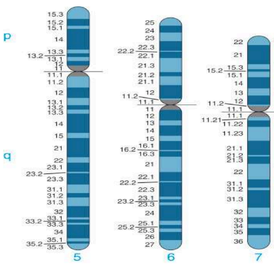
Chromosomal Nomenclature



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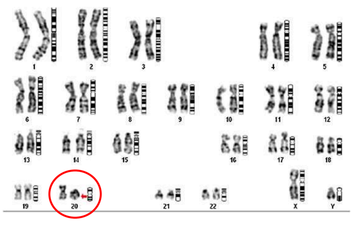
Karyotypes are Best for Identifying Structural Problems



Kalane U, Datar C, Kalane S. Ring chromosome 20 syndrome-a rare chromosomal cause of refractory epilepsy in children. *International Journal of Epilepsy*. 4(2017):87-89.

Fluorescence In Situ Hybridization (FISH)

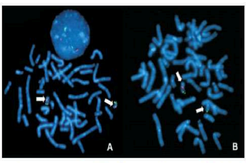
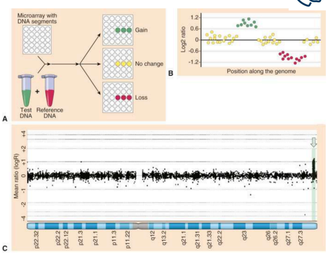


Figure 1 – FISH - metaphase plates showing (A) the expected signs in both chromosomes 22 (normal pattern) and (B) absence of the signal corresponding to region 11.2 of the long arm (q) of chromosome 22, consistent with a 22q11.2 microdeletion (the arrows indicate the chromosomes 22). In (A) note an interphase nucleus with two red signals (normal pattern).

Rosa et al. 2011

Genome Analysis Using Microarray



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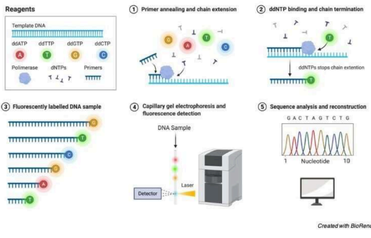
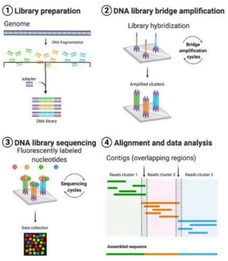
Sanger Sequencing



DNA Sequencing: Definition, Type, Mechanism and Significance (researchtweet.com)

Next Generation Sequencing



DNA Sequencing: Definition, Type, Mechanism and Significance (researchtweet.com)

Next-Gen Sequencing

- NGS often won't identify:
  - Deletions/duplications >100 bp (newer NGS technology is better at this)
    - Specific deletion/duplication testing at a gene level available through MLPA or custom arrays
  - Trinucleotide repeats
    - PCR +Southern blot
  - Imprinting disorders
    - Methylation
  - Whether mutations are in cis or trans
    - Parental samples

Next-Gen Sequencing

- These are the tests that use NGS:
  - Gene panels
  - Exome sequencing
  - Genome sequencing