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GENETIC DISORDERS, DEVELOPMENTAL ANOMALIES AND MONSTERS

- Genetics
  - Chromosomes
- Genetic disorders
  - Aberrations in chromosomes
- Anomalies
- Monsters
- Model Questions
GENETICS
Genetics is the branch of science deals with study of genes, chromosomes and transmittance of characters from one to another generation.

CHROMOSOMES
Chromosomes are thread like structures present in the form of short pieces in nucleus of a cell. They are in pairs; of which one pair is sex chromosome and others are autosomes.

Table 3.1: Number of chromosomes in different species of animals

<table>
<thead>
<tr>
<th>Sl. No.</th>
<th>Animal</th>
<th>Chromosomes Pairs</th>
<th>Total</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Cattle</td>
<td>30</td>
<td>60</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>2.</td>
<td>Sheep</td>
<td>27</td>
<td>54</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>3.</td>
<td>Goat</td>
<td>30</td>
<td>60</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>4.</td>
<td>Pig</td>
<td>19</td>
<td>38</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>5.</td>
<td>Dog</td>
<td>39</td>
<td>78</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>6.</td>
<td>Cat</td>
<td>19</td>
<td>38</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>7.</td>
<td>Horse</td>
<td>32</td>
<td>64</td>
<td>XY</td>
<td>XX</td>
</tr>
<tr>
<td>8.</td>
<td>Poultry</td>
<td>39</td>
<td>78</td>
<td>ZZ</td>
<td>ZW</td>
</tr>
</tbody>
</table>

- Each chromosome is composed of two chromatids connected at centromere.
- Chromosomes are grouped together on the basis of their length, location of centromere and this procedure is known as Karyotyping.
- The study of Karyotyping is known as cytogenetics.
- Chromosomes are composed of 3 components:
  - DNA - 20%
  - RNA - 10%
  - Nuclear Proteins - 70%

Deoxyribo nucleic acid (DNA)
- Double helix structure of polynucleotide chain.
- A nucleotide consists of phosphate, sugar and base of either purine (Adenine, Guanine) or pyrimidine (Thymine, Cytosine).

- A sequence of 3 nucleotide determines the synthesis of an amino acid and is known as genetic code/codon.
- During cell division, one half of DNA molecule acts as template for the synthesis of other half by an enzyme DNA polymerase to transmit the genetic information which may also have some disorders to next progeny.

Gene
- Sequence of nucleotides which controls the synthesis of one specific protein is known as gene. It is a unit of function. Study of genes is termed as Genetics. In higher animals about 1.0 million genes are present.
- Genes located on X or Y chromosomes are termed as sex linked and all other genes are autosomal genes.
- When the genes at one locus are same from both parents they are termed as homozygous but when they are different at one locus they are known as heterozygous.
- In heterozygous, characters of one gene are manifested in phenotype and such gene is known as dominant while unexpressed gene is called as recessive.
Karyotyping
- Karyotyping is the study of chromosomes in cell.
- Collection of blood, separation of lymphocytes using Histopaque-1077 gradient.
- The lymphocytes are cultured with mitogen concanavalin A (ConA) or phytohemagglutinin - M (PHA-M) for 72 hrs.
- Colchicine is used after 72hrs to arrest the cell division at metaphase stage.
- Hypotonic solution is added to allow cells for swelling which causes separation of chromosomes.
- Prepare glass slides and stain with Giemsa or other special stain.
- Identify the chromosomes and photograph them.
- Cut photographs having homologous chromosomes and make pairs.

GENETIC DISORDERS
ABERRATION IN CHROMOSOMES
- A large number of chromosomal aberrations are removed due to death of gamete or zygote which is termed as “species cleansing effect”. Even though some aberrations do persist and expressed in phenotype leading to illness.

1. Aberration in number
- Chromosomes are in pairs (2n). When number of chromosomes are other than (n) or (2n). It is known as heteroploidy.

(a) Heteroploidy
- The number of chromosomes are other than (n) or (2n).
- When abnormal number is exact multiples of the haploid set due to errors in mitosis. The polar body may fail to be extruded from ovum leaving diploid set to be fertilized by sperm (n) i.e. 2n + n = 3n (Triploid zygote).
- When abnormal number is not the exact multiples of haploid set. It may have specific chromosome in triple number (trisomy) or in single number (monosomy).

