Vincenzo Leuzzi

List of Publications by Year in Descending Order

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	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	
122	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	
121	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
120	Clinical variability at the mild end of BRAT1-related spectrum: Evidence from two families with genotype-phenotype discordance. *Human Mutation*, **2021**,	4.7	1
119	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. *Cerebellum*, **2021**, 1	4.3	1
118	Italian national consensus statement on management and pharmacological treatment of phenylketonuria. *Orphanet Journal of Rare Diseases*, **2021**, 16, 476	4.2	1
117	haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. *Journal of Medical Genetics*, **2021**,	5.8	3
116	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
115	Functional Classification of the ATM Variant c.7157C>A and Effects of Dexamethasone. *Frontiers in Genetics*, **2021**, 12, 759467	4.5	
114	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
113	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
112	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
111	Refining the mutational spectrum and gene-phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. *Journal of Medical Genetics*, **2021**,	5.8	3
110	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
109	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
108	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
107	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. *Brain*, **2021**, 144, 3020-3035	11.2	1

106	Acute ischemic stroke in childhood: a comprehensive review. *European Journal of Pediatrics*, **2021**, 1	4.1	4
105	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
104	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
103	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
102	Parkinsonism in children: Clinical classification and etiological spectrum. *Parkinsonism and Related Disorders*, **2021**, 82, 150-157	3.6	5
101	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. *Brain*, **2021**,	11.2	2
100	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
99	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
98	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1575-1581	2.5	3
97	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
96	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
95	NGS in Hereditary Ataxia: When Rare Becomes Frequent. *International Journal of Molecular Sciences* , **2021**, 22,	6.3	3
94	Intellectual Disability and Brain Creatine Deficit: Phenotyping of the Genetic Mouse Model for GAMT Deficiency. *Genes*, **2021**, 12,	4.2	1
93	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
92	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clinical Epigenetics*, **2021**, 13, 157	7.7	0
91	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. *Nature Communications*, **2021**, 12, 5529	17.4	3
90	Aromatic L-amino Acid Decarboxylase (AADC) deficiency: results from an Italian modified Delphi consensus. *Italian Journal of Pediatrics*, **2021**, 47, 13	3.2	5
89	"Protenuria in SLE: Is it always lupus?". *Lupus*, **2021**, 30, 664-668	2.6	

88	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
87	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH) deficiencies. *Orphanet Journal of Rare Diseases*, **2020**, 15, 126	4.2	31
86	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
85	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
84	Treatable Inherited Movement Disorders in Children: Spotlight on Clinical and Biochemical Features. *Movement Disorders Clinical Practice*, **2020**, 7, 154-166	2.2	7
83	Multiple sclerosis and intracellular cobalamin defect (/) comorbidity in a young male. *Molecular Genetics and Metabolism Reports*, **2020**, 22, 100560	1.8	2
82	Parkinsonism, Intellectual Disability, and Catatonia in a Young Male With Variant. *Movement Disorders Clinical Practice*, **2020**, 7, 118-119	2.2	5
81	The expanding spectrum of movement disorders in genetic epilepsies. *Developmental Medicine and Child Neurology*, **2020**, 62, 178-191	3.3	15
80	KCNQ2 encephalopathy manifesting with Rett-like features: A follow-up into adulthood. *Neurology: Genetics*, **2020**, 6, e510	3.8	1
79	Cognitive Outcomes and Relationships with Phenylalanine in Phenylketonuria: A Comparison between Italian and English Adult Samples. *Nutrients*, **2020**, 12,	6.7	2
78	The Genetic Landscape and Epidemiology of Phenylketonuria. *American Journal of Human Genetics*, **2020**, 107, 234-250	11	44
77	Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. *Molecular Genetics and Metabolism*, **2020**, 131, 155-162	3.7	4
76	-Related Neurological Disorders: From Old to Emerging Clinical Phenotypes. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	8
75	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
74	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
73	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
72	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A single-center cohort study. *Movement Disorders*, **2019**, 34, 1516-1527	7	32
71	Clinical characterization of tremor in patients with phenylketonuria. *Molecular Genetics and Metabolism*, **2019**, 128, 53-56	3.7	5

70	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
69	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. *Molecular Genetics and Metabolism*, **2019**, 127, 12-22	3.7	34
68	Newborn screening for homocystinurias: Recent recommendations versus current practice. *Journal of Inherited Metabolic Disease*, **2019**, 42, 128-139	5.4	24
67	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
66	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
65	Untreated PKU Patients without Intellectual Disability: What Do They Teach Us?. *Nutrients*, **2019**, 11,	6.7	10
64	Adult cognitive outcomes in phenylketonuria: explaining causes of variability beyond average Phe levels. *Orphanet Journal of Rare Diseases*, **2019**, 14, 273	4.2	16
63	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	4.4	3
62	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
61	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
60	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
59	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
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	3.6	9
57	Ataxia-telangiectasia: A new remitting form with a peculiar transcriptome signature. *Neurology: Genetics*, **2018**, 4, e228	3.8	8
56	A new therapy prevents intellectual disability in mouse with phenylketonuria. *Molecular Genetics and Metabolism*, **2018**, 124, 39-49	3.7	16
55	In vivo effects of dexamethasone on blood gene expression in ataxia telangiectasia. *Molecular and Cellular Biochemistry*, **2018**, 438, 153-166	4.2	19
54	Living with phenylketonuria in adulthood: The PKU ATTITUDE study. *Molecular Genetics and Metabolism Reports*, **2018**, 16, 39-45	1.8	36
53	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

