

Genetic Counseling

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Introduction

Genetic Counseling is a growing career within healthcare. It is a fascinating blend between medicine and psychology.

Genetic Counselors must be up to date on the newest research surrounding their specific field of study. They are experts on genetic conditions and serve as a valuable resource to physicians and other healthcare providers. Genetic Counselors work in several different areas within the healthcare. This includes cardiology, obstetrics, pediatrics, neurological, etc.

This semester, I had the opportunity to work with a board-certified genetic counselor, Jessica Hidalgo. She specializes in obstetrics and works in a Maternal Fetal Medicine office through Intermountain Healthcare.

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Responsibilities:

- Research the condition indicated by the ultrasound and/or referring physician
- Take a family history and make a pedigree chart
- Explain the Invitae test as well as the potential condition and what a negative/positive result means
- Call patient when results are returned

Figures 1 & 2–



Conditions seen:

- MTHFR gene mutation
- Eye problems, abnormal blood clotting, skeletal abnormalities, learning problems
- Turner Syndrome
- One X chromosome is fully or partially missing
 - Results in short height, failure of ovaries to develop, heart defects, High levels of Alpha-Fetoprotein (AFP)
 - Increased risk of fetus having a neural tube defect (e.g., Spina bifida or anencephaly)
- Joubert Syndrome
- Brain malformation caused by the absence of underdevelopment of the cerebella vermis and malformed brain stem
- Dandy-Walker Syndrome
- Development delay, ataxia, enlarged head circumference, spasticity and hypotonia
- Trisomy 21 (Down Syndrome)
- Extra copy of chromosome 21
 - Distinctive facial features, intellectual disabilities, heart and kidney issues
- Prader-Willi Syndrome
- Caused by the loss of function of genes on chromosome 15
 - Results in hypotonia, feeding difficulties, poor growth, and delayed development
- Angel Man Syndrome
- Caused by the loss of function of genes on chromosome 15
 - Results in hyperactivity, seizures, small head size and jerky movements

Figure 3– Invitae Test



An Invitae test is used as a first step to check for genetic anomalies.

Conclusions

Through my semester internship, I have been able to learn much more about the growing field of Genetic Counseling. I was able to shadow a wide variety of genetic counselors from different fields, and with that, an array of medical cases.

I have learned what I like about the career, and things that I did not enjoy as much. I think it was a very valuable experience, considering it is one of my top choices for a future career.

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