**KARYOTYPING**

Karyotyping refers to a procedure conducted in the laboratory to sort and identify chromosomes in an attempt to learn more about one’s chromosomes and to know someone was born with any type of syndrome (Anon, 2018). Standardized staining procedures are used during the preparation of the karyotypes, and these make it possible for the chromosomes to show their unique structural features. Human beings have 23 pairs of chromosomes. These chromosomes can be seen with the help of a light microscope. The easiest stage of the cell cycle to distinguish and examine a chromosome is during the metaphase stage because the chromosomes are in their maximum condensed stage (Nlm.nih.gov, 2018). During a karyotype analysis, Giemsa dye is used to stain the areas of the chromosomes that have a lot of Adenine (A) and Thymine (T), and the result is a dark band. The thinnest band has more than one million base pairs and could have zero to hundreds of genes. Three things are usually analysed during Karyotyping, and these are: comparing the lengths of the chromosomes, where the centromeres are placed and where G-bands are located with their sizes so each chromosome can be matched with its pair. The primary goal of this work is to use case histories of three patients to come up with completed karyotypes.

**Patient 1**

I would diagnose patient 1 with Down syndrome which is also referred to as trisomy 21

**The phenotype, molecular aetiology**

Notation 47, XX, +21 is used to identify patient one which means that the patient being diagnosed is a female, and she has 47 chromosomes. The extra chromosome is 21. An average person usually has two copies of the chromosome 21 but this patient with Down syndrome has three copies of the chromosomes. Down syndrome refers to a medical condition whereby an individual has an additional chromosome (Nlm.nih.gov, 2018).An average person has forty-six chromosomes while someone with Down syndrome has forty-seven chromosomes. The extra chromosome that such person has is the chromosome 21, and this is what gives them the Down syndrome (Nlm.nih.gov, 2018).  The diagnosis and different components were arrived at through karyotyping. The additional chromosome 21 change the way his brain and body develop. That is why babies with this syndrome face some mental and physical difficulties, and example is learning disability. All people with Down syndrome have different abilities even though their appearance and behaviour might appear similar. The intelligence quotient (IQ) of individuals with Down syndrome typically falls between mild to moderate (Anon, 2018). These children usually take longer to speak compared with the rest of the children.

**Clinical picture**

Symptoms of Down syndrome vary from one person to another. The physical symptoms include poor muscle tone, small ears, heads and mouth, flattened facial profile, short hands with short fingers, and slanting eyes with skin fold from upper eyelid (Anon, 2018). Others include intellectual and developmental symptoms that present with cognitive impairment, short attention span, slow learning and delayed speech or language.

**Clinical outcome**

Children diagnosed with Down syndrome are at risk of developing different types of leukaemia, congenital heart diseases and Hirschsprung’s disease.

**Prognosis**

According to prognosis of Down syndrome, the life expectancy of people with Down syndrome has improved. The average lifespan of a person with Down syndrome was nine years but currently they can live to fifty years and beyond.

**Patient 2**

Patient 2 would be diagnosed with Klinefelter’s Syndrome (KS) which is responsible for his infertility

**The phenotype, molecular aetiology**

The syndrome occurs due to random error that caused male to be born with extra sex chromosomes. The condition is not inherited. The patient has an additional X sex chromosome. The notation 47, XXY suits patient 2. This notation is an indication that the patient is male and he has 47 chromosomes whereby the additional one is a sex chromosome. Klinefelter’s Syndrome is the most common form of X chromosome aneuploidy syndrome that most males are diagnosed with. One of the symptoms that frequently accompany Klinefelter’s Syndrome is infertility which can be seen in the case of patient 2 (Nlm.nih.gov, 2018). Other symptoms of this condition include less muscle tone, reduced energy, smaller testes and penis and the shoulders may become narrower, and the hips widen. These signs are usually noticed in boys with Klinefelter’s Syndrome after they have passed the stage of puberty. Although KS is not curable, some treatments can help to manage the syndrome (Anon, 2018).That is why patients are advised to get started on treatment the earliest time possible.

**Clinical picture**

The clinical features of Klinefelter’s Syndrome include delayed or incomplete puberty, taller than normal stature, less muscle and less facial hair compared with other teens after puberty, shorter torso, longer legs and broader hips compared to other boys, small penis, small but firm testicles and gynecomastia.

**Clinical outcome**

Patients diagnosed with the syndrome are at risk of weak bones, infertility issues, depression, anxiety, heart diseases, lung diseases, breast cancer, and autoimmune diseases like rheumatoid arthritis and endocrine conditions like diabetes.

**Prognosis**

The syndrome is a result of problems with chromosomes, which means there is no cure for the disease. The prognosis is good as long as the patient has consistent monitoring of their health and treatment. People with the syndrome can live a normal lifespan.

**Patient 3**

The diagnosis of patient 3 was Trisomy 13 syndrome. Another name for Trisomy 13 syndrome is Patau’s syndrome.

**The phenotype, molecular aetiology**

Trisomy is caused when there is a defect in fertilization where the egg and the sperm contain twenty chromosomes and an error occurs during fertilization. The best notation for patient 3 was 47, XY, +13. This notation means that patient 3 was a male and he had 47 chromosomes and among these chromosomes was an additional chromosome 13. The complications associated with having an additional chromosome 13 could have led to the patient developing Varadi-Papp Syndrome (Nlm.nih.gov, 2018).A Cleft lip is one of the signs of the Varadi-Papp Syndrome and patient 3 had it. Research shows that boys have higher chances of being born with Trisomy 13 syndrome compared to girls. Further research indicates that approximately 80% to 90% of children born with the Patau syndrome die a few months after their birth. Out of every 10,000 children born, 2 of them have Trisomy 13 syndrome (Nlm.nih.gov, 2018).There are no specific people who are susceptible to giving birth to children with Patau’s syndrome. However, older mothers have a higher possibility of their children being born with this syndrome (Anon, 2018).

**Clinical picture**

Patients with trisomy present with small head and sloping forehead as babies. The nose appears bulbous, eye defects occur, ears with low set and unusual shape. Babies with trisomy have structural brain problems. Some have a sac attached to the abdomen around the umbilical cord. Girls may present with abnormal uterus and boys have testis that may fail to descend.

**Prognosis**

Approximately fifty percent of babies with trisomy survive beyond twelve days and about twenty percent survive on their first year. Predicting the life expectancy is difficult if there are no immediate life threatening issues. Approximately thirteen percent of babies born with trisomy survive until age 10 (Anon, 2018).

**Bibliography**

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