(b) Duplication and deficiencies
- Duplication or deficiency may occur in a section of chromosome and total number of chromosomes remains same.
- Translocation is the rearrangement of a part of chromosome in two non-homologous chromosomes. It may be reciprocal or non-reciprocal. Absence of a piece of chromosome is known as deletion.

(c) Mosaicism
- In mosaicism, there is more than one population of cells in body; each population differs in their chromosomes/ genes due to error during development.
- May be due to chromosomal non-disjunction e.g. XXY in some cells, XY in other cells.

(d) Chimerism
- In this, one type of cells are acquired in utero from a twin e.g. Bovine twin 1 male and 1 female with joint placenta. The blood cells of male may go in female counterpart. Then the female will have two type of cell population, one of its own and another acquired from twin. Similarly, male may also have XX leucocytes in its blood. Such chimeric bulls are sterile.

2. Abnormalities in sex chromosomes
(a) Klinefelters syndrome
- Male have sex chromatin i.e. XXY = 47(2n) in man.
- In some cells, different number of chromosomes i.e. XX, XXY, XXXY, XXXYY
- It is recognized in adolescence by small testes, tall body, and low sexual characters, mostly infertile.
- May occur in sheep, cattle and horse.

(b) Tortoiseshell male cat
- Male cat has small testes, lack of libido and absence of spermatozoa in testes with 3n chromosomes (XXY).
(c) **Turner’s syndrome**
- Mare are with XO karyotype having gonadal dysgenesis and such animals are sterile and do not have sex chromatin.
- In mice XO karyotype is normal.

(d) **Intersexes**
- In this condition ambiguity occurs in genitalia or the secondary sex characters are present for both the sexes including male and female.
- Hermaphrodites have male and female genitalia while pseudohermaphrodites are having external genitalia of one sex and gonads of opposite sex.

(e) **Freemartinism**
- In bovine twins, one male with (XY) and one female (XX) karyotype but they share placental circulation so cells of embryo establish in other co-twin.

(f) **Testicular feminization**
- Animal is having female genitalia as external and internal organs but in place of ovaries, there are testes. It occurs due to single gene defect and makes tissues unresponsive to androgenic hormones.

3. **Abnormalities in autosomal chromosomes**
   (a) **Down’s syndrome/ Mongolism**
   - It occurs as a result of trisomy, number of a particular chromosome increases leaving 2n, as 61 in bovines, 77 in dogs and 47 in man e.g. bovine lymphosarcoma occurs in animals with 2n=61. Male dog with 2n= 77 are prone to lymphoma.

   (b) **Sterility in hybrids**
   - Donkey has 2n=62 and horse has 2n=64. Their cross mule has 2n=63.
   - Sterility in mules, cause is not known, may be due to uneven number of chromosomes.

4. **Abnormalities in genes**
- Lethal genes are those genes, which are responsible for death of zygote.
- Sublethal genes
- X-linked or sex linked: Diseases transmitted by heterozygous carrier females only to male offsprings who are homozygous for X-chromosome.

**ANOMALIES**
Anomaly is a developmental abnormality occurs in any organ/tissue. It may be due to genetic disorder and may affect the zygote itself within few days after fertilization or may occur during any stage of pregnancy. It may be classified as under:

1. **Imperfect development**
   (a) **Agenesis**
   Agenesis is incomplete development of an organ or mostly it is associated with absence of any organ.
   - Acrania is absence of cranium.
   - Anencephalia is absence of brain.
   - Hemicrania is absence of half of head.
   - Agnathia is absence of lower jaw.
   - Anophathalmia is absence of one or both eyes.
   - Abrachia is absence of fore limbs.
   - Abrachiocephalia is absence of forelimbs and head.
   - Adactylia is absence of digits.
   - Atresia is absence of normal opening e.g. Atresia ani is absence of anus opening.

   (b) **Fissures**
   Fissures are a cleft or narrow opening in an organ on the median line of head, thorax and abdomen.
   - Cranioschisis is a cleft in skull.
   - Chelioschisis is a cleft in lips also known as harelip.
   - Palatoschisis is a cleft in palates; also known as cleft palate.
   - Rachischisis is a cleft in spinal column.
   - Schistothorax is a fissure in thorax.
   - Schistosomus is a fissure in abdomen.

