Clinical and Genetic Findings in Children with Neurofib and Other Related Neurocutaneous Disorders

Genes

10, 580

DOI: 10.3390/genes10080580

Citation Report

#	Article	IF	Citations
1	Understanding the Biological Activities of Vitamin D in Type 1 Neurofibromatosis: New Insights into Disease Pathogenesis and Therapeutic Design. Cancers, 2020, 12, 2965.	1.7	12
2	Constitutional mismatch repair deficiency is the diagnosis in 0.41% of pathogenic NF1/SPRED1 variant negative children suspected of sporadic neurofibromatosis type 1. Genetics in Medicine, 2020, 22, 2081-2088.	1.1	14
3	A novel RAB39B mutation and concurrent de novo NF1 mutation in a boy with neurofibromatosis type 1, intellectual disability, and autism: a case report. BMC Neurology, 2020, 20, 327.	0.8	21
4	Retrospective Multicentric Study on Non-Optic CNS Tumors in Children and Adolescents with Neurofibromatosis Type 1. Cancers, 2020, 12, 1426.	1.7	8
5	Identification and Characterization of Splicing Defects by Single-Molecule Real-Time Sequencing Technology (PacBio). Journal of Neuromuscular Diseases, 2020, 7, 477-481.	1.1	7
6	Simultaneous Detection of NF1, SPRED1, LZTR1, and NF2 Gene Mutations by Targeted NGS in an Italian Cohort of Suspected NF1 Patients. Genes, 2020, 11, 671.	1.0	5
7	Bioactive Phenolic Compounds in the Modulation of Central and Peripheral Nervous System Cancers: Facts and Misdeeds. Cancers, 2020, 12, 454.	1.7	12
8	Evaluation of clinical findings and neurofibromatosis type 1 bright objects on brain magnetic resonance images of 60 Turkish patients with NF1 gene variants. Neurological Sciences, 2021, 42, 2045-2057.	0.9	1
9	Olfactory bulb enlargement in neurofibromatosis type 1: report of a novel finding. Child's Nervous System, 2021, 37, 2927-2930.	0.6	1
10	Central precocious puberty in a girl with LEGIUS syndrome: an accidental association?. Italian Journal of Pediatrics, 2021, 47, 50.	1.0	0
11	Ampliando el diagnÃ³stico de las manchas cafÃ© con leche. Piel, 2022, 37, 59-60.	0.0	0
12	Mutational spectrum of NF1 gene in 24 unrelated Egyptian families with neurofibromatosis type 1. Molecular Genetics & Genomic Medicine, 2021, 9, e1631.	0.6	5
15	Reviewing the occurrence of large genomic rearrangements in patients with inherited cancer predisposing syndromes: importance of a comprehensive molecular diagnosis. Expert Review of Molecular Diagnostics, 2022, 22, 319-346.	1.5	1
16	Challenges in the diagnosis of neurofibromatosis type 1 (NF1) in young children facilitated by means of revised diagnostic criteria including genetic testing for pathogenic NF1 gene variants. Human Genetics, 2022, 141, 177-191.	1.8	29
17	RASopathies: Dermatologistsâ€™ viewpoints. Indian Journal of Dermatology, Venereology and Leprology, 2021, 88, 452-463.	0.2	1
18	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: Identification of Novel and Recurrent NF1 Gene Variants and Correlations with Neurocognitive Phenotype. Genes, 2022, 13, 1130.	1.0	10
19	Inside the Noonan â€œuniverseâ€: Literature review on growth, GH/IGF axis and rhGH treatment: Facts and concerns. Frontiers in Endocrinology, 0, 13, .	1.5	5
20	Neurofibromatosis Type 1 and Hypospadias in a Male 46, XY with a Mutation in the NF1 Gene and a Mutation in NR5A1. Pharmacogenomics and Personalized Medicine, 0, Volume 15, 873-878.	0.4	0

#	Article	IF	Citations
21	Identifying a novel frameshift pathogenic variant in a Chinese family with neurofibromatosis type 1 and review of literature. International Journal of Ophthalmology, 2023, 16, 47-52.	0.5	0
22	Neurofibromatosis Type 1: Pediatric Aspects and Review of Genotypeâ€“Phenotype Correlations. Cancers, 2023, 15, 1217.	1.7	5