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Karyotypes

- karyogram: an image of chromosomes arranged in a standard formate
- karyotype: the description of the chromosomes as seen in the karyogram
- karyotyping: the process of determining the karyotype by creating a karyogram
- karyotyping can reveal changes in chromosome number, such as trisomy 21 (Down syndrome).
- karyotypes can indicate chromosomal sex
- karyograms are a source of diagnostic information for specific birth defects, genetic disorders, and cancers.

Karyotyping:

- Sample collection and cell culture
- Inhibition of mitosis with drugs
- Seperate the chromosomes
- Staining
- Photography
- karyogram preparation

Sample collection - white cells from a blood sample or amniotic cells from a developing fetus are ideal.

Inhibition of mitosis with drugs - The cells in the samples are grown in a lab ("cultured") and then treated with drugs which stop the mitotic process at metaphase. Chromosomes become condensed and microscopically visible during mitosis metaphase.

Certain drugs interfere with the spindle fiber apparatus and the chromosomes will remain paired at the metaphase plate; hence, these drugs are used to 'freeze' the chromosomes in place at a time when they are most visible.

These same types of drugs are commonly used as 'chemotherapeutic agents' to stop rapidly-dividing cells characteristic of most cancers.

Seperate the chromosomes - A hypotonic salt solution is added to the cultured cells which causes the cells to take in water and in effect swell up and burst. This process causes the chromosomes to spill out of the cell and seperate from one another.

Staining - Chemicals are used to stain the DNA which makes up most of the chromosome composition; in this way the chromosomes are highly visible under the microscope.

Photography - With a camera fitted to a microscope, photographs are taken of several 'mitotic plates' in which the chromosomes are visible and individually seperated. Enlargments are made of the images, and the chromosomes are digitally cut out.

The individual chromosomes are paired up according to size and banding pattern. The larger chromosomes (#1-5) are arranged on the top row, and in order of descending size, four more rows are constructed with the paired chromosomes. The remaining sex chromosomes (XX or XY) are positioned at the bottom right of the karyotype.

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Unity - on a larger scale unity means that all species on earth use the same genetic code based on two nucleotide pairs and the same set of codons for amino acids. Also, from genetics point of view humans share more similarities than differences.

Diversity - means that regardless of how close we are genetically, there is no 2 individuals that share the same genetic code (except for some archaea and identical twins)

Mitochondrial DNA is ideal for comparing organisms within a species or those who have diverged recently (> 20 million years).

Evidence from mtDNA has been used to divide up the human population into distinct haplogroups.

Humans can trace their origins back to one of seven super-haplogroups originating from an original 'mitochondrial Eve'

Amino acids sequences are also used to see the differences and similarities between species.

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Frameshift mutations

- Caused by insertion or deletion of bases.
- Occurs when the change is not in multiples of three bases.
- Alters the reading frame of codons during translation.
- Results in incorrect amino acids down stream of the mutation.
- Typically leads to nonfunctional prokins due to premature stop codons or misfolding.

Development of HIV

- CCR5 - (C-c chemokine receptor type 5) - used by HIV1 virus as a point of entry to infect CD4 lymphocytes aka Helper T cells.
- Normally CCR5 is a chemokine that help leucocytes to locate the infection.

Delta 32 mutation

CCR5 delta 32 mutation:

- A 32-base pair deletion in the CCR5 gene, causes frameshift and accidentally formed STOP-codon.
- Results in a truncated, non-functional CCR5 receptor on immune cells.
- HIV uses CCR5 as a co-receptor to enter cells; absence of functional CCR5 prevents the virus from infecting these cells.
- Individuals with homozygous delta 32 are resistant to HIV infection.

Location mutations

Germline mutations:

- Occur in gametes (sperm and eggs).
- Heritable; can be passed to offspring.
- Impact future generations.

Somatic Mutations:

- Occur in non-germline cells (body cells).
- Not heritable; affect only the individual.
- Can lead to diseases like cancer.

Coding region mutations:

- Affect the sequence of genes.
- May result in changes to protein structure and function.
- Include missense, nonsense, and silent mutations.

Non-coding region mutations:

- Occur in regulatory regions (promoters, enhancers).
- Can affect gene expression without altering protein coding.
- May influence traits and susceptibility to diseases.

Mutation Hotspots - zones where mutations are more frequent

- **Repetitive sequences**: Areas with microsatellites and minisatellites are prone to slippage during DNA replication, leading to insertions or deletions
- **CpG Dinucleotides**: these are pairs of cytosine and guanine nucleotides linked by a phosphate bond. In the genome, cytosines in CpG sites (or islands) can be methylated, making them more likely to undergo a chemical change (deamination) that converts cytosine to thymine. This process results in a common type of mutation called a transition which can lead to genetic disorders and contribute to cancer development.

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