Vincenzo Leuzzi

List of Publications by Citations

123 papers	2,263 citations	25 h-index	43 g-index
128 ext. papers	3,001 ext. citations	4.4 avg, IF	4.78 L-index

#	Paper	IF	Citations
123	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
122	Key European guidelines for the diagnosis and management of patients with phenylketonuria. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 743-756	18.1	180
121	White matter pathology in phenylketonuria. *Molecular Genetics and Metabolism*, **2010**, 99 Suppl 1, S3-9	3.7	114
120	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	4.2	96
119	Genes of early-onset epileptic encephalopathies: from genotype to phenotype. *Pediatric Neurology*, **2012**, 46, 24-31	2.9	88
118	Creatine depletion in a new case with AGAT deficiency: clinical and genetic study in a large pedigree. *Molecular Genetics and Metabolism*, **2002**, 77, 326-31	3.7	86
117	Guanidinoacetate methyltransferase (GAMT) deficiency: outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. *Molecular Genetics and Metabolism*, **2014**, 111, 16-25	3.7	77
116	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	5.5	55
115	Quantitative determination of guanidinoacetate and creatine in dried blood spot by flow injection analysis-electrospray tandem mass spectrometry. *Clinica Chimica Acta*, **2006**, 364, 180-7	6.2	54
114	encephalopathy: Broadening the phenotype and evaluating treatment and outcome. *Neurology: Genetics*, **2017**, 3, e143	3.8	51
113	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. *Neurology: Neuroimmunology and NeuroInflammation*, **2015**, 2, e98	9.1	45
112	Neurocognitive and neuroimaging outcome of early treated young adult PKU patients: A longitudinal study. *Molecular Genetics and Metabolism*, **2015**, 115, 84-90	3.7	44
111	The Genetic Landscape and Epidemiology of Phenylketonuria. *American Journal of Human Genetics*, **2020**, 107, 234-250	11	44
110	Treatment with L-arginine improves neuropsychological disorders in a child with creatine transporter defect. *Neurocase*, **2008**, 14, 151-61	0.8	38
109	Inborn errors of creatine metabolism and epilepsy. *Epilepsia*, **2013**, 54, 217-27	6.4	37
108	Living with phenylketonuria in adulthood: The PKU ATTITUDE study. *Molecular Genetics and Metabolism Reports*, **2018**, 16, 39-45	1.8	36
107	Metabolic epilepsy: an update. *Brain and Development*, **2013**, 35, 827-41	2.2	36

106	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. *Molecular Genetics and Metabolism*, **2019**, 127, 12-22	3.7	34
105	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A single-center cohort study. *Movement Disorders*, **2019**, 34, 1516-1527	7	32
104	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH) deficiencies. *Orphanet Journal of Rare Diseases*, **2020**, 15, 126	4.2	31
103	Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. *Human Mutation*, **2015**, 36, 842-50	4.7	31
102	Erythrocyte-mediated delivery of phenylalanine ammonia lyase for the treatment of phenylketonuria in BTBR-Pah(enu2) mice. *Journal of Controlled Release*, **2014**, 194, 37-44	11.7	31
101	Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. *Parkinsonism and Related Disorders*, **2019**, 61, 19-25	3.6	31
100	Clinical and Biochemical Features in a Patient With Gene Alteration. *Frontiers in Genetics*, **2018**, 9, 625	4.5	27
99	AADC 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	5.4	26
98	A novel mouse model of creatine transporter deficiency. *F1000Research*, **2014**, 3, 228	3.6	25
97	Newborn screening for homocystinurias: Recent recommendations versus current practice. *Journal of Inherited Metabolic Disease*, **2019**, 42, 128-139	5.4	24
96	Psychiatric disorders in adolescent and young adult patients with phenylketonuria. *Molecular Genetics and Metabolism*, **2016**, 117, 12-8	3.7	24
95	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*, **2015**, 15, 39-45	1.9	24
94	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	5.6	23
93	Epilepsy in KCNH1-related syndromes. *Epileptic Disorders*, **2016**, 18, 123-36	1.9	21
92	Can untreated PKU patients escape from intellectual disability? A systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 149	4.2	21
91	In vivo effects of dexamethasone on blood gene expression in ataxia telangiectasia. *Molecular and Cellular Biochemistry*, **2018**, 438, 153-166	4.2	19
90	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	5.5	19
89	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study. *Molecular Genetics and Metabolism*, **2015**, 116, 171-7	3.7	18

