

Davide Tonduti

List of Publications by Year in descending order

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Version: 2024-02-01

76
papers

2,857
citations

236925

25
h-index

197818

49
g-index

78
all docs

78
docs citations

78
times ranked

4295
citing authors

#	ARTICLE	IF	CITATIONS
1	How to look for intracranial calcification in children with neurological disorders: CT, MRI, or both of them?. <i>Neurological Sciences</i> , 2022, 43, 2043-2050.	1.9	1
2	Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e1136-e1147.	3.6	15
3	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS. <i>Annals of the Rheumatic Diseases</i> , 2022, 81, 601-613.	0.9	31
4	Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 109-113.	1.1	17
5	Correspondence on "Expanded phenotype of AARS1-related white matter disease" by Helman et al. <i>Genetics in Medicine</i> , 2022, , .	2.4	2
6	Mucopolysaccharidosis-Plus Syndrome, a Rapidly Progressive Disease: Favorable Impact of a Very Prolonged Steroid Treatment on the Clinical Course in a Child. <i>Genes</i> , 2022, 13, 442.	2.4	8
7	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: <sc>CANDLE</sc>/<sc>PRAAS</sc>, <sc>SAVI</sc>, and <sc>AGS</sc>. <i>Arthritis and Rheumatology</i> , 2022, 74, 735-751.	5.6	23
8	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i>, <i>POLR3B</i>, and <i>POLR1C</i>. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e660-e674.	3.6	26
9	Impact of COVID-19 lockdown in children with neurological disorders in Italy. <i>Disability and Health Journal</i> , 2021, 14, 101053.	2.8	16
10	Late-Onset Aicardi-Goutières Syndrome: A Characterization of Presenting Clinical Features. <i>Pediatric Neurology</i> , 2021, 115, 1-6.	2.1	18
11	<i>PRKRA</i>-Related Disorders: Bilateral Striatal Degeneration in Addition to DYT16 Spectrum. <i>Movement Disorders</i> , 2021, 36, 1038-1040.	3.9	10
12	Ruxolitinib in Aicardi-Goutières syndrome. <i>Metabolic Brain Disease</i> , 2021, 36, 859-863.	2.9	12
13	Hydroxychloroquine modulates immunological pathways activated by RNA:DNA hybrids in Aicardi-Goutières syndrome patients carrying RNASEH2 mutations. <i>Cellular and Molecular Immunology</i> , 2021, 18, 1593-1595.	10.5	3
14	Spinal cord involvement and paroxysmal events in "Infantile Onset Transient Hypomyelination" due to TMEM63A mutation. <i>Journal of Human Genetics</i> , 2021, 66, 1035-1037.	2.3	12
15	The epileptology of Aicardi-Goutières syndrome: electro-clinical-radiological findings. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 86, 197-209.	2.0	2
16	Case Report: Novel Compound Heterozygous RNASEH2B Mutations Cause Aicardi-Goutières Syndrome. <i>Frontiers in Immunology</i> , 2021, 12, 672952.	4.8	1
17	Neurodevelopmental outcome of preterm very low birth weight infants admitted to an Italian tertiary center over an 11-year period. <i>Scientific Reports</i> , 2021, 11, 16316.	3.3	11
18	Alexander disease evolution over time: data from an Italian cohort of pediatric-onset patients. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 353-358.	1.1	6

