James S. Amoss
L2
LSU Health Sciences Center, New Orleans, LA

Dr. Regina Zambrano:
Children's Hospital of New Orleans

"Chart-Based Insights into Neurofibromatosis Type 1: A Single-Center Database"

Introduction: Neurofibromatosis type 1 (NF1) is a common (1:2500) autosomal dominant disorder, caused by heterozygous pathogenic variants (PV) in *NF1* (a tumor suppressor gene). NF1 is a neurocutaneous disorder, presenting with pigmentary lesions (café-au-lait macules, skinfold freckling and Lisch nodules) and dermal neurofibromas. Other systems can be affected including the skeleton (scoliosis, tibial pseudarthrosis and orbital dysplasia), central nervous system (optic pathway gliomas and glioblastoma), peripheral nervous system (spinal neurofibromas, plexiform neurofibromas, and malignant peripheral nerve sheath tumors), learning disabilities, attention deficits, and social and behavioral problems. Patients have an increased risk for tumor development and potential for malignant transformation. A related disorder, Legius syndrome, has significant clinical overlap with NF1. It is also inherited in an autosomal dominant pattern and characterized by multiple CALMs with or without skinfold freckling, caused by heterozygous PV in *SPRED1.*

There is no current cure for NF1. However, in April 2020, a treatment option (selumetinib) for children with NF1-related symptomatic plexiform neurofibromas, which arise in approximately 50% of patients, was approved by the Food and Drug Administration (FDA).

Objective: The primary objectives of this database are to characterize and identify trends within the patient population at the Children's Hospital New Orleans (CHNOLA) NF1 and related disorders clinic. By consolidating patient data, this database aims to enhance clinical care and inform patient decision-making. Additionally, the centralized data will facilitate and support future research projects related to the NF1 and related disorders population.

Methods: A retrospective chart review was conducted on all patients seen in the CHNOLA NF1 and related disorders clinic from July 2018 to May 2024. Data was collected and stored using LSU (Louisiana State University) REDCap software encompassing demographics, clinical manifestations, laboratory test results, treatments, interventions (including the use of selumetinib), dermatoglyphics, molecular testing results, and inheritance patterns.