Davide Tonduti

List of Publications by Year
in descending order

76	2,857	236925 25	197818 49
papers	citations	h-index	g-index

78	78	78	4295
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
2	Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency. Neurology, 2010, 75, 64-71.	1.1	198
3	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
4	A De Novo Mutation in the Î²-Tubulin Gene TUBB4A Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. American Journal of Human Genetics, 2013, 92, 767-773.	6.2	174
5	Recessive Mutations in POLR3B, Encoding the Second Largest Subunit of Pol III, Cause a Rare Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 652-655.	6.2	139
6	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	21.4	105
7	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	6.2	91
8	Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with MCT8 deficiency: an international, single-arm, open-label, phase 2 trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 695-706.	11.4	77
9	Characteristic brain magnetic resonance imaging pattern in patients with macrocephaly and PTEN mutations. American Journal of Medical Genetics, Part A, 2014, 164, 627-633.	1.2	70
10	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
11	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
12	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
13	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	11.4	50
14	MCT8 Deficiency. Journal of Child Neurology, 2013, 28, 795-800.	1.4	48
15	Brain Magnetic Resonance Imaging (MRI) Pattern Recognition in Pol III-Related Leukodystrophies. Journal of Child Neurology, 2014, 29, 214-220.	1.4	47
16	Neonatal detection of Aicardi GoutiÃ¨res Syndrome by increased C26:0 lysophosphatidylcholine and interferon signature on newborn screening blood spots. Molecular Genetics and Metabolism, 2017, 122, 134-139.	1.1	43
17	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	1.1	42
18	Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. European Journal of Medical Genetics, 2019, 62, 103596.	1.3	39

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19	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
20	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants. Neurology: Genetics, 2019, 5, e369.	1.9	38
21	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
22	KARS-related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature. Orphanet Journal of Rare Diseases, 2018, 13, 45.	2.7	32
23	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. Annals of the Rheumatic Diseases, 2022, 81, 601-613.	0.9	31
24	Bilateral striatal necrosis in two subjects with Aicardiâ€“GoutiÃ¨res syndrome due to mutations in ADAR1 (AGS6). American Journal of Medical Genetics, Part A, 2014, 164, 815-819.	1.2	30
25	New spastic paraplegia phenotype associated to mutation of NFU1. Orphanet Journal of Rare Diseases, 2015, 10, 13.	2.7	30
26	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610.	1.6	29
27	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
28	More Than Hypomyelination in Pol-III Disorder. Journal of Neuropathology and Experimental Neurology, 2013, 72, 67-75.	1.7	27
29	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
30	Novel and emerging treatments for Aicardi-GoutiÃ¨res syndrome. Expert Review of Clinical Immunology, 2020, 16, 189-198.	3.0	27
31	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
32	TUBB4A-related hypomyelinating leukodystrophy: New insights from a series of 12 patients. European Journal of Paediatric Neurology, 2016, 20, 323-330.	1.6	24
33	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Current Neurology and Neuroscience Reports, 2016, 16, 54.	4.2	23
34	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: <scp>CANDLE</scp>/<scp>PRAAS</scp>, <scp>SAVI</scp>, and <scp>AGS</scp>. Arthritis and Rheumatology, 2022, 74, 735-751.	5.6	23
35	Dysregulation of the immune system in Aicardi-GoutiÃ¨res syndrome: another example in a TREX1-mutated patient. Lupus, 2013, 22, 1064-1069.	1.6	22
36	New Case of 4H Syndrome and a Review of the Literature. Pediatric Neurology, 2010, 42, 359-364.	2.1	21