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19	Novel and emerging treatments for Aicardi-Goutières syndrome. <i>Expert Review of Clinical Immunology</i> , 2020, 16, 189-198.	3.0	27
20	<i>RARS1</i> -related hypomyelinating leukodystrophy: Expanding the spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 83-93.	3.7	18
21	Genetic and phenotypic spectrum associated with <i>IFIH1</i> gain-of-function. <i>Human Mutation</i> , 2020, 41, 837-849.	2.5	63
22	Disease characteristics of <i>MCT8</i> deficiency: an international, retrospective, multicentre cohort study. <i>Lancet Diabetes and Endocrinology</i> , 2020, 8, 594-605.	11.4	50
23	<i>cGAS</i> -mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. <i>Nature Genetics</i> , 2020, 52, 1364-1372.	21.4	105
24	Phenotypic spectrum of short-chain enoyl-Coa hydratase-1 (<i>ECHS1</i>) deficiency. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 151-158.	1.6	18
25	De novo <i>EIF2AK1</i> and <i>EIF2AK2</i> Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	6.2	37
26	Expanding the phenotypic spectrum of Allan-Herndon-Dudley syndrome in patients with <i>SLC16A2</i> mutations. <i>Developmental Medicine and Child Neurology</i> , 2019, 61, 1439-1447.	2.1	53
27	Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with <i>MCT8</i> deficiency: an international, single-arm, open-label, phase 2 trial. <i>Lancet Diabetes and Endocrinology</i> , 2019, 7, 695-706.	11.4	77
28	Novel mutations in <i>SLC16A2</i> associated with a less severe phenotype of <i>MCT8</i> deficiency. <i>Metabolic Brain Disease</i> , 2019, 34, 1565-1575.	2.9	12
29	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. <i>Journal of Clinical Medicine</i> , 2019, 8, 750.	2.4	29
30	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-Goutières syndrome: Diagnostic and disease-monitoring implications. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 489-494.	1.1	10
31	Cortical malformations and <i>COL4A1</i> mutation: Three new cases. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 410-417.	1.6	7
32	Clinical spectrum of <i>PTEN</i> mutation in pediatric patients. A bicenter experience. <i>European Journal of Medical Genetics</i> , 2019, 62, 103596.	1.3	39
33	Clinical spectrum of <i>POLR3</i> -related leukodystrophy caused by biallelic <i>POLR1C</i> pathogenic variants. <i>Neurology: Genetics</i> , 2019, 5, e369.	1.9	38
34	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. <i>Pediatric Neurology</i> , 2018, 84, 21-26.	2.1	9
35	<i>SLC19A3</i> related disorder: Treatment implication and clinical outcome of 2 new patients. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 332-335.	1.6	6
36	<i>KARS</i> -related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 45.	2.7	32

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37	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. <i>Journal of Human Genetics</i> , 2018, 63, 563-568.	2.3	15
38	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 135.	2.7	17
39	A <i>PDE10A</i> de novo mutation causes childhood-onset chorea with diurnal fluctuations. <i>Movement Disorders</i> , 2017, 32, 1646-1647.	3.9	13
40	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 18-32.	1.1	42
41	Neonatal detection of Aicardi Goutières Syndrome by increased C26:0 lysophosphatidylcholine and interferon signature on newborn screening blood spots. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 134-139.	1.1	43
42	Thiamine-responsive disease due to mutation of <i>tpk1</i> : Importance of avoiding misdiagnosis. <i>Neurology</i> , 2017, 89, 870-871.	1.1	13
43	Time-course of myelination and atrophy on cerebral imaging in 35 patients with <i>PLP1</i> -related disorders. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 706-713.	2.1	20
44	Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in <i>PLA2G6</i> -associated neurodegeneration. <i>European Journal of Neurology</i> , 2016, 23, e24-5.	3.3	2
45	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 604-610.	1.6	29
46	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. <i>Current Neurology and Neuroscience Reports</i> , 2016, 16, 54.	4.2	23
47	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. <i>American Journal of Human Genetics</i> , 2016, 99, 1229-1244.	6.2	91
48	Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease. <i>Neurological Sciences</i> , 2016, 37, 973-977.	1.9	7
49	Early-onset progressive spastic paraplegia caused by a novel TUBB4A mutation: brain MRI and FDG-PET findings. <i>Journal of Neurology</i> , 2016, 263, 591-593.	3.6	17
50	TUBB4A-related hypomyelinating leukodystrophy: New insights from a series of 12 patients. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 323-330.	1.6	24
51	Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. <i>Neurology</i> , 2016, 86, 28-35.	1.1	59
52	Altered <i>PLP1</i> splicing causes hypomyelination of early myelinating structures. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 648-661.	3.7	27
53	A novel mutation in <i>COL4A1</i> gene: A possible cause of early postnatal cerebrovascular events. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 810-815.	1.2	7
54	Cystic leukoencephalopathy with cortical dysplasia related to <i>LAMB1</i> mutations. <i>Neurology</i> , 2015, 84, 2195-2197.	1.1	17

