The Use of Karyotypes to Investigate Genetic Disorders

Question
How can a karyotype be used to identify chromosomal abnormalities in humans?

Background
Karyotyping is the way geneticists identify, organize, and study human chromosomes. Cells from tissue are made to reproduce in a culture and then chemically treated to stop mitosis at metaphase, when the chromosomes can be spread easily for observation.

The cells are stained for examination under a microscope, and the chromosomes are photographed. The photo is enlarged and the individual chromosomes are cut out. Normally, there are 22 pairs of autosomes and one pair of sex chromosomes. They are divided into seven groups according to their length, the position of their centromeres, and their banding patterns.

The distinctive banding patterns created after the chromosomes have been stained are used to organize the chromosomes into homologous, or matching, pairs. This arrangement of chromosomes is called a karyotype. In this lab, you will study a karyotype and analyze the information it provides.

Procedure
1. Study the normal karyotype of human chromosomes in figure 1. Notice that the chromosomes are arranged in pairs, which are numbered. Observe all characteristics of each pair. Note whether all the pairs match.
2. Figure 2 shows a chart of human chromosomes from a person with a genetic disorder. Using the photocopy provided, cut out the chromosomes. Arrange the chromosomes in matching pairs using figure 1 as a guide.
3. Make a karyotype by gluing or taping the chromosomes onto a piece of blank paper. Compare the karyotype you made with the karyotype shown in Figure 1.

Analyses and Conclusions (answer in complete sentences)
1. Is the karyotype in Figure 1 from a male or a female? The karyotype you constructed? Explain your answers.
2. Explain why you think banding patterns might be an important way to make sure that two chromosomes are truly homologous.
3. How does the karyotype you made differ from the karyotype in Figure 1? Can you identify the disorder?
Diseases Caused by Abnormal Number of Chromosomes

Down Syndrome (trisomy 21)
A person with Down syndrome has 3 of chromosome #21. People with Down syndrome have distinct facial characteristics, short stature, heart defects, susceptibility to respiratory infection, and mental retardation. Although people with Down syndrome, on average, have a lifespan much shorter than normal, some live to middle age and beyond. Down syndrome strikes 0.04% of children born to women under age 30. The risk climbs to 1.25% for mothers in their early thirties and is even higher for older mothers. Because of this relatively high risk, pregnant women who are over 35 are candidates for amniocentesis in order to check for trisomy 21.

Patau Syndrome (trisomy 13)
Patau syndrome, caused by having 3 of chromosome #13, is characterized by serious eye, brain, and circulatory defects, as well as harelip and cleft palate. Patau syndrome occurs once every 5,000 live births. Most victims survive less than a year.

Edwards Syndrome (trisomy 18)
Edwards syndrome, caused by having 3 of chromosome #18, affects almost every organ system in the body. It occurs about once in every 10,000 live births. Most victims survive less than a year.