Vincenzo Leuzzi

List of Publications by Year
in descending order

124 papers	3,675 citations	159585 30 h-index	168389 53 g-index
128 all docs	128 docs citations	128 times ranked	4871 citing authors

#	Article	IF	Citations
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756.	11.4	272
3	White matter pathology in phenylketonuriaâˆ†. Molecular Genetics and Metabolism, 2010, 99, S3-S9.	1.1	154
4	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
5	Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotype. Pediatric Neurology, 2012, 46, 24-31.	2.1	114
6	Intra-Erythrocyte Infusion of Dexamethasone Reduces Neurological Symptoms in Ataxia Teleangiectasia Patients: Results of a Phase 2 Trial. Orphanet Journal of Rare Diseases, 2014, 9, 5.	2.7	114
7	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25.	1.1	111
8	Creatine depletion in a new case with AGAT deficiency: clinical and genetic study in a large pedigree. Molecular Genetics and Metabolism, 2002, 77, 326-331.	1.1	95
9	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
10	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	1.9	84
11	Living with phenylketonuria in adulthood: The PKU ATTITUDE study. Molecular Genetics and Metabolism Reports, 2018, 16, 39-45.	1.1	67
12	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. Molecular Genetics and Metabolism, 2019, 127, 12-22.	1.1	66
13	Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. Parkinsonism and Related Disorders, 2019, 61, 19-25.	2.2	64
14	Quantitative determination of guanidinoacetate and creatine in dried blood spot by flow injection analysis-electrospray tandem mass spectrometry. Clinica Chimica Acta, 2006, 364, 180-187.	1.1	61
15	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e98.	6.0	59
16	<scp>AADC</scp> deficiency from infancy to adulthood: Symptoms and developmental outcome in an international cohort of 63 patients. Journal of Inherited Metabolic Disease, 2020, 43, 1121-1130.	3.6	59
17	Neurocognitive and neuroimaging outcome of early treated young adult PKU patients: A longitudinal study. Molecular Genetics and Metabolism, 2015, 115, 84-90.	1.1	58
18	Guanidinoacetate and Creatine plus Creatinine Assessment in Physiologic Fluids: An Effective Diagnostic Tool for the Biochemical Diagnosis of Arginine:Glycine Amidinotransferase and Guanidinoacetate Methyltransferase Deficiencies. Clinical Chemistry, 2002, 48, 1772-1778.	3.2	57

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19	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
20	Inborn errors of creatine metabolism and epilepsy. Epilepsia, 2013, 54, 217-227.	5.1	54
21	Erythrocyte-mediated delivery of phenylalanine ammonia lyase for the treatment of phenylketonuria in BTBR-Pahenu2 mice. Journal of Controlled Release, 2014, 194, 37-44.	9.9	45
22	Treatment with<scp>l</scp>-Arginine improves neuropsychological disorders in a child with Creatine transporter defect. Neurocase, 2008, 14, 151-161.	0.6	44
23	Metabolic epilepsy: An update. Brain and Development, 2013, 35, 827-841.	1.1	43
24	A novel mouse model of creatine transporter deficiency. F1000Research, 2014, 3, 228.	1.6	42
25	Microdeletions ofELP4Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. Human Mutation, 2015, 36, 842-850.	2.5	41
26	A mouse model for creatine transporter deficiency reveals early onset cognitive impairment and neuropathology associated with brain aging. Human Molecular Genetics, 2016, 25, 4186-4200.	2.9	39
27	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
28	Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149.	2.7	36
29	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
30	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
31	Clinical and Biochemical Features in a Patient With Mitochondrial Fission Factor Gene Alteration. Frontiers in Genetics, 2018, 9, 625.	2.3	34
32	Psychiatric disorders in adolescent and young adult patients with phenylketonuria. Molecular Genetics and Metabolism, 2016, 117, 12-18.	1.1	32
33	Caenorhabditis elegans provides an efficient drug screening platform for GNAO1-related disorders and highlights the potential role of caffeine in controlling dyskinesia. Human Molecular Genetics, 2022, 31, 929-941.	2.9	32
34	The expanding spectrum of movement disorders in genetic epilepsies. Developmental Medicine and Child Neurology, 2020, 62, 178-191.	2.1	31
35	Adult cognitive outcomes in phenylketonuria: explaining causes of variability beyond average Phe levels. Orphanet Journal of Rare Diseases, 2019, 14, 273.	2.7	30
36	Report of Two Never Treated Adult Sisters with Aromatic l-Amino Acid Decarboxylase Deficiency: A Portrait of the Natural History of the Disease or an Expanding Phenotype?. JIMD Reports, 2014, 15, 39-45.	1.5	29

