Evolution of cytogenetic techniques

1956
- Karyotyping
- FISH
- Microarrays
- NGS

20,000 genes
46 chromosomes
23 pairs

Innovation and discovery

1870
- Walther Flemming

1905
- Karl Nägeli

1960
- Edwin Southern

1980
- Joe W. Gray, PhD & Daniel Pinkel, PhD

1990
- Joe Hin Tjio, PhD

1991
- Southern files a UK patent application for DNA "blotting." A basic technique used in Southern blotting to transfer nucleic acid from gel to membrane.

1994
- FISH

1995
- Pinkel and Gray add interphase and metaphase FISH (fluorescence in situ hybridization).

2010
- Schena publishes the first use of microarrays as an expression analysis tool.

Current and future applications

Building on history and forging new paths

Technologies have been applied and advanced for more than a century, helping scientists understand chromosome defects and rearrangements. Their ability to examine genetic material at the nucleotide level has opened a world of exciting possibilities.

Genetic diseases

- Duchenne muscular dystrophy
- Down syndrome
- Fragile X syndrome
- Huntington’s disease
- Cystic fibrosis
- Sickle-cell disease
- Birth defects
- Developmental delays
- Fetal loss
- Cancer

Common genetic disorders

- Cancer predisposition
- Early-onset aging
- Neurodevelopmental disorders
- Cystic fibrosis
- Huntington’s disease

Chromosome research

- Whole-exome sequencing
- Targeted gene panels
- Custom testing options
- Long-read sequencing continue to drive variant discovery in cytogenetic research

Comparative genomic hybridization

- Whole-genome sequencing
- Exome sequencing
- Targeted gene panels

Next-generation sequencing

- Short- and long-read sequencing
- RNA-seq
- Blue-print discovery in the focused exome
- Whole-genome sequencing

High level of data complexity

Microarrays

- Basic techniques
- Customization
- High throughput
- High level of data complexity

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