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37	COL4A1-Related Disease: Raised Creatine Kinase and Cerebral Calcification as Useful Pointers. Neuropediatrics, 2012, 43, 283-288.	0.6	20
38	Timeâ€course of myelination and atrophy on cerebral imaging in 35 patients with <scp>PLP</scp>1â€related disorders. Developmental Medicine and Child Neurology, 2016, 58, 706-713.	2.1	20
39	Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G > A mitochondrial DNA mutation: a case report. BMC Neurology, 2011, 11, 85.	1.8	18
40	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	3.7	18
41	Phenotypic spectrum of short-chain enoyl-Coa hydratase-1 (ECHS1) deficiency. European Journal of Paediatric Neurology, 2020, 28, 151-158.	1.6	18
42	Late-Onset Aicardi-GoutiÃ¨res Syndrome: A Characterization of Presenting Clinical Features. Pediatric Neurology, 2021, 115, 1-6.	2.1	18
43	Neurotransmitter abnormalities and response to supplementation in SPG11. Molecular Genetics and Metabolism, 2012, 107, 229-233.	1.1	17
44	Cystic leukoencephalopathy with cortical dysplasia related to LAMB1 mutations. Neurology, 2015, 84, 2195-2197.	1.1	17
45	Early-onset progressive spastic paraplegia caused by a novel TUBB4A mutation: brain MRI and FDG-PET findings. Journal of Neurology, 2016, 263, 591-593.	3.6	17
46	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
47	Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome. Molecular Genetics and Metabolism, 2022, 135, 109-113.	1.1	17
48	Impact of COVID-19 lockdown in children with neurological disorders in Italy. Disability and Health Journal, 2021, 14, 101053.	2.8	16
49	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	2.3	15
50	Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1136-e1147.	3.6	15
51	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	3.9	13
52	Thiamine-responsive disease due to mutation of tpk1 : Importance of avoiding misdiagnosis. Neurology, 2017, 89, 870-871.	1.1	13
53	Novel mutations in SLC16A2 associated with a less severe phenotype of MCT8 deficiency. Metabolic Brain Disease, 2019, 34, 1565-1575.	2.9	12
54	Ruxolitinib in Aicardi-GoutiÃ¨res syndrome. Metabolic Brain Disease, 2021, 36, 859-863.	2.9	12

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55	Spinal cord involvement and paroxysmal events in â€œInfantile Onset Transient Hypomyelinationâ€•due to TMEM63A mutation. Journal of Human Genetics, 2021, 66, 1035-1037.	2.3	12
56	Neurodevelopmental outcome of preterm very low birth weight infants admitted to an Italian tertiary center over an 11-year period. Scientific Reports, 2021, 11, 16316.	3.3	11
57	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-GoutiÃ¨res syndrome: Diagnostic and disease-monitoring implications. Molecular Genetics and Metabolism, 2019, 126, 489-494.	1.1	10
58	PRKRAâ€Related Disorders: Bilateral Striatal Degeneration in Addition to DYT16 Spectrum. Movement Disorders, 2021, 36, 1038-1040.	3.9	10
59	Spinal Cord Calcification in an Early-Onset Progressive Leukoencephalopathy. Journal of Child Neurology, 2011, 26, 876-880.	1.4	9
60	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	2.1	9
61	Mucopolysaccharidosis-Plus Syndrome, a Rapidly Progressive Disease: Favorable Impact of a Very Prolonged Steroid Treatment on the Clinical Course in a Child. Genes, 2022, 13, 442.	2.4	8
62	A novel mutation in COL4A1 gene: A possible cause of early postnatal cerebrovascular events. American Journal of Medical Genetics, Part A, 2015, 167, 810-815.	1.2	7
63	Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease. Neurological Sciences, 2016, 37, 973-977.	1.9	7
64	Cortical malformations and COL4A1 mutation: Three new cases. European Journal of Paediatric Neurology, 2019, 23, 410-417.	1.6	7
65	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
66	Alexander disease evolution over time: data from an Italian cohort of pediatric-onset patients. Molecular Genetics and Metabolism, 2021, 134, 353-358.	1.1	6
67	Novel Hypomyelinating Leukoencephalopathy Affecting Early Myelinating Structures: Clinical Course in Two Brothers. Neuropediatrics, 2013, 44, 213-217.	0.6	4
68	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. Journal of Child Neurology, 2015, 30, 1800-1805.	1.4	3
69	Hydroxychloroquine modulates immunological pathways activated by RNA:DNA hybrids in Aicardiâ€“GoutiÃ¨res syndrome patients carrying RNASEH2 mutations. Cellular and Molecular Immunology, 2021, 18, 1593-1595.	10.5	3
70	Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in <scp>PLA</scp>2G6â€associated neurodegeneration. European Journal of Neurology, 2016, 23, e24-5.	3.3	2
71	The epileptology of Aicardi-GoutiÃ¨res syndrome: electro-clinical-radiological findings. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 197-209.	2.0	2
72	Correspondence on â€œExpanded phenotype of AARS1-related white matter diseaseâ€•by Helman etÂal. Genetics in Medicine, 2022, , .	2.4	2

#	Article	IF	Citations
73	Case Report: Novel Compound Heterozygous RNASEH2B Mutations Cause Aicardiâ€“GoutiÃ¨res Syndrome. Frontiers in Immunology, 2021, 12, 672952.	4.8	1
74	How to look for intracranial calcification in children with neurological disorders: CT, MRI, or both of them?. Neurological Sciences, 2022, 43, 2043-2050.	1.9	1
75	Calcifying leukoencephalopathies: New overlapping phenotypes. American Journal of Medical Genetics, Part A, 2012, 158A, 964-965.	1.2	0
76	Leukodystrophies and Epilepsy. , 0, , .		0