# Veronica Saletti

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9493953/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 57 papers | 2,053 citations | 331670 21 h-index | 254184 43 g-index |
| 59 all docs | 59 docs citations | 59 times ranked | 3474 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46. | 21.4 | 245 |
| 2 | The molecular landscape of glioma in patients with Neurofibromatosis 1. Nature Medicine, 2019, 25, 176-187. | 30.7 | 145 |
| 3 | Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87. | 6.2 | 144 |
| 4 | Intrathecal methotrexate affects cognitive function in children with medulloblastoma. Neurology, 2002, 59, 48-53. | 1.1 | 138 |
| 5 | Sepiapterin reductase deficiency: A Treatable Mimic of Cerebral Palsy. Annals of Neurology, 2012, 71, 520-530. | 5.3 | 125 |
| 6 | Late neuropsychological and behavioural outcome of children surgically treated for craniopharyngioma. Child's Nervous System, 1998, 14, 179-184. | 1.1 | 92 |
| 7 | Clinical spectrum of individuals with pathogenic  N F1  missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315. | 2.5 | 80 |
| 8 | MRI features of cerebral lesions and cognitive functions in preterm spastic diplegic children. Pediatric Neurology, 1996, 15, 207-212. | 2.1 | 77 |
| 9 | Neuropsychologic Effects of Frontal Lobe Epilepsy in Children. Journal of Child Neurology, 2002, 17, 661-667. | 1.4 | 63 |
| 10 | Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849. | 2.5 | 63 |
| 11 | Novel mutations in the CDKL5 gene, predicted effects and associated phenotypes. Neurogenetics, 2009, 10, 241-250. | 1.4 | 62 |
| 12 | Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876. | 2.4 | 62 |
| 13 | Unilateral frontal lobe epilepsy affects executive functions in children. Neurological Sciences, 2005, 26, 263-270. | 1.9 | 53 |
| 14 | A novelCLN8mutation in late-infantile-onset neuronal ceroid lipofuscinosis (LINCL) reveals aspects of CLN8 neurobiological function. Human Mutation, 2009, 30, 1104-1116. | 2.5 | 53 |
| 15 | Expanding the phenotypic spectrum of Allanâ€“Herndonâ€“Dudley syndrome in patients with <scp>SLC</scp>16A2 mutations. Developmental Medicine and Child Neurology, 2019, 61, 1439-1447. | 2.1 | 53 |
| 16 | Diagnosis and treatment of Chiari malformation and syringomyelia in adults: international consensus document. Neurological Sciences, 2022, 43, 1327-1342. | 1.9 | 42 |
| 17 | Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. European Journal of Medical Genetics, 2019, 62, 103596. | 1.3 | 39 |
| 18 | Neuroradiological diagnosis of Chiari malformations. Neurological Sciences, 2011, 32, 283-286. | 1.9 | 34 |

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|---|---|---|---|
| 19 | Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750. | 2.4 | 29 |
| 20 | Treatment for Chiari 1 malformation (CIM): analysis of a pediatric surgical series. Neurological Sciences, 2011, 32, 321-324. | 1.9 | 25 |
| 21 | 126 novel mutations in Italian patients with neurofibromatosis type 1. Molecular Genetics & Genomic Medicine, 2015, 3, 513-525. | 1.2 | 25 |
| 22 | Progressive bone impairment with age and pubertal development in neurofibromatosis type I. Archives of Osteoporosis, 2018, 13, 93. | 2.4 | 25 |
| 23 | Diagnosis and treatment of Chiari malformation type 1 in children: the International Consensus Document. Neurological Sciences, 2022, 43, 1311-1326. | 1.9 | 24 |
| 24 | Can Chiari malformation negatively affect higher mental functioning in developmental age?. Neurological Sciences, 2011, 32, 307-309. | 1.9 | 22 |
| 25 | A CDKL5 mutated child with precocious puberty. American Journal of Medical Genetics, Part A, 2009, 149A, 1046-1051. | 1.2 | 21 |
| 26 | Risk of Optic Pathway Glioma in Neurofibromatosis Type 1: No Evidence of Genotypeâ€“Phenotype Correlations in a Large Independent Cohort. Cancers, 2019, 11, 1838. | 3.7 | 19 |
| 27 | Neurological pictures in Paediatric Chiari I malformation. Neurological Sciences, 2011, 32, 295-298. | 1.9 | 17 |
| 28 | Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135. | 2.7 | 17 |
| 29 | GEN-O-MA project: an Italian network studying clinical course and pathogenic pathways of moyamoya diseaseâ€”study protocol and preliminary results. Neurological Sciences, 2019, 40, 561-570. | 1.9 | 15 |
| 30 | Vascular Remodeling in Moyamoya Angiopathy: From Peripheral Blood Mononuclear Cells to Endothelial Cells. International Journal of Molecular Sciences, 2020, 21, 5763. | 4.1 | 15 |
| 31 | Chiari I malformation in a child with PTEN hamartoma tumor syndrome: Association or coincidence?. European Journal of Medical Genetics, 2017, 60, 261-264. | 1.3 | 14 |
| 32 | Non-Coding RNA and Tumor Development in Neurofibromatosis Type 1: ANRIL Rs2151280 Is Associated with Optic Glioma Development and a Mild Phenotype in Neurofibromatosis Type 1 Patients. Genes, 2019, 10, 892. | 2.4 | 14 |
| 33 | Visuoperceptual Impairment in Children with NF1: From Early Visual Processing to Procedural Strategies. Behavioural Neurology, 2019, 2019, 1-10. | 2.1 | 14 |
| 34 | A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647. | 3.9 | 13 |
| 35 | Chiari I malformation in defined genetic syndromes in children: are there common pathways?. Child's Nervous System, 2019, 35, 1727-1739. | 1.1 | 13 |
| 36 | Verbal and Gestural Communication in Children With Bilateral Perisylvian Polymicrogyria. Journal of Child Neurology, 2007, 22, 1090-1098. | 1.4 | 12 |

