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Lab Report: Cytogenetics and Karyotyping

Abstract:

The purpose of this lab was to understand chromosomal abnormalities through the study of cytogenetics and karyotyping. Cytogenetics is the study of chromosomes in regard to their structure and function. Karyotyping is a specific technique used to visualize stained chromosomes during metaphase stage (*Blair, C., Genetics Lab Manual, p.15*). For this lab the role of cytogeneticist was assigned to each student with a case study in order to diagnose or determine any abnormalities for each patient using the karyotyping technique. In conclusion, the diagnosis for case study H was identified as Klinefelter syndrome.

Introduction:

Chromosomes are the genetic composition of all organisms. However, this lab focuses on eukaryotic nuclear chromosomes specifically in human somatic cells. Chromosomes are composed of DNA and proteins and form a threadlike structure. 46 chromosomes are responsible for the makeup of human cells; 23 maternal and 23 paternal chromosomes such making humans diploid. Chromosomes are responsible in the determination of different species. In humans' chromosomes arrange from largest to smallest. Cytogenetics is the study of chromosomes using microscopes. Karyotyping is a technique cytogeneticist use to study a set of chromosomes during the metaphase stage with the use of a light microscope to detect any structural or numerical abnormalities. Some structural changes a cytogeneticist might observe are deletion, duplication, inversion, insertion and translocation. Some numerical abnormalities can include the loss or gain of one (euploidy) or a whole set (monoploidy) of chromosomes. Numerical changes (aneuploidy) can occur due to non-disjunctions which refer to the failure of separation of sister chromatids or of homologous chromosomes. This lab analyzes the numerical and structural changes in each case in order to detect the abnormalities and diagnose each case.

Methods:

A case study was assigned with chromosome decals. Each chromosome was analyzed and paired with the corresponding chromosome on chromoscan board presented. After each homologous chromosome were paired, any numerical abnormalities or structural changes were reported.

Results:

Figure 1 demonstrates an extra X chromosome on the 23rd homologous pair. Thus, this patient has two X chromosome and one Y chromosome. Concluding that this patient has Klinefelter syndrome (KS). KS is one of the most common causes of male infertility due to chromosomal changes. *(Hawksworth,D.J, Szaran, A.,(2018)…*)

Discussion:

In conclusion after analyzing case study H and the set of chromosomes an extra X chromosome was present thus giving a total of 47 chromosomes. Klinefelter Syndrome is one of the common aneuploidy errors known in infertile men due to its characteristics of one or more X chromosome *(Hawksworth, D.J.(2018)...).* Along with infertility some other consequences of KS is cognitive impairment, enlarged boobs (gynecomastia) and small testes (Hawksworth, D.J., 2018). Based on Santos medical history and the findings presented one can conclude that KS is the proper diagnosis. Karyotyping can help determine genetic disorders such as in Santos case where some disorders go undiagnosed most of the time.

References:

Blair, Christopher, (2018). Bio 2450L Genetics Laboratory Manual., pg. 15-22

Hawksworth, D. J., Szafran, A. A., Jordan, P. W., Dobs, A. S., & Herati, A. S. (2018). Infertility in Patients With Klinefelter Syndrome: Optimal Timing for Sperm and Testicular Tissue Cryopreservation. *Reviews in urology*, *20*(2), 56-62.