What you need to know about 22q11 deletion syndrome

The 22q11 deletion syndrome (22q11DS) is a common genetic condition currently estimated to occur in 1 out of every 4000 live births. It has been known by many different names in the past, such as velocardiofacial syndrome, DiGeorge syndrome, and Shprintzen syndrome. All of these conditions are actually caused by the same partial deletion of chromosome 22. Complications from this deletion can lead to a variety of clinical problems, which vary from person to person. This diagnosis should be considered in any individual with one or more of the following features:

- Congenital heart defect, particularly conotruncal malformations, such as tetralogy of Fallot, truncus arteriosus, and interrupted aortic arch
- Hypocalcemia, particularly in the newborn period or at times of physiologic stress
- Immunologic deficiency or dysfunction
- Cleft palate
- Velopharyngeal insufficiency, which often results in hypernasal speech and/or swallowing difficulties
- Learning disabilities and psychological disturbances, including psychosis

Testing for this deletion requires that the individual submit 3 to 5 ml of blood drawn in a sodium heparin (green top) tube, which is sent to a cytogenetics laboratory with experience in performing fluorescence in situ hybridization (FISH) for this particular deletion. Emory Genetics Laboratory (1-800-366-1502 or www.genetics.emory.edu) offers this service, as do a number of cytogenetic laboratories around the country.

Recently, the Southeastern Regional Center of Excellence for 22q11 Deletion Syndrome was created. This clinic is geared toward a multidisciplinary approach to the evaluation and treatment of individuals with the 22q11 DS. The clinical team includes members from the following specialties: cardiology, immunology, clinical genetics, endocrinology, craniofacial surgery, speech/language pathology, and psychiatry. To refer a patient to our multidisciplinary clinic, please call:

- 404-778-8570 (English-speaking patients)
- 404-785-2672 (Spanish-speaking patients)