Physicians in the Division of Child Neurology bring together expertise in neurogenetics from both clinical and research perspectives. Our physicians, in collaboration with clinical geneticists, refer patients to a weekly neurogenetics research clinic for advanced genetic evaluation and testing. Conditions evaluated in this clinic include epilepsy, developmental delay, neuromuscular disorders, microcephaly, and movement disorders. The comprehensive evaluation varies from patient to patient but can include complete neurological and genetic exams, improved genotyping using whole exome sequencing (WES) or whole genome sequencing (WGS), and skin biopsies for research analysis of skin fibroblasts. After diagnosis, children can continue to receive neurological care with Dr. Devorah Segal in a weekly clinical neurogenetics clinic.
The Division has three neuro-oncologists with expertise in diagnosing and treating tumors of the brain and spinal cord. They work together with our pediatric neurosurgeons to provide comprehensive care to children with tumors of the central nervous system. Bridging neuro-oncology and neurogenetics is our neurofibromatosis program. Dr. Segal sees children with known or suspected NF1 in her weekly neurogenetics clinic and coordinates their care together with pediatric neurosurgeons, ophthalmologists, and endocrinologists. She works together with neurogeneticists and genetic counselors to arrange genetic testing and counseling as indicated.

Services & Programs
In our neurogenetics clinic, we evaluate children with syndromic or familial epilepsy, multiple neurologic abnormalities, undiagnosed neuromuscular or movement disorders, and global developmental delay. We treat patients with neurofibromatosis types 1 and 2 and coordinate their care among multiple specialties. We provide surveillance and management of children with benign or incidentally found brain lesions and coordinate care with neurosurgeons when necessary.

Diagnosis & Treatment
At your initial visit to our neurogenetics clinic, a genetic counselor and research associate will meet with you and your child to obtain a comprehensive personal and family history. A physician from our team will perform a thorough neurologic and physical exam. We offer comprehensive genetic testing that may include specialized genetic panels, whole exome sequencing, whole genome sequencing, and parental testing. Blood draws can be performed on site at the time of the visit and do not require an additional visit to a lab.

What Sets us Apart
- Dr. Devorah Segal’s expertise in the diagnosis and clinical management of patients with neurofibromatosis type 1 and other brain tumor predisposition syndromes.
- Dr. Srishti Nangia’s extensive experience in the diagnosis and clinical management of patients with tuberous sclerosis and epileptic encephalopathies.
- Three clinical epileptologists to investigate the genetics of epilepsy and provide clinical care to these patients.
- Three clinical pediatric geneticists and one genetics counselor with extensive expertise in neurogenetics.
- Research-based geneticist and a research laboratory that performs preclinical translational research.
Pediatric Services

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