

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
 Meiotic 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

