

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 ${\sf G}$ or ${\sf R}$ 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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