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37	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study. Molecular Genetics and Metabolism, 2015, 116, 171-177.	1.1	27
38	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	2.2	25
39	Predictability and inconsistencies of cognitive outcome in patients with phenylketonuria and personalised therapy: the challenge for the future guidelines. Journal of Medical Genetics, 2020, 57, 145-150.	3.2	24
40	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	3.6	23
41	Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124.	1.2	22
42	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	4.1	22
43	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
44	Guanidinoacetate and creatine plus creatinine assessment in physiologic fluids: an effective diagnostic tool for the biochemical diagnosis of arginine:glycine amidinotransferase and guanidinoacetate methyltransferase deficiencies. Clinical Chemistry, 2002, 48, 1772-8.	3.2	21
45	In vivo effects of dexamethasone on blood gene expression in ataxia telangiectasia. Molecular and Cellular Biochemistry, 2018, 438, 153-166.	3.1	20
46	Arginine and glycine stimulate creatine synthesis in creatine transporter 1-deficient lymphoblasts. Analytical Biochemistry, 2008, 375, 153-155.	2.4	19
47	Very early pattern of movement disorders in sepiapterin reductase deficiency. Neurology, 2013, 81, 2141-2142.	1.1	19
48	A novel compound heterozygous genotype associated with aromatic amino acid decarboxylase deficiency: Clinical aspects and biochemical studies. Molecular Genetics and Metabolism, 2019, 127, 132-137.	1.1	19
49	KCND3-Related Neurological Disorders: From Old to Emerging Clinical Phenotypes. International Journal of Molecular Sciences, 2020, 21, 5802.	4.1	19
50	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
51	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2015, 115, 157-160.	1.1	18
52	Erythrocyteâ€mediated delivery of recombinant enzymes. Journal of Inherited Metabolic Disease, 2016, 39, 519-530.	3.6	18
53	Development of global rating instruments for pediatric patients with ataxia telangiectasia. European Journal of Paediatric Neurology, 2016, 20, 140-146.	1.6	18
54	Predictability and inconsistencies in the cognitive outcome of early treated PKU patients. Journal of Inherited Metabolic Disease, 2017, 40, 793-799.	3.6	18

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55	A new therapy prevents intellectual disability in mouse with phenylketonuria. Molecular Genetics and Metabolism, 2018, 124, 39-49.	1.1	18
56	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. Metabolic Brain Disease, 2018, 33, 261-269.	2.9	18
57	In vitro study of uptake and synthesis of creatine and its precursors by cerebellar granule cells and astrocytes suggests some hypotheses on the physiopathology of the inherited disorders of creatine metabolism. BMC Neuroscience, 2012, 13, 41.	1.9	17
58	Neurotransmitter trafficking defect in a patient with clathrin (CLTC) variation presenting with intellectual disability and early-onset parkinsonism. Parkinsonism and Related Disorders, 2019, 61, 207-210.	2.2	17
59	New pathogenic variants in COQ4 cause ataxia and neurodevelopmental disorder without detectable CoQ10 deficiency in muscle or skin fibroblasts. Journal of Neurology, 2021, 268, 3381-3389.	3.6	17
60	Behavioral and Neurochemical Characterization of New Mouse Model of Hyperphenylalaninemia. PLoS ONE, 2013, 8, e84697.	2.5	17
61	Untreated PKU Patients without Intellectual Disability: What Do They Teach Us?. Nutrients, 2019, 11, 2572.	4.1	16
62	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	2.2	16
63	Progressive myoclonus epilepsy and ceroidolipofuscinosis 14: The multifaceted phenotypic spectrum of KCTD7-related disorders. European Journal of Medical Genetics, 2019, 62, 103591.	1.3	15
64	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
65	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
66	Acute ischemic stroke in childhood: a comprehensive review. European Journal of Pediatrics, 2022, 181, 45-58.	2.7	15
67	Aromatic L-amino Acid Decarboxylase (AADC) deficiency: results from an Italian modified Delphi consensus. Italian Journal of Pediatrics, 2021, 47, 13.	2.6	15
68	A Nervous System-Specific Model of Creatine Transporter Deficiency Recapitulates the Cognitive Endophenotype of the Disease: a Longitudinal Study. Scientific Reports, 2019, 9, 62.	3.3	14
69	Parkinsonism in children: Clinical classification and etiological spectrum. Parkinsonism and Related Disorders, 2021, 82, 150-157.	2.2	14
70	A mutation on exon 6 of guanidinoacetate methyltransferase (GAMT) gene supports a different function for isoform a and b of GAMT enzyme. Molecular Genetics and Metabolism, 2006, 87, 88-90.	1.1	13
71	Severe early onset ethylmalonic encephalopathy with West syndrome. Metabolic Brain Disease, 2015, 30, 1537-1545.	2.9	13
72	ATM splicing variants as biomarkers for low dose dexamethasone treatment of A-T. Orphanet Journal of Rare Diseases, 2017, 12, 126.	2.7	13

