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| **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 | |  | | --- | | **Our Team**  **MJ Hajianpour, MD, PhD, FACMGG**  **Stefanie Yoon, FNP-C**  423-433-6848  **Jennifer Carver, FNP-C**  423-433-6848  **Heather Marshall, LPN**  423-433-6801  **Amber Hall, LPN**  423-433-6810 | | |  |  | | --- | --- | |  | ETSU Medical genetics  325 N. State of Franklin Rd.  Johnson City, TN 37604 | | | |  | | --- | |  | |  | | ETSU Medical genetics | | **Clinical Genetics**  **Pediatrics & Adults** | |
| **What is Clinical Genetic?**   * Evaluation of babies/ infants/ Children and adults with birth defects/ multiple congenital anomalies, developmental delay, speech and language deficit, intellectual disability, and behavioral disorders (ADHD, Autism, etc.). * Diagnosis of genetic disorders verified with genetic studies (if available). * Identify at-risk relative for the known genetic condition in the family by analysis of family history/ Pedigree * Provide genetic counseling including description of identified genetic disorder, mode of inheritance, and recurrence risk | **Clinical Genetics Services**   * **Abnormal Newborn Screening (NBS):** The ETSU Medical Genetics is a follow-up center for abnormal newborn screening tests which is a State program for diagnosis of certain genetic disorders/ inborn errors of metabolism in newborns. * Clinical diagnosis of inherited disorders/ genetic disorders verified by specialized genetic testing including Chromosome Microarray (CMA), Gene Sequencing with deletion/ duplication analysis, Next Generation Sequencing (NGS), Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS). * **Carrier and pre-symptomatic testing:** these tests are used to identify genetic changes when disorders are suspected after birth, or sometimes much later in life. Tests are useful to identify at-risk family members or asymptomatic carriers of the known gene alteration (mutation) in the family. | **Our Process Involves:**   * Review reason for the referral for genetic consultation. * Mailing a package to the patient/ guardian to collect the medical and family history * Pre-clinic review of the gestational history, present illness and past medical history * Prepare and analyze a three-generation pedigree (family history) * Schedule the patient for genetic evaluation and genetic testing * Provide pre-test and post-test counseling * Referring and coordinating patient care based on identified genetic condition including physical/ occupational/ speech therapies. * Providing emotional support and connection to support groups * Continued follow-up as needed throughout the lifespan |