Yu Tun Ng
BIO 2450L
Professor Michael Gotesman
February 14th, 2019

Lab Report 1: Cytogenetics and Karyotyping

Abstract: Karyotyping is a type of technique used to analyze and identify any chromosomal abnormalities during the development of a fetus. A karyotype case study was given to detect whether the fetus was carrying certain genetic diseases or traits. As a result, the karyotype verified that the fetus is under normal conditions and showed no signs of abnormalities.

Introduction: A karyotype is an organized representation on the number of chromosomes presented in an individual. These chromosomes are extracted from the individual's cells, stained, and photographed under the microscope for genetic analysis. Doctors and scientists often use karyotyping to diagnose any genetic disorder that may be passed down from the parents to their offspring. Karyotyping is a common way for parents to check for congenital anomaly that may be detected during their fetal development. Examples of some congenital abnormalities include, Down syndrome, Turner syndrome, and Klinefelter syndrome.

Case Study ID K presented 23-year old female, Sherry, that is on her first pregnancy. She was scheduled for a 20-week ultrasound to ensure that her fetus' heartbeat and development was normal. Upon her ultrasound, the technician finds a minor heart malformation, and a cleft palate. The patient was warned and was referred for karyotyping. The type and number of chromosomes were examined.

Methods: A blue color-coded Chromoscan Board containing a case study was given. The board included a set of colored chromosome decals. The chromosome decals are first randomly selected to make a sketch of the chromosome on the assigned Cytogenetics Report. The sketched chromosome was examined to determine what type of chromosome is presented, either as metacentric, submetacentric, acrocentric, or telocentric. Secondly, a karyotype was created using the decals given and then correctly matched with the chromosomes that are already presented on the board. Lastly, the number of chromosomes were observed to check whether there are any abnormalities or not. The patient's diagnosis can be identified based on the observation of the karyotype and the information given from the case study,

Results: Based on the karyotype, the fetus presented two complete set of 23 chromosomes, 22 of which are autosomes and 2 of which are sex chromosomes. In this case, the fetus presented XY chromosomes for males. The chromosomes are arranged from largest to smallest and shape. There were no abnormal chromosomal findings. Therefore, the fetus is normal.

Discussion: An amniocentesis was performed, and amniotic fluid was extracted from around the fetus of the pregnant woman. This fluid contains amniocytes, in which was used to examine and develop the karyotype. Based on the karyotype, the type of chromosome that is present in the fetus is metacentric. Metacentric chromosomes have equal lengths and their centromere is located near the middle. All these chromosomal characteristics are within normal. Thus, no genetic disease or trait was found in the fetus.

References

1. Brooker, R. J. (2016). *Concepts of genetics*. New York, NY: McGraw Hill Education.