Name _____ Date _____

Using Karyotypes to Determine Genetic Disorders

Background Information

A normal set of human chromosomes contains twenty-three **pairs**. Strictly speaking, the twenty-third pair in males is not really a pair, as it is made up of one X-chromosome and one Y-chromosome. In females, the twenty-third pair is made up of two X-chromosomes and is therefore a homologous pair. The X and Y chromosomes in humans are called the "sex chromosomes" as they are responsible for determining the sexual morphology of an individual. People sometimes think that having an extra chromosome or two would result in a superhuman. This is not the case, however. Extra chromosomes, damaged chromosomes, and missing chromosomes almost always cause severe abnormalities or disorders, some of which are so detrimental that they will cause complete arrest of embryo/ fetal growth. Studies of chromosomes are made by enlarging photographs of an individual's chromosomes. The chromosome images are cut out of the picture and one by one they are matched in pairs and glued into place by an expert. The image this process creates is called a **karyotype**.

Directions

1. Complete the table below by identifying the affected chromosome, the change in the chromosome(s), the symptoms of the disorder, who is affected, and your source of information.
2. You may use any available resource to research each disorder. The National Institute of Health online could be extremely helpful. The website is www.nih.gov Cite every source you use.
3. Identify the disorder that corresponds to each karyotype on the attached karyotype page. Write the name of the disorder in the space below the karyotype.

Note: The first disorder has been filled in for you as an example.

Disorder	**Chromo-some(s) Affected**	**Change in chromosomes**	**Symptoms of Disorder**	**Males, Females, or Both**	**Source(s)**
Down's Syndrome (Trisomy 21)	#21	An extra chromosome #21, so there are 3 of them instead of the normal 2	Slowed physical and mental development, decreased muscle tone, smaller/ more rounded features	Both	National Institute of Health online www.nih.gov
Turner's Syndrome					

Disorder	Chromo-some(s) Affected	Change in chromosomes	Symptoms of Syndrome	Males, Females, or Both	Source(s)
Cri du Chat Syndrome (Cry of the Cat)					
Klinefelter's Syndrome					
Edward's Syndrome (Trisomy 18)					
Patau Syndrome (Trisomy 13)					