Genetic Counselor Analysis Questions

1. What is the gender of the patient?
2. Are there any abnormal chromosomes?
3. What is the reason for the abnormality? If there is no abnormality, what is the reason for everything being normal?
4. Will there be a normal or abnormal phenotype?
5. Is the child expected to be healthy or unhealthy?
6. Are there indications in the parent's karyotypes that are seen in their child's karyotype?
7. Based on your analysis, write a letter to the parents with your findings. Be sure to include:
    a. Whether there are abnormalities or not
    b. What the abnormalities mean/the disorder
    c. Recommendations for the parents based on the analysis (ie, therapy for the child, expected health of the child, etc.)

Sample Genetic Disorders:

Turner's Syndrome (X0 Disorder)

- Results from having only one X chromosome
- Affects 1 in 5,000 females
- Individuals will most often have a learning disability but is usually not intellectually disabled
- Physical features include shorter than average, swelling hands/feet, may also have health problems such as heart disease and misshaped kidneys
- Many females do not undergo puberty unless given hormones and most are infertile.

Down's Syndrome (Trisomy 21)

- Results from having an extra 21st chromosome
- Large variation in severity of associated problems (very mild to very severe)
- All individuals have some form of intellectual disability (ranges from mild to severe)
- Can have heart problems, hearing loss, feeding issues
- With modern medicine, can live into their 50's and 60's; elderly show easier susceptibility to acquiring Alzheimer's Disease

Robertsonian Translocation

- Results when a chromosome, usually 13, 14, 20, or 21 attaches to another chromosome
- Depending on the translocation, the fetus will either miscarry or live to be pretty healthy or normal.
- All of the genetic material is available, and in the right amounts, but the translocation may result in the material not being able to be read properly.