52	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
51	Clinical and Biochemical Features in a Patient With Gene Alteration. *Frontiers in Genetics*, **2018**, 9, 625	4.5	27
50	Can untreated PKU patients escape from intellectual disability? A systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 149	4.2	21
49	PRICKLE1-related early onset epileptic encephalopathy. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2841-2845	2.5	8
48	Key European guidelines for the diagnosis and management of patients with phenylketonuria. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 743-756	18.1	180
47	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
46	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
45	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency. *European Journal of Pediatrics*, **2017**, 176, 917-924	4.1	0
44	encephalopathy: Broadening the phenotype and evaluating treatment and outcome. *Neurology: Genetics*, **2017**, 3, e143	3.8	51
43	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
42	ATM splicing variants as biomarkers for low dose dexamethasone treatment of A-T. *Orphanet Journal of Rare Diseases*, **2017**, 12, 126	4.2	9
41	Predictability and inconsistencies in the cognitive outcome of early treated PKU patients. *Journal of Inherited Metabolic Disease*, **2017**, 40, 793-799	5.4	14
40	Issues with European guidelines for phenylketonuria - AuthorsPreply. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 683-684	18.1	6
39	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
38	Cognitive, adaptive, and behavioral features in Joubert syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3115-3124	2.5	13
37	Development of global rating instruments for pediatric patients with ataxia telangiectasia. *European Journal of Paediatric Neurology*, **2016**, 20, 140-6	3.8	13
36	Psychiatric disorders in adolescent and young adult patients with phenylketonuria. *Molecular Genetics and Metabolism*, **2016**, 117, 12-8	3.7	24
35	Epilepsy in KCNH1-related syndromes. *Epileptic Disorders*, **2016**, 18, 123-36	1.9	21

34	Erythrocyte-mediated delivery of recombinant enzymes. *Journal of Inherited Metabolic Disease*, **2016**, 39, 519-30	5.4	16
33	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
32	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. *Molecular Genetics and Metabolism*, **2015**, 115, 157-60	3.7	15
31	Severe early onset ethylmalonic encephalopathy with West syndrome. *Metabolic Brain Disease*, **2015**, 30, 1537-45	3.9	11
30	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
29	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. *Journal of Pediatric Neurology*, **2015**, 13, 213-224	0.2	1
28	Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. *Human Mutation*, **2015**, 36, 842-50	4.7	31
27	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
26	Metabolic Causes of Epilepsy **2015**, 71-99		
25	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. *Neurology: Neuroimmunology and NeuroInflammation*, **2015**, 2, e98	9.1	45
24	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
23	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
22	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
21	Age-related psychophysiological vulnerability to phenylalanine in phenylketonuria. *Frontiers in Pediatrics*, **2014**, 2, 57	3.4	8
20	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. *Movement Disorders*, **2014**, 29, 277-8	7	1
19	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
18	A novel mouse model of creatine transporter deficiency. *F1000Research*, **2014**, 3, 228	3.6	25
17	Inborn errors of creatine metabolism and epilepsy. *Epilepsia*, **2013**, 54, 217-27	6.4	37

16	Metabolic epilepsy: an update. *Brain and Development*, **2013**, 35, 827-41	2.2	36
15	Very early pattern of movement disorders in sepiapterin reductase deficiency. *Neurology*, **2013**, 81, 214-62	6.5	16
14	Urinary neopterin and phenylalanine loading test as tools for the biochemical diagnosis of segawa disease. *JIMD Reports*, **2013**, 7, 67-75	1.9	6
13	Behavioral and neurochemical characterization of new mouse model of hyperphenylalaninemia. *PLoS ONE*, **2013**, 8, e84697	3.7	16
12	Genes of early-onset epileptic encephalopathies: from genotype to phenotype. *Pediatric Neurology*, **2012**, 46, 24-31	2.9	88
11	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
10	White matter pathology in phenylketonuria. *Molecular Genetics and Metabolism*, **2010**, 99 Suppl 1, S3-9	3.7	114
9	Treatment with L-arginine improves neuropsychological disorders in a child with creatine transporter defect. *Neurocase*, **2008**, 14, 151-61	0.8	38
8	Arginine and glycine stimulate creatine synthesis in creatine transporter 1-deficient lymphoblasts. *Analytical Biochemistry*, **2008**, 375, 153-5	3.1	18
7	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
6	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
5	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
4	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
3	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
2	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
1	Skeletal Findings in Neurometabolic Disease117-122		