   (c) **Fusion**
   Fusion is joining of paired organs.
   - Cyclopia is fusion of eyes.
• **Renarcuatus** is fusion of kidneys; also known as horse shoe kidneys.

2. Excess of development
- Congenital hypertrophy of any organ.
- Increase in the number of any organ or part/tissue.
  - **Polyotia** is increased number of ears.
  - **Polypodontia** is increased number of teeth.
  - **Polymelia** is increased number of limbs.
  - **Polydactyly** is increased number of digits.
  - **Polymastia** is increased number of mammary gland.
  - **Polythelia** is increased number of teats.

3. Displacement during development
   (a) **Displacement of organ**
   - **Dextrocardia** is the transposition of heart into right side instead of left side of thoracic cavity.
   - **Ectopia cordis** is the displacement of heart into neck.

   (b) **Displacement of tissues**
   - **Teratoma** is a tumor arising due to some embryonic defect and composed of two or more types of tissues. In this at least two tissues should be of origin.
   - **Dermoid cyst** is a mass containing skin, hair, feathers or teeth depending on the species and often arranged as cyst. It mostly occurs in the subcutaneous tissues.

**MONSTERS**

Monster is a disturbance of development in several organs and causes distortion of the foetus *e.g.* Duplication of all or most of the organs (Fig. 3.3).
- Monsters develop from a single ovum; these are the product of incomplete twinning.
- Monsters are classified as under:

1. **Separate twins**
   One twin is well developed while another is malformed and lack the heart, lungs or trunk, head, limbs.

   ![Fig.3.3. Photograph showing monster calf.](image)

2. **United twins**
   These twins are united with symmetrical development and are further classified as:

   (a) **Anterior twinning**
   Anterior portion of foetus is having double structures while posterior remains as single.
   - **Pyopagus** is a monster twin united in the pelvic region with the bodies side by side.
   - **Ischiopagus** is a monster twin united in the pelvic region with the bodies at more than a right angle.
   - **Dicephalus** is a monster having two separate heads, neck, thorax, and trunk.
   - **Diprosopus** is a monster having double organs in cephalic region without complete separation of heads and with double face.

   (b) **Posterior twinning**
   When in monsters, the anterior portion remains single and posterior parts become double.
   - **Craniopagus** is a monster having separate brain with separate bodies arranged at an acute angle.
   - **Cephalothoracopagus** is the monster having united head and thorax.
   - **Dipygus** is the monster having double posterior extremities and posterior parts of body.

   (c) **Almost complete twining**
   In monster, twins are having complete development with joining in thorax and abdomen.
• **Thoracopagus** is a monster united in thorax region.
• **Prosopothoracopagus** is the monster twin united at thorax, head, neck and abdomen.

• **Rachipagus** is the monster in which thoracic and lumbar portion of vertebral column are united in twin.

**MODEL QUESTIONS**

**Q. 1. Fill in the blanks with suitable word(s).**
1. Chromosomes are grouped together on the basis of ……………. and ………………. and this procedures is known as ………………...
2. ………………. is the rearrangement of a part of chromosome in the non-homologous chromosomes and it may be ………………. or ………………. 
3. Acrania is absence of ………………. while ………………. is absence of forelimbs.
4. ………………. is absence of normal opening; for example, ………………. is absence of anus opening.
5. ………………. is a fissure in lips which is also known as ………………..
6. Palatoschisis is a ………………. in palates and is also known as ………………. 
7. ………………. is transposition of heart into ………………. of thoracic cavity.
8. Monsters develop from ………………. and are the products of ………………. twinning.
9. ………………. is a monster united in the pelvic region with the bodies side by side.
10. ………………. is fusion of kidneys and is also known as ………………..

