Q8(b)

Describe the causes of structural abnormalities of chromosomes with suitable examples.

UPSC Civil Services Mains 2023 · Anthropology GS 1 · 15 marks · Numerical and structural aberrations (disorders).

(a) Numerical and structural aberrations (disorders).

Core demand of the question

- Describe the causes of structural abnormalities of chromosomes with suitable examples
- Write at 15-mark length

Revision summary

Structural abnormalities rearrange chromosome parts after breakage or unequal crossing-over.

Types are deletion, duplication, inversion, translocation, ring, and isochromosome.

Cri-du-chat is 5p deletion; 22q11 is a common microdeletion.

Philadelphia t(9;22) is a somatic translocation in CML.

Robertsonian 14;21 can cause translocation Down syndrome.

Balanced parental carriers explain some recurrent unbalanced offspring.

Introduction

Structural chromosomal abnormalities are changes in the shape and arrangement of chromosomes, not in the simple count of 46. They arise when breakage and repair go wrong in meiosis or mitosis, and the clinical picture depends on how much DNA is lost, gained, or moved.

Body

Causes at the chromosome

- A **break** in the DNA-protein chromosome, from errors of recombination, radiation, some chemicals, or fragile sites, is the starting event.
- **Repair** may join the wrong ends, producing deletions, duplications, inversions, translocations, rings, or isochromosomes.
- Unequal crossing-over between repeated sequences is a common meiotic cause of deletion and duplication.
- **Robertsonian** fusion joins two acrocentric long arms, which is a structural change that also alters count.
- Parental balanced rearrangements can become unbalanced gametes, which is why family karyotypes matter in counselling.

Main types with examples

• **Deletion:** loss of a segment. **Cri-du-chat** is a 5p deletion with a characteristic cry and developmental delay. **22q11** deletion (DiGeorge / velocardiofacial) is a common microdeletion with heart and immune findings.

• **Duplication:** extra segment, sometimes the partner of a deletion from unequal crossing-over. Charcot-Marie-Tooth type 1A is a teaching duplication of a myelin gene region on chromosome 17.

• **Inversion:** a reversed segment. Pericentric inversions can be balanced in a parent and still produce unbalanced recombinant children.

• **Translocation:** exchange between chromosomes. The **Philadelphia chromosome** t(9;22) fuses BCR-ABL in chronic myeloid leukaemia, a somatic structural change. Reciprocal translocations in the germline can be balanced in a parent and unbalanced in a child.

• **Robertsonian translocation** between 14 and 21 is a cause of translocation **Down syndrome**, distinct from free trisomy 21, which is numerical.

• **Ring chromosome** forms when two ends of one chromosome fuse after terminal breaks.

• **Isochromosome** is a mirror chromosome with two identical arms, seen in some **Turner** variants involving X.

Anthropological and clinical note

• These are cytogenetic facts used in medical anthropology of disability, stigma, and prenatal testing, not racial traits.

• Mosaicism can blunt or patch the phenotype when the rearrangement is post-zygotic.

Flow diagram

Chromosome break -> Deletion

Chromosome break -> Duplication

Chromosome break -> Inversion

Chromosome break -> Translocation

Translocation -> Philadelphia CML

Deletion -> Cri-du-chat 5p

Conclusion

Structural abnormalities come from breakage, unequal recombination, and mis-repair, yielding deletions, duplications, inversions, translocations, rings, and isochromosomes. Cri-du-chat, 22q11, Philadelphia t(9;22), and 14;21 Robertsonian Down syndrome are the standard examples.

Facts & figures

Cri-du-chat

A deletion of the short arm of chromosome 5, named for the infant cry and associated developmental findings.

Philadelphia chromosome

t(9;22) creating a BCR-ABL fusion, the classic structural change of chronic myeloid leukaemia.

Robertsonian translocation

Fusion of acrocentric long arms; 14;21 translocation is a cause of translocation Down syndrome.

22q11 deletion

A frequent microdeletion syndrome with cardiac, palatal, and immune features.

Unequal crossing-over

A meiotic mechanism that produces complementary deletions and duplications at repeated sequences.