Magee-Womens
Research Institute

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Disorders in Sex Development Study

Goal: The goal of this research study is to investigate possible genetic causes of disorders in sexual development (DSD).

Study Overview: Disorders in sexual development are a complex group of congenital conditions where chromosomal, gonadal or anatomical sex is atypical. These conditions could exhibit a variety of phenotypes. DSD is caused by a range of genetic etiologies including chromosome abnormalities or sex chromosome mosaicism and gonadal dysgenesis in patients with normal 46,XY or 46,XX karyotype with changes in the genes, responsible for gonadal development and function. By further studying these conditions we can better understand the molecular causes of DSD. This research study is being conducted by the University of Pittsburgh.

Qualifications for the Study:

- A clinical diagnosis of a disorder in sex development of unknown cause
- Parents of the diagnosed patient can also be included

What will take place in the study?

Once you have consented to be included in the research you will be asked to provide a blood sample. This blood sample will be approximately 10 cc's (1-2 purple top tubes) and will be used to obtain DNA and RNA. At the time of collection we will also obtain a brief family history and a family pedigree.

Outcomes:

Your participation in this research does not have a direct benefit to you. Your sample will hopefully help us to further investigate disorders in sex development and help patients in the future with possible causes.

Study Coordinator Contact Info:

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