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Anthropology short notes

Autosomal aberrations

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

(c) Autosomal aberrations-Down syndrome, Patau, Edward and Cri-du-chat syndromes.

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/mains-answers/anthropology-gs-1/autosomal-aberrations/>

Autosomal Aberrations

Definition

$2n + 1 \rightarrow$ Trisomy, $2n - 1 \rightarrow$ Monosomy.

Example / case study

Cri-du-chat Syndrome (5p-) - deletion on short arm of chromosome 5. □ (2021) "Chromosomal aberrations can play havoc with the human body and mind." Explain.

Definition

Risk increases with maternal age.

Example / case study

Mosaic Down Syndrome - some cells 47,+21; others normal 46. Turner mosaic - some cells 45,X; others 46,XX. □ (2021) "Chromosomal aberrations can play havoc with the human body and mind." Explain. Chromosomal aberrations disturb the normal gene dosage and gene regulation, thereby affecting multiple organ systems simultaneously. Because many genes are involved, their impact on the human body and mind can be profound.

Definition

Micrognathia (small jaw), clenched fists with overlapping fingers.

Definition

PTC Tasting: The ability to taste the bitter chemical Phenylthiocarbamide is a dominant trait (TT, Tt).

Example / case study

A study of a local tribe that ignores the impact of global capitalism or state-led industrialization (such as the mining in the DRC or phosphate excavation on Banaba Island) fails to capture the true reality of that culture's survival.

Thinkers

Bronislaw Malinowski | Fieldwork; Cultural Anthropology Franz Boas | Cultural Anthropology; Historical Particularism Benjamin Lee Whorf | Linguistic Relativity Individual as Active Agent: Scholars like Alexander Goldenweiser and Edward Sapir argued that culture is not an abstract force independent of humans. By assuming a closed system, anthropologists overlook individual biographies and the "personal particularity" that creative individuals inject into the cultural stream to drive innovation.

Definition

Entire sets of chromosomes gained or lost.

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.

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