An investigation of the clinical care practices of Neurofibromatosis type I in North America

Kristin Levandoski, Seema Jamal, Kathleen Berentsen, Christina Flora, MaryAnn W. Campion
Boston University School of Medicine, Boston, Massachusetts

Neurofibromatosis type 1 (NF1) is one of the most common genetic disorders. The diagnostic criteria and clinical findings of NF1 have been well documented in the scientific literature. These criteria, as initially presented in the National Institutes of Health Consensus Development Conference Statement on Neurofibromatosis in 1987, include seven key findings. Two or more of these findings are needed for a diagnosis of NF1. Specific recommendations regarding the surveillance, management and treatment of patients with NF1 have been made for a number of specialties. This information is often broken down into age-dependent categories, as many of the clinical findings of NF1 are clearly age-related. A number of sources recommend the lifelong care of NF1 patients to be coordinated by a medical home or set at a multidisciplinary clinic. However, within all the available information on how the care of NF1 patients should be managed, there is no investigation into how this is actually occurring. Some information is available on how certain findings have been managed, but no information is available on how clinics or sites go about the management and screening of individuals with NF1. This study uncovered the current clinical practices, compared them to published recommendations, as well as between sites, in order to work towards improving the standard of care for those diagnosed with NF1.