage imbalance of genomic regions containing one or more genes. These imbalances are caused by the structural rearrangement of genomes as a result of deletions, duplications, translocations and inversions.

Traditional Cytogenetics:

Traditional cytogenetics is one of the greatest innovations of the 20th century. Routine cytogenetic studies can detect only relatively large (>5Mb) rearrangements or numerical changes such as absence or presence of an extra chromosome. The diagnostic yield of cytogenetic analysis is 2-5% depending on the patient recruitment criteria.

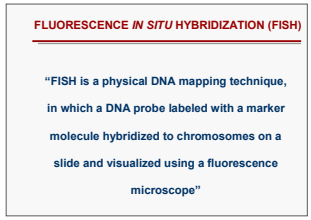


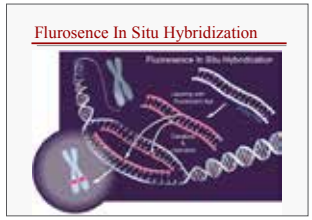


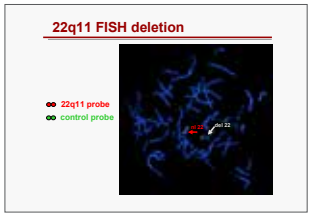


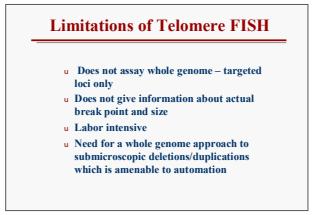
Molecular Cytogenetic 1:

Molecular cytogenetic techniques provide a way to detect complicated, cryptic and submicroscopic rearrangements that remain undetected by conventional cytogenetic studies. Molecular cytogenetic methods increase the level of resolution and the most commonly used method is FISH. FISH diagnostic tests are directed at detecting a specific abnormality in the number or structure of a particular chromosome. FISH testing is mainly used for the detection of known microdeletion syndromes and the screening of subtelomeric abnormalities.









Molecular Cytogenetic 2:

Array-based comparative genomic hybridization (aCGH) has recently become a widely used approach in the genome-wide analysis of chromosomal abnormalities. The aCGH analysis has surpassed traditional chromosome analysis and FISH as the method of choice for identifying chromosome abnormalities.