88	Arginine and glycine stimulate creatine synthesis in creatine transporter 1-deficient lymphoblasts. *Analytical Biochemistry*, **2008**, 375, 153-5	3.1	18
87	A new therapy prevents intellectual disability in mouse with phenylketonuria. *Molecular Genetics and Metabolism*, **2018**, 124, 39-49	3.7	16
86	Very early pattern of movement disorders in sepiapterin reductase deficiency. *Neurology*, **2013**, 81, 2141-43	6.5	16
85	Behavioral and neurochemical characterization of new mouse model of hyperphenylalaninemia. *PLoS ONE*, **2013**, 8, e84697	3.7	16
84	Erythrocyte-mediated delivery of recombinant enzymes. *Journal of Inherited Metabolic Disease*, **2016**, 39, 519-30	5.4	16
83	Adult cognitive outcomes in phenylketonuria: explaining causes of variability beyond average Phe levels. *Orphanet Journal of Rare Diseases*, **2019**, 14, 273	4.2	16
82	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. *Molecular Genetics and Metabolism*, **2015**, 115, 157-60	3.7	15
81	The expanding spectrum of movement disorders in genetic epilepsies. *Developmental Medicine and Child Neurology*, **2020**, 62, 178-191	3.3	15
80	Predictability and inconsistencies in the cognitive outcome of early treated PKU patients. *Journal of Inherited Metabolic Disease*, **2017**, 40, 793-799	5.4	14
79	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	3.2	14
78	Cognitive, adaptive, and behavioral features in Joubert syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3115-3124	2.5	13
77	Development of global rating instruments for pediatric patients with ataxia telangiectasia. *European Journal of Paediatric Neurology*, **2016**, 20, 140-6	3.8	13
76	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. *Parkinsonism and Related Disorders*, **2019**, 68, 8-16	3.6	13
75	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	3.7	12
74	Severe early onset ethylmalonic encephalopathy with West syndrome. *Metabolic Brain Disease*, **2015**, 30, 1537-45	3.9	11
73	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	5.8	11
72	Untreated PKU Patients without Intellectual Disability: What Do They Teach Us?. *Nutrients*, **2019**, 11,	6.7	10
71	Progressive myoclonus epilepsy and ceroidolipofuscinosis 14: The multifaceted phenotypic spectrum of KCTD7-related disorders. *European Journal of Medical Genetics*, **2019**, 62, 103591	2.6	10

70	Delineating the neurological phenotype in children with defects in the ECHS1 or HIBCH gene. *Journal of Inherited Metabolic Disease*, **2021**, 44, 401-414	5.4	10
69	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. *Parkinsonism and Related Disorders*, **2020**, 72, 75-79	3.6	9
68	ATM splicing variants as biomarkers for low dose dexamethasone treatment of A-T. *Orphanet Journal of Rare Diseases*, **2017**, 12, 126	4.2	9
67	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	3.7	9
66	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	3.6	9
65	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. *Metabolic Brain Disease*, **2018**, 33, 261-269	3.9	9
64	Inborn errors of creatine metabolism and epilepsy: clinical features, diagnosis, and treatment. *Journal of Child Neurology*, **2002**, 17 Suppl 3, 3S89-97; discussion 3S97	2.5	9
63	Ataxia-telangiectasia: A new remitting form with a peculiar transcriptome signature. *Neurology: Genetics*, **2018**, 4, e228	3.8	8
62	Age-related psychophysiological vulnerability to phenylalanine in phenylketonuria. *Frontiers in Pediatrics*, **2014**, 2, 57	3.4	8
61	-Related Neurological Disorders: From Old to Emerging Clinical Phenotypes. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	8
60	PRICKLE1-related early onset epileptic encephalopathy. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2841-2845	2.5	8
59	Treatable Inherited Movement Disorders in Children: Spotlight on Clinical and Biochemical Features. *Movement Disorders Clinical Practice*, **2020**, 7, 154-166	2.2	7
58	Targeting mGlu5 Metabotropic Glutamate Receptors in the Treatment of Cognitive Dysfunction in a Mouse Model of Phenylketonuria. *Frontiers in Neuroscience*, **2018**, 12, 154	5.1	7
57	New pathogenic variants in COQ4 cause ataxia and neurodevelopmental disorder without detectable CoQ deficiency in muscle or skin fibroblasts. *Journal of Neurology*, **2021**, 268, 3381-3389	5.5	7
56	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	6.2	6
55	Issues with European guidelines for phenylketonuria - AuthorsPreply. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 683-684	18.1	6
54	Urinary neopterin and phenylalanine loading test as tools for the biochemical diagnosis of segawa disease. *JIMD Reports*, **2013**, 7, 67-75	1.9	6
53	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. *Nephrology Dialysis Transplantation*, **2020**, 35, 1195-1202	4.3	6

