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Anthropology short notes

Sex chromosomal aberration

UPSC Civil Services (Main) Examination · Anthropology Paper 1 · Anthropology GS 1 (Paper I)

(b) Sex chromosomal aberration-Klinefelter (XXY), Turner (XO), Super female (XXX), intersex and other syndromic disorders.

Short notes by official syllabus heading.

Revision notes for the Anthropology optional. Confirm thinkers and examples against standard texts and recent papers.

<https://wrapexams.com/upsc/archives/anthropology-notes/paper-1/sex-chromosomal-aberrations/>

Sex Chromosomal Aberration

Definition

Understanding evolutionary change through chromosomal studies.

Example / case study

Humans and chimpanzees share a common ancestor with 24 pairs of chromosomes; two fused to form chromosome #2 in humans. Discovered by: Murray F. Lyon (Lyon Hypothesis, 1961). (T: M.F Leone) Refers to the inactivated X chromosome in female somatic cells (random inactivation).

Definition

A karyotype is the complete set of chromosomes of an individual, arranged in homologous pairs according to their size, shape and banding pattern.

Example / case study

Cri-du-chat Syndrome (5p-) - deletion on short arm of chromosome 5. The brain is highly sensitive to gene dosage imbalance.

Thinkers

Turner's Syndrome (XO): Female with only one X → no Barr body.

Definition

A karyotype is the complete set of chromosomes of an individual, arranged in homologous pairs according to their size, shape and banding pattern.

Example / case study

Cri-du-chat Syndrome (5p-) - deletion on short arm of chromosome 5. Severity depends on proportion and distribution of abnormal cells. Mosaic Down Syndrome - some cells 47,+21; others normal 46. Turner mosaic - some cells 45,X; others 46,XX.

Thinkers

Turner's Syndrome (XO): Female with only one X → no Barr body.

Definition

Refers to the inactivated X chromosome in female somatic cells (random inactivation).

Example / case study

Calico Cats: Different coat patches due to random X inactivation. Turner's Syndrome (XO): Female with only one X → no Barr body.

Definition

Diagnosis: Karyotype (47,XXX); two Barr bodies.

Example / case study

Cri-du-chat Syndrome (5p-) - deletion on short arm of chromosome 5. Baby after birth cry like cat, microcephaly, wide-set eyes, low-set ears. Intellectual disability, delayed development.

Definition

Understanding evolutionary change through chromosomal studies.

Example / case study

Humans and chimpanzees share a common ancestor with 24 pairs of chromosomes; two fused to form chromosome #2 in humans. Discovered by: Murray F. Lyon (Lyon Hypothesis, 1961). (T: M.F Leone) Refers to the inactivated X chromosome in female somatic cells (random inactivation).