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73	Brain <scp>MR</scp> patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1070-1082.	3.6	13
74	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
75	Expanding the genetic and phenotypic spectrum of <scp>CHD2</scp>â€related disease: From early neurodevelopmental disorders to adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 522-533.	1.2	13
76	PRICKLE1â€related early onset epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 2841-2845.	1.2	12
77	Treatable Inherited Movement Disorders in Children: Spotlight on Clinical and Biochemical Features. Movement Disorders Clinical Practice, 2020, 7, 154-166.	1.5	12
78	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
79	Italian national consensus statement on management and pharmacological treatment of phenylketonuria. Orphanet Journal of Rare Diseases, 2021, 16, 476.	2.7	12
80	Ataxia-telangiectasia. Neurology: Genetics, 2018, 4, e228.	1.9	11
81	Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. Molecular Genetics and Metabolism, 2020, 131, 155-162.	1.1	11
82	The management of phenylketonuria in adult patients in Italy: a survey of six specialist metabolic centers. Current Medical Research and Opinion, 2021, 37, 411-421.	1.9	11
83	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	7.6	11
84	Targeting mGlu5 Metabotropic Glutamate Receptors in the Treatment of Cognitive Dysfunction in a Mouse Model of Phenylketonuria. Frontiers in Neuroscience, 2018, 12, 154.	2.8	10
85	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. International Journal of Molecular Sciences, 2021, 22, 4202.	4.1	10
86	Compound heterozygosis in AADC deficiency: A complex phenotype dissected through comparison among heterodimeric and homodimeric AADC proteins. Molecular Genetics and Metabolism, 2021, 134, 147-155.	1.1	10
87	Age-Related Psychophysiological Vulnerability to Phenylalanine in Phenylketonuria. Frontiers in Pediatrics, 2014, 2, 57.	1.9	9
88	Clinical characterization of tremor in patients with phenylketonuria. Molecular Genetics and Metabolism, 2019, 128, 53-56.	1.1	9
89	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	2.5	9
90	Inborn errors of creatine metabolism and epilepsy: clinical features, diagnosis, and treatment. Journal of Child Neurology, 2002, 17 Suppl 3, 3S89-97; discussion 3S97.	1.4	9

