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Better clinical management improves quality of life for neurofibromatosis patients

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A genetic disorder called neurofibromatosis (NF) causes benign tumors to grow on the brain, spinal cord, and other parts of the nervous system.

There are no effective drugs to prevent or reverse NF. But increasing scientific knowledge has allowed for better clinical management and fewer complications, resulting in a higher quality of life for neurofibromatosis patients, NF specialists report in the *Journal of Neuropsychiatry and Clinical Neurosciences*.

Pediatric neurologist and NF specialist Nikolas Mata-Machado, MD, of Loyola University Medical Center, is a co-author of the paper, which proposes guidelines for the clinical management of NF. Writing the guidelines was a collaboration among the top neurofibromatosis groups in Brazil, where Dr. Mata-Machado grew up and went to medical school.

At least 100,000 people in the United States have NF, making it one of the most common genetic disorders. NF affects roughly 1 in every 3,000 babies born in the U.S. There are two types of neurofibromatosis (NF1 and NF2) and a related condition called schwannomatosis.

People with NF typically have brown spots on the skin called café-au-lait spots; soft bumps on or under the skin called neurofibromas; and tiny bumps on the iris called Lisch nodules. NF can cause a wide range of effects throughout the body, including learning disabilities; hyperactivity; headaches; hearing problems; short stature; facial drop; heart problems; numbness and weakness in arms or legs; balance

problems; and bone deformities such as curvature of the spine.

"Most medical doctors are able to perform NF diagnosis, but the wide range of clinical manifestations and the inability to predict the onset or severity of new features, consequences, or complications make NF management a real clinical challenge," the guidelines say.

Treating neurofibromatosis often requires specialists from multiple disciplines, especially for patients with NF2 and schwannomatosis.

Each NF patient is unique, and there's no single standard clinical approach that applies to all patients, the guidelines say. "Considering the natural history of NF as individualized, distinctive, and unpredictable, the main clinical procedure for all NF forms is periodic medical examination throughout life, aiming for the early detection and treatment of possible complications."

The guidelines recommend that patients undergo annual medical visits, unless new signs or symptoms hasten the schedule. All NF patients should have their medical, developmental and familial histories reviewed periodically, and receive appropriate genetic counseling. They also should have a complete physical exam, with emphasis on the cardiovascular and nervous systems.

The guidelines conclude that neurofibromatosis is among the thousands of rare diseases that "progressively demand well-connected reference centers for information, treatment, genetic counseling and improvement in the quality of life of the affected individuals."

Provided by Loyola University Health System

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