Genetic Services at ETSU Medical Genetics (Partial List)

- Genetic evaluation/consultation by a board-certified clinical geneticist (pediatrics and adults)
- Diagnostic Genetic testing including Chromosome Microarray analysis and molecular studies
- Genetic counseling/recommendations for management, and regular follow-up of patients with genetic conditions

PEDIATRICS
- Multiple Congenital Anomalies
- Dysmorphic Features
- Intellectual Disability
- Behavioral Disorders/ADHD
- Autism Spectrum Disorder
- Abnormal Growth & Development

ADULTS
- Intellectual Disability
- Behavioral Disorders
- Family History of Inherited Disorders
- Short/Tall Stature

CANCER GENETICS
- Personal and/or Family History of Cancer: Breast, Ovarian, Colon Cancers, etc.
- Syndromic Cancers: Multiple Endocrine Neoplasia, Macrocephaly-Overgrowth Syndromes, etc.

PRENATAL GENETICS
- Pre-test and post-test counseling for:
  - Maternal serum screening (AFP/Quad test)
  - Noninvasive Prenatal Testing/Screening (NIPT/NIPS)
  - Invasive Prenatal Diagnosis (Amniocentesis, CVS)
- Multiple Miscarriages/Infertility
- Personal and/or Family History of Inherited/Genetic Conditions

For Scheduling and Referrals:
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Genetic counseling is the process of determining the risk you have of passing on an inheritable disease to your baby, which involves:
- Identifying families at risk
- Identifying and explaining the hereditary disorder present in the family
- Analyzing inheritance patterns and risk of recurrence
- Reviewing available options with the family

Indications for Prenatal Genetic Counseling

- Pre-test and post-test counseling for:
  - Maternal serum screening (AFP/Quad test)
  - Noninvasive Prenatal Testing/Screening (NIPT/NIPS)
  - Invasive Prenatal Diagnosis (Amniocentesis, CVS)
- Previous child or a close family member with a birth defect or genetic disorder
- Advanced maternal age (~35 years old)
- Have a history of miscarriages, stillbirths and/or infertility
- Increased risk of being a carrier of a genetic condition because of your ethnic background (some disease are more common in certain ethnicities)
- Prenatal exposure to medication, chemicals, drugs or alcohol during pregnancy

What is noninvasive prenatal screening/testing (NIPS/NIPT)?

Noninvasive prenatal testing (NIPT), also known as noninvasive prenatal screening (NIPS), analyzes small fragments of fetal cell-free DNA from the placenta that are circulating in a pregnant woman's blood starting around 10 weeks gestation. Noninvasive prenatal fetal screening/testing has higher sensitivity and specificity than traditional first and second trimester screening for Down syndrome. It can be used to screen for certain chromosomal disorders, including:
- Trisomy 21 (Down Syndrome)
- Trisomy 18
- Trisomy 13

It can also be used to screen for fetal rhesus (Rh) blood type and fetal sex. NIPT/NIPS might also screen for the increased chance for:
- Trisomy 16 and Trisomy 22
- Sex chromosome disorders such as Turner syndrome (45,X) and Klinefelter syndrome (47,XXY)
- Certain disorders caused by a chromosomal microdeletion syndrome, such as chromosome 22q11.2 deletion syndrome.
- Certain single-gene disorders associated with a known familial mutation