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91	Development of a new UPLC-ESI-MS/MS method for the determination of biopterin and neopterin in dried blood spot. Clinica Chimica Acta, 2017, 466, 145-151.	1.1	8
92	Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684.	11.4	8
93	Urinary Neopterin and Phenylalanine Loading Test as Tools for the Biochemical Diagnosis of Segawa Disease. JIMD Reports, 2012, 7, 67-75.	1.5	7
94	Paradoxical sleep deprivation in rats causes a selective reduction in the expression of type-2 metabotropic glutamate receptors in the hippocampus. Pharmacological Research, 2017, 117, 46-53.	7.1	7
95	Parkinsonism, Intellectual Disability, and Catatonia in a Young Male With MECP2 Variant. Movement Disorders Clinical Practice, 2020, 7, 118-119.	1.5	7
96	Cognitive Outcomes and Relationships with Phenylalanine in Phenylketonuria: A Comparison between Italian and English Adult Samples. Nutrients, 2020, 12, 3033.	4.1	7
97	Missense PDSS1 mutations in CoenzymeQ10 synthesis cause optic atrophy and sensorineural deafness. Annals of Clinical and Translational Neurology, 2021, 8, 247-251.	3.7	7
98	Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. Journal of Neurology, 2022, 269, 1476-1484.	3.6	7
99	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	3.6	7
100	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. Cerebellum, 2022, 21, 1144-1150.	2.5	7
101	Validity and reliability of Italian version of the Non-Communicating Children's Pain Checklist: revised version. European Journal of Physical and Rehabilitation Medicine, 2019, 55, 89-94.	2.2	6
102	A next generation sequencingâ€based analysis of a large cohort of ataxic patients refines the clinical spectrum associated with spinocerebellar ataxia 21. European Journal of Neurology, 2021, 28, 2784-2788.	3.3	6
103	Uniparental disomy of chromosome 1 unmasks recessive mutations of PPT1 in a boy with neuronal ceroid lipofuscinosis type 1. Brain and Development, 2017, 39, 182-183.	1.1	5
104	Intellectual Disability and Brain Creatine Deficit: Phenotyping of the Genetic Mouse Model for GAMT Deficiency. Genes, 2021, 12, 1201.	2.4	4
105	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. Molecular Genetics and Metabolism Reports, 2019, 21, 100502.	1.1	3
106	KCNQ2 encephalopathy manifesting with Rett-like features. Neurology: Genetics, 2020, 6, e510.	1.9	3
107	Executive functioning, adaptive skills, emotional and behavioral profile: A comparison between autism spectrum disorder and phenylketonuria. Molecular Genetics and Metabolism Reports, 2020, 23, 100577.	1.1	3
108	Multiple sclerosis and intracellular cobalamin defect (MMACHC/PRDX1) comorbidity in a young male. Molecular Genetics and Metabolism Reports, 2020, 22, 100560.	1.1	3

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109	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. American Journal of Medical Genetics, Part A, 2021, 185, 1575-1581.	1.2	3
110	Presenting Patterns of Genetically Determined Developmental Encephalopathies With Epilepsy and Movement Disorders: A Single Tertiary Center Retrospective Cohort Study. Frontiers in Neurology, 0, 13, .	2.4	3
111	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. Journal of Pediatric Neurology, 2015, 13, 213-224.	0.2	2
112	A novel developmental encephalopathy with epilepsy and hyperkinetic movement disorders associated with a deletion of the sodium channel gene cluster on chromosome 2q24.3. Parkinsonism and Related Disorders, 2019, 68, 1-3.	2.2	2
113	<scp>AP</scp>1S2â€truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	1.5	2
114	TSC1 as a Novel Gene for Sleep-Related Hypermotor Epilepsy: A Child with a Mild Phenotype of Tuberous Sclerosis. Neuropediatrics, 2021, 52, 146-149.	0.6	2
115	Simultaneous determination of 5-hydroxytryptophan and 3-O-methyldopa in dried blood spot by UPLC-MS/MS: A useful tool for the diagnosis of L-amino acid decarboxylase deficiency. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2021, 1185, 122999.	2.3	2
116	3-Methylglutaconic Aciduria Type I Due to AUH Defect: The Case Report of a Diagnostic Odyssey and a Review of the Literature. International Journal of Molecular Sciences, 2022, 23, 4422.	4.1	2
117	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. Movement Disorders, 2014, 29, 277-278.	3.9	1
118	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency. European Journal of Pediatrics, 2017, 176, 917-924.	2.7	1
119	Creatine Levels in Patients with Phenylketonuria and Mild Hyperphenylalaninemia: A Pilot Study. Life, 2021, 11, 425.	2.4	1
120	Engineering new metabolic pathways in isolated cells for the degradation of guanidinoacetic acid and simultaneous production of creatine. Molecular Therapy - Methods and Clinical Development, 2022, 25, 26-40.	4.1	1
121	Neurodevelopmental Impairment As the Main Phenotypic Hallmark Associated with the Translocation t(7;10)(7p22.3;q26.11). Journal of Pediatric Genetics, 2020, 11, 68-73.	0.7	0
122	â€œProtenuria in SLE: Is it always lupus?â€• Lupus, 2021, 30, 664-668.	1.6	0
123	Functional Classification of the ATM Variant c.7157C>A and In Vitro Effects of Dexamethasone. Frontiers in Genetics, 2021, 12, 759467.	2.3	0
124	Looking back at the neonatal period in early-treated phenylketonuric patients. Pediatric Research, 2022, , .	2.3	0