**Review Worksheet Answers: Chromosomal Mutations**

1: List and describe the types of mutation that can occur to chromosomes

(5 marks)

*Deletion: Part of the chromosome is lost*

*Duplication: Part of the chromosome is doubled (occurs twice)*

*Inversion: Part of the chromosome is flipped*

*Translocation: Part of the chromosome breaks off and joins to another chromosome*

*Non-disjunction: A whole chromosome is added or absent (aneuploidy)*

2: What are the differences between a deletion that occurs as part of a gene mutation, and a deletion that occurs as part of a chromosomal mutation?

(6 marks)

*Deletion as part of a gene mutation involves removal of one base (1) in the sequence that forms the gene. It results in frame shift (1) and a change in the protein produced by that gene.(1)*

*Deletion as part of a chromosomal mutation involves part of a chromosome being lost during meiosis (1). Many genes are lost (1) and the production of many proteins (1) is affected.*

3: Fill in the following table:

(10 marks)

|  |  |  |
| --- | --- | --- |
| **Syndrome** | **Chromosomal abnormality** | **Features** |
| Down Syndrome | *Trisomy 21* | *Characteristic facial features*  *Low Muscle tone*  *Intellectual Disability*  *Heart Defects* |
| Patau Syndrome | *Trisomy 13* | *Wide range of severe defects to face, heart, body systems and brain. Death in utero or just after birth* |
| Klinefelter’s Syndrome | *Extra X Chromosome (XXY)* | *Male genitalia, with female hormonal development eg gynecomastia, feminine body shape* |
| Cri-du-chat Syndrome | *Missing part of chromosome 5* | *Cat like cry*  *Cleft palate*  *Intellectual disability*  *Fused/Webbed digits*  *Developmental delay* |
| Turner’s Syndrome | *Missing Y chromosome (XO)* | *Short stature*  *Lack secondary sex characteristics*  *Infertile*  *Fold of skin between shoulders and neck* |



4: Fill in the blank cells to identify the lobes and areas of the cerebrum

(10 marks)

|  |  |  |
| --- | --- | --- |
| **Function** | **Lobe** | **Area** |
| Thinking, problem solving, emotions, personality, language, motor control | *Frontal lobe* | *Frontal Association Area* |
| Receiving information about temperature, touch, pain and taste | *Parietal lobe* | *Primary Sensory Area* |
| Processing sensory information | *Parietal lobe* | *Sensory Association Area* |
| Receiving Visual Information | *Occipital lobe* | *Primary Visual Area* |
| Processing Visual Information | *Occipital lobe* | *Visual Association Area* |
| Receiving Auditory Information | *Temporal lobe* | *Primary Auditory Area* |
| Processing Auditory Information | *Temporal lobe* | *Auditory Association Area* |
| Initiating Motor Movement | *Parietal Lobe* | *Primary Motor Area* |
| Processing Motor Movement | *Parietal Lobe* | *Motor Association Area* |
| Receiving Olfactory information | *Frontal lobe* | *Primary Olfactory Area* |

5: Label the following areas on the diagram of the cerebrum below