52	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	10.2	5
51	Clinical characterization of tremor in patients with phenylketonuria. *Molecular Genetics and Metabolism*, **2019**, 128, 53-56	3.7	5
50	Parkinsonism, Intellectual Disability, and Catatonia in a Young Male With Variant. *Movement Disorders Clinical Practice*, **2020**, 7, 118-119	2.2	5
49	Parkinsonism in children: Clinical classification and etiological spectrum. *Parkinsonism and Related Disorders*, **2021**, 82, 150-157	3.6	5
48	Brain MR patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. *Journal of Inherited Metabolic Disease*, **2021**, 44, 1070-1082	5.4	5
47	Aromatic L-amino Acid Decarboxylase (AADC) deficiency: results from an Italian modified Delphi consensus. *Italian Journal of Pediatrics*, **2021**, 47, 13	3.2	5
46	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	2.2	4
45	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*, **2021**,	5.6	4
44	Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. *Molecular Genetics and Metabolism*, **2020**, 131, 155-162	3.7	4
43	A next generation sequencing-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	6	4
42	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
41	Acute ischemic stroke in childhood: a comprehensive review. *European Journal of Pediatrics*, **2021**, 1	4.1	4
40	A Nervous System-Specific Model of Creatine Transporter Deficiency Recapitulates the Cognitive Endophenotype of the Disease: a Longitudinal Study. *Scientific Reports*, **2019**, 9, 62	4.9	4
39	haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. *Journal of Medical Genetics*, **2021**,	5.8	3
38	Refining the mutational spectrum and gene-phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. *Journal of Medical Genetics*, **2021**,	5.8	3
37	Validity and reliability of Italian version of the Non-Communicating Children‚Ä≤s Pain Checklist: revised version. *European Journal of Physical and Rehabilitation Medicine*, **2019**, 55, 89-94	4.4	3
36	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1575-1581	2.5	3
35	NGS in Hereditary Ataxia: When Rare Becomes Frequent. *International Journal of Molecular Sciences* , **2021**, 22,	6.3	3

34	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	3.7	3
33	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. *Nature Communications*, **2021**, 12, 5529	17.4	3
32	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. *Molecular Genetics and Metabolism Reports*, **2019**, 21, 100502	1.8	2
31	Executive functioning, adaptive skills, emotional and behavioral profile: A comparison between autism spectrum disorder and phenylketonuria. *Molecular Genetics and Metabolism Reports*, **2020**, 23, 100577	1.8	2
30	Multiple sclerosis and intracellular cobalamin defect (/) comorbidity in a young male. *Molecular Genetics and Metabolism Reports*, **2020**, 22, 100560	1.8	2
29	Expanding the genetic and phenotypic spectrum of CHD2-related disease: From early neurodevelopmental disorders to adult-onset epilepsy. *American Journal of Medical Genetics, Part A* , **2021**,	2.5	2
28	Cognitive Outcomes and Relationships with Phenylalanine in Phenylketonuria: A Comparison between Italian and English Adult Samples. *Nutrients*, **2020**, 12,	6.7	2
27	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
26	Missense PDSS1 mutations in CoenzymeQ10 synthesis cause optic atrophy and sensorineural deafness. *Annals of Clinical and Translational Neurology*, **2021**, 8, 247-251	5.3	2
25	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. *Brain*, **2021**,	11.2	2
24	TSC1 as a Novel Gene for Sleep-Related Hypermotor Epilepsy: A Child with a Mild Phenotype of Tuberous Sclerosis. *Neuropediatrics*, **2021**, 52, 146-149	1.6	2
23	Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. *Journal of Neurology*, **2021**, 1	5.5	2
22	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	3.6	1
21	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. *Journal of Pediatric Neurology*, **2015**, 13, 213-224	0.2	1
20	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. *Movement Disorders*, **2014**, 29, 277-8	7	1
19	Clinical variability at the mild end of BRAT1-related spectrum: Evidence from two families with genotype-phenotype discordance. *Human Mutation*, **2021**,	4.7	1
18	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. *Cerebellum*, **2021**, 1	4.3	1
17	Italian national consensus statement on management and pharmacological treatment of phenylketonuria. *Orphanet Journal of Rare Diseases*, **2021**, 16, 476	4.2	1

16	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	3.2	1
15	KCNQ2 encephalopathy manifesting with Rett-like features: A follow-up into adulthood. *Neurology: Genetics*, **2020**, 6, e510	3.8	1
14	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. *Brain*, **2021**, 144, 3020-3035	11.2	1
13	AP1S2-truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. *Acta Paediatrica, International Journal of Paediatrics*, **2019**, 108, 564-565	3.1	1
12	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	2.5	1
11	Intellectual Disability and Brain Creatine Deficit: Phenotyping of the Genetic Mouse Model for GAMT Deficiency. *Genes*, **2021**, 12,	4.2	1
10	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency. *European Journal of Pediatrics*, **2017**, 176, 917-924	4.1	0
9	Assessment of intellectual impairment, health-related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the iNTD registry. *Journal of Inherited Metabolic Disease*, **2021**, 44, 1489-1502	5.4	0
8	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clinical Epigenetics*, **2021**, 13, 157	7.7	0
7	3-Methylglutaconic Aciduria Type I Due to Defect: The Case Report of a Diagnostic Odyssey and a Review of the Literature.. *International Journal of Molecular Sciences*, **2022**, 23,	6.3	0
6	Metabolic Causes of Epilepsy **2015**, 71-99		
5	Functional Classification of the ATM Variant c.7157C>A and Effects of Dexamethasone. *Frontiers in Genetics*, **2021**, 12, 759467	4.5	
4	Skeletal Findings in Neurometabolic Disease117-122		
3	Neurodevelopmental Impairment As the Main Phenotypic Hallmark Associated with the Translocation t(7;10)(7p22.3;q26.11).. *Journal of Pediatric Genetics*, **2022**, 11, 68-73	0.7	
2	"Protenuria in SLE: Is it always lupus?". *Lupus*, **2021**, 30, 664-668	2.6	
1	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	6.4	