**Q. 2. Write true or false against each statement. Correct the false statement.**
1. ……………..Hemicrania is absence of head.
2. …………….Polyotia is decreased number of ears.
3. …………….Each chromosome contains about 70% DNA.
4. …………….Monsters develop from a single ovum.
5. …………….Abrachiocephalia is a absence of forelimbs and head.
6. …………….Chromosomes are thread like structures, composed to two chromatids connected with a centromere.
7. …………….Dipygus is a monster having double anterior extremities and other parts of body.
8. …………….Schistosomus is a fissure in spinal column.
9. …………….Dicephalus is a monster having two separate head, neck, thorax and trunk.
10. …………….Prosopothoracopagus is a monster, which is not united at head.

**Q. 3. Write short notes on the following.**
1. Draw a diagram of DNA structure
2. Karyotyping
3. Freemartin
4. Anomalies
5. Monsters
6. Dermoid cyst
7. Teratoma
8. Aberration in chromosomes
9. Testicular feminization
10. Mosaicism

**Q. 4. Define the followings with suitable examples.**
1. Cytogenetics
2. Heteroploidy
3. Agnathia
4. Chromosome
5. Monosomy
6. Polysomy
7. Telocentric
8. Metacentric
9. Acrocentric
10. Carcinogenesis
11. Thoracopagus
12. Abrachia
13. Renarcuatus
5. Cyclopia     15. Rachischisis
7. Ischiopagus  17. Chimerism
8. Ectopia cordis 18. Rachipagus

Q. 5. Each question is provided with four options. Select most appropriate option to fill in or answer the question.

1. Each chromosome contains the DNA content as……………….
   (a) 20%  (b) 10%  (c) 70%  (d) 30%
2. The study of karyotyping of chromosomes falls under ……………
   (a) Immunogenetics (b) Cytogenetics (c) Molecular genetics (d) Nuclear genetics.
3. In heterozygous, one gene character is manifested in phenotype and such gene is called as…
   (a) Autosomal (b) Recessive (c) Dominant (d) Sex linked
4. In karyotyping, colchicine is added in culture of peripheral blood lymphocytes for arresting the cell division in ……………
   (a) Telophase (b) Meiosis (c) Anaphase (d) Metaphase
5. In heteroploidy, the chromosome number will be ……………in cells.
   (a) n  (b) 2n  (c) 3n  (d) All of them
6. Intersexes is the condition in animals which occurs due to ambiguity in…………..
   (a) Genitalia (b) Bones (c) Ears (d) Eyes
7. In Turner’s syndrome, mare is having karyotype as………………
   (a) XX  (b) XXX  (c) XXXX  (d) XO
8. Mules are having chromosome number as………
   (a) 61  (b) 62  (c) 63  (d) 64
9. Bovine lymphosarcoma occurs in animals having chromosome number……
   (a) 60  (b) 61  (c) 62  (d) 64.
10. Dogs are more prone to lymphoma with chromosome number………
    (a) 76  (b) 78  (c) 77  (d) 75
11. Absence of lower jaw in foetus is known as…………
    (a) Acrania  (b) Adactylia  (c) Agnathia  (d) Abrachia
12. Rachischisis is a cleft in ……………
    (a) Spinal column (b) Abdomen (c) Skull (d) Lips
13. Harelip is due to fissure in lips and also known as…………
    (a) Palatoschisis (b) Cranioschisis (c) Schistosomus (d) Cheilioschisis
14. Fusion of eyes occurs in monsters and is known as………………
    (a) Renarcuatus (b) Columbia (c) Cyclopia (d) Anophthalmia
15. Increased number of limbs in monsters is known as ……………
    (a) Polythelia (b) Polymastia (c) Polymelia (d) Polydactyly
16. Dextrocardia is transposition of heart in…………
    (a) Right thorax (b) Left thorax (c) Neck (d) Abdomen
17. Tumor arising from embryonic defect and composed of more than two tissue……
    (a) Dermatoma (b) Hematoma (c) Papilloma (d) Teratoma

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18. A monster having two separate brains with bodies separately arranged at an acute angle……
   (a) Cephalothoracopagus  (b) Dicephalus  (c) Craniopagus  (d) Cranioschisis
19. A monster united at thorax region and with complete development as twin is known as………
   (a) Prosopothoracopagus  (b) Thoracopagus  (c) Dipygus  (d) Cephalothoracopagus
20. A monster having thorax and lumbar portion of vertebral column united in twin is known as…..
   (a) Rachipagus  (b) Craniopagus  (c) Thoracopagus  (d) Dipagus