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|---|---|---|---|
| 37 | Electroencephalographic (EEG) Photoparoxysmal Responses Under 5 Years of Age. Journal of Child Neurology, 2015, 30, 1824-1830. | 1.4 | 12 |
| 38 | Novel mutations in SLC16A2 associated with a less severe phenotype of MCT8 deficiency. Metabolic Brain Disease, 2019, 34, 1565-1575. | 2.9 | 12 |
| 39 | The TAND checklist: a useful screening tool in children with tuberous sclerosis and neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2020, 15, 237. | 2.7 | 12 |
| 40 | The management of Chiari malformation type 1 and syringomyelia in children: a review of the literature. Neurological Sciences, 2021, 42, 4965-4995. | 1.9 | 12 |
| 41 | Chiari 1 malformation and untreated sagittal synostosis: a new subset of complex Chiari?. Child's Nervous System, 2019, 35, 1741-1753. | 1.1 | 11 |
| 42 | Plasma Lipid Profiling Contributes to Untangle the Complexity of Moyamoya Arteriopathy. International Journal of Molecular Sciences, 2021, 22, 13410. | 4.1 | 11 |
| 43 | Brain Tumors in NF1 Children: Influence on Neurocognitive and Behavioral Outcome. Cancers, 2019, 11, 1772. | 3.7 | 10 |
| 44 | The Key Search Subtest of the Behavioural Assessment of the Dysexecutive Syndrome in Children (BADS-C) Instrument Reveals Impaired Planning Without External Constraints in Children With Neurofibromatosis Type 1. Journal of Child Neurology, 2017, 32, 387-396. | 1.4 | 9 |
| 45 | Fluorescein-guided resection of plexiform neurofibromas: how I do it. Acta Neurochirurgica, 2019, 161, 2141-2145. | 1.7 | 8 |
| 46 | Paediatric Stroke: Review of the Literature and Possible Treatment Options, including Endovascular Approach. Stroke Research and Treatment, 2011, 2011, 1-11. | 0.8 | 7 |
| 47 | The absence that makes the difference: choroidal abnormalities in Legius syndrome. Journal of Human Genetics, 2017, 62, 1001-1004. | 2.3 | 7 |
| 48 | Clinical and Molecular Spectrum of Myotonia and Periodic Paralyses Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. Frontiers in Neurology, 2020, 11, 646. | 2.4 | 7 |
| 49 | Neurological malignancies in neurofibromatosis type 1. Current Opinion in Oncology, 2019, 31, 554-561. | 2.4 | 5 |
| 50 | 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. | 2.4 | 5 |
| 51 | Response to â€œcharacteristics of 2p15-p16.1 microdeletion syndrome: review and description of two additional patientsâ€• Congenital Anomalies (discontinued), 2015, 55, 191-192. | 0.6 | 4 |
| 52 | The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 557-562. | 1.7 | 4 |
| 53 | Language abilities and gestural communication in a girl with bilateral perisylvian syndrome: a clinical and rehabilitative follow-up. Neurological Sciences, 2010, 31, 471-481. | 1.9 | 3 |
| 54 | Legius Syndrome: two novel mutations in the SPRED1 gene. Human Genome Variation, 2015, 2, 15051. | 0.7 | 3 |

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|---|---|---|---|
| 55 | CGH Findings in Children with Complex and Essential Autistic Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, , 1. | 2.7 | 2 |
| 56 | The Lipid Asset Is Unbalanced in Peripheral Nerve Sheath Tumors. International Journal of Molecular Sciences, 2022, 23, 61. | 4.1 | 2 |
| 57 | Unique combination of myxopapillary ependymoma and conus lipoma with subcutaneous extension in an 11-month-old child. Child's Nervous System, 2018, 34, 597-599. | 1.1 | 1 |