Medical Genetics 327 Cheat Sheet

by Varda via cheatography.com/165279/cs/37076/

Architecture of Human Genome

DNA - Transcription - RNA - Translation - Protein
23 Pairs, XX - Female XY - Male
mtDNA - Mitochondria DNA, heavy and light strands
Displacement-Loop is a triple-stranded region, due to a short third strand (7s DNA), contains the mtDNA control region
mtDNA - minimum spacers
Nuclear DNA (Chromosomal)
HGP - Human Genome Projects, collaborative research program
Conjoined genes - genes can make both protein-coding mRNA and functional noncoding RNA transcripts
Repetitive DNA sequence - patterns of nucleic acids (DNA or RNA) that occur in multiple copies throughout the genome
Repetitive DNA families : Alu, LINE, Segmental duplication
Cell Division - Mitosis and Meiosis
Ploidy - number of different copies of each chromosome present in a cell
Meeiotic nondisjunction - failed to separate from one another to travel to the opposite poles.

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Clinical Cytogenetics

Clinical Cytogenetics - Practice of medical Genetics by studying the STRUCTURE and NUMBER of chromosomes to identify chromosome abnormalities
Indications for Chromosomal Disorders
- Problems with early growth and development
- stillbirth and neonatal death
- fertility problems
Insertion/Deletions
Translocation - transfer a segment of one chromosome to another chromosome (Robertsonian translocation)
Diagnose for Chromosomal Disorders
Karyotyping
- high-resolution banding (prometaphase) - higher resolution of G or R banding
- G banding - light regions (GC-rich regions) Dark regions (AT- rich regions)
- Ideogram (Computer imaging of G,R,Q, C, banding)
- Q banding - detects Heteromorphism
- R banding - reverse of G and Q banding, analyze the distal ends of chromosomes
- C banding - Centromeric regions, constitutive heterochromatin
Fluorescence In Situ Hybridization
- fluorescent labelled ssDNA probes to hybridize with chromosomes, gene specific or locus-specific probes used to detect chromosomes
SKY - FISH, all chromosomes have a colour
Comparative Genome Hybridization
- determine the copy number differences between two distinct DNA samples - DELETIONS AND DUPLICATIONS that are too small for cytogenetic analysis
DNA Microarray
LARGEST TO SMALLEST - Banding - FISH/SKY - Microarray - Allele-specific oligonucleotide hybridization
Chromosome Abnormalities

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