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55	Case definition and classification of leukodystrophies and leukoencephalopathies. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 494-500.	1.1	185
56	Characterization of human disease phenotypes associated with mutations in <i>TREX1</i> , <i>RNASEH2A</i> , <i>RNASEH2B</i> , <i>RNASEH2C</i> , <i>SAMHD1</i> , <i>ADAR</i> , and <i>IFIH1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 296-312.	1.2	447
57	New spastic paraplegia phenotype associated to mutation of <i>NFU1</i> . <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 13.	2.7	30
58	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. <i>Journal of Child Neurology</i> , 2015, 30, 1800-1805.	1.4	3
59	Brain Magnetic Resonance Imaging (MRI) Pattern Recognition in Pol III-Related Leukodystrophies. <i>Journal of Child Neurology</i> , 2014, 29, 214-220.	1.4	47
60	Characteristic brain magnetic resonance imaging pattern in patients with macrocephaly and <i>PTEN</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 627-633.	1.2	70
61	Bilateral striatal necrosis in two subjects with Aicardi-Goutières syndrome due to mutations in <i>ADAR1</i> (<i>AGS6</i>). <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 815-819.	1.2	30
62	<i>MCT8</i> Deficiency. <i>Journal of Child Neurology</i> , 2013, 28, 795-800.	1.4	48
63	A De Novo Mutation in the β -Tubulin Gene <i>TUBB4A</i> Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. <i>American Journal of Human Genetics</i> , 2013, 92, 767-773.	6.2	174
64	Novel Hypomyelinating Leukoencephalopathy Affecting Early Myelinating Structures: Clinical Course in Two Brothers. <i>Neuropediatrics</i> , 2013, 44, 213-217.	0.6	4
65	Dysregulation of the immune system in Aicardi-Goutières syndrome: another example in a <i>TREX1</i> -mutated patient. <i>Lupus</i> , 2013, 22, 1064-1069.	1.6	22
66	More Than Hypomyelination in Pol-III Disorder. <i>Journal of Neuropathology and Experimental Neurology</i> , 2013, 72, 67-75.	1.7	27
67	<i>COL4A1</i> -Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. <i>Neuropediatrics</i> , 2012, 43, 283-288.	0.6	20
68	Neurotransmitter abnormalities and response to supplementation in <i>SPG11</i> . <i>Molecular Genetics and Metabolism</i> , 2012, 107, 229-233.	1.1	17
69	Calcifying leukoencephalopathies: New overlapping phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 964-965.	1.2	0
70	Recessive Mutations in <i>POLR3B</i> , Encoding the Second Largest Subunit of Pol III, Cause a Rare Hypomyelinating Leukodystrophy. <i>American Journal of Human Genetics</i> , 2011, 89, 652-655.	6.2	139
71	Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G > A mitochondrial DNA mutation: a case report. <i>BMC Neurology</i> , 2011, 11, 85.	1.8	18
72	<i>COL4A1</i> Mutations Associated with a Characteristic Pattern of Intracranial Calcification. <i>Neuropediatrics</i> , 2011, 42, 227-233.	0.6	38

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73	Spinal Cord Calcification in an Early-Onset Progressive Leukoencephalopathy. Journal of Child Neurology, 2011, 26, 876-880.	1.4	9
74	Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency. Neurology, 2010, 75, 64-71.	1.1	198
75	New Case of 4H Syndrome and a Review of the Literature. Pediatric Neurology, 2010, 42, 359-364.	2.1	21
76	Leukodystrophies and Epilepsy. , 0, , .		0