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INTRODUCTION: We aimed to (1) characterize the spectrum of clinical phenotypes of NF1 in a random pediatric population, (2) correlate genotype with phenotypic expression for those with a genetic diagnosis, and (3) explore radiological features of NF1 in the central nervous system (CNS) by radiomics analyses to predict clinical course. METHODS: We performed a database search in the hospital information system of the University Children´s Hospital between January 2017 and December 2020 for patients with NF1 and evaluated the clinical phenotype by retrospective chart review. RESULTS: 75 children/adolescents were identified with suspicion/clinical diagnosis of NF1 (median age 10.0 years (range, 1.1-22.6); 35 female), confirmatory revised "diagnostic criteria" were met in 57 patients at the last follow-up. Per number of documented items, major signs were detected as 73/75 café-au-lait macules, 31/63 freckling, 38/71 neurofibromas (thereof 21 plexiform neurofibromas), 18/43 optic pathway glioma, 5/66 Lisch nodules, and two patients with sphenoid dysplasia. Genetic analysis (31/75) identified pathogenic NF1 variants in 27 patients. In 20/66 cases a parent met diagnostic criteria. Cognitive symptoms included developmental delay (28/68), learning deficits (12/48), attention-deficit hyperactivity disorder (3/53), and behavior anomalies (7/63). Classical unidentified bright objects were seen in 29/43, other intracranial tumors in 7/43, and cerebrovascular abnormalities in 5/43. Analysis of imaging features of the CNS in these patients will involve lesion segmentation and radiomics features. Symptomatic/progressive low-grade glioma necessitated neurosurgical resection (4/25) and/or chemotherapy (12/25). In 10/25 neuropsychological functions were assessed by the German neuropsychological basic diagnostic instrument. Until June 30th, 2021, one patient died of progressive plexiform neurofibroma. CONCLUSIONS: A wide range of neurological manifestations, including neuropsychological deficits, should raise the suspicion of NF1 in an unselected pediatric population. We expect imaging features of the CNS to better predict the clinical course and enhance decision-making.

NFB-03. NEUROLOGICAL MANIFESTATIONS IN CHILDREN AND ADOLESCENTS WITH NEUROFIBROMATOSIS TYPE 1 - IMPLICATIONS FOR MANAGEMENT AND SURVEILLANCE

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