Ivo BariÄ

List of Publications by Year in descending order

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60 papers	2,781 citations	201674 27 h-index	182427 51 g-index
65 all docs	65 docs citations	65 times ranked	4226 citing authors

#	Article	IF	Citations
1	Case Report: Advanced Skeletal Muscle Imaging in S-Adenosylhomocysteine Hydrolase Deficiency and Further Insight Into Muscle Pathology. Frontiers in Pediatrics, 2022, 10, 847445.	1.9	1
2	Longâ€Term Sebelipase Alfa Treatment in Children and Adults With Lysosomal Acid Lipase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2022, 74, 757-764.	1.8	6
3	Inborn Errors of Metabolism Associated With Autism Spectrum Disorders: Approaches to Intervention. Frontiers in Neuroscience, 2021, 15, 673600.	2.8	33
4	Early initiation of ambroxol treatment diminishes neurological manifestations of type 3 Gaucher disease: A long-term outcome of two siblings. European Journal of Paediatric Neurology, 2021, 32, 66-72.	1.6	8
5	Current Status of Newborn Screening in Southeastern Europe. Frontiers in Pediatrics, 2021, 9, 648939.	1.9	10
6	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	3.8	10
7	86â€…Floppy infant syndrome due to connective tissue disorder. Case report of a patient with kyphoscoliotic Ehlers-Danlos syndrome. , 2021, , .		0
8	101â€…Inherited autoinflammatory encephalopathy in the differential diagnosis of conatal viral infections- newborn with Aicardi-GoutiÃ¨res syndrome. , 2021, , .		0
9	ATP synthase deficiency due to m.8528T>C mutationÂâ€“ a novel cause of severe neonatal hyperammonemia requiring hemodialysis. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 389-393.	0.9	0
10	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
11	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	2.5	14
12	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
13	Diagnosis and the importance of early treatment of tyrosinemia type 1: A case report. Clinical Mass Spectrometry, 2019, 12, 1-6.	1.9	0
14	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	3.6	30
15	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
16	A novel PGAP3 mutation in a Croatian boy with brachytelephalangy and a thin corpus callosum. Human Genome Variation, 2018, 5, 18005.	0.7	17
17	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
18	Hypogammaglobulinemia and imaging features in a patient with infantile free sialic acid storage disease (ISSD) and a novel mutation in the SLC17A5 gene. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1155-1159.	0.9	1

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19	Metabolic follow-up of a Croatian patient with gyrate atrophy and a new mutation in the OAT gene: a case report. Biochemia Medica, 2018, 28, 030801.	2.7	3
20	Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. Human Mutation, 2017, 38, 524-531.	2.5	18
21	The ethical framework for performing research with rare inherited neurometabolic disease patients. European Journal of Pediatrics, 2017, 176, 395-405.	2.7	11
22	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
23	Consensus recommendations for the diagnosis, treatment and followâ€up of inherited methylation disorders. Journal of Inherited Metabolic Disease, 2017, 40, 5-20.	3.6	47
24	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. PLoS ONE, 2016, 11, e0167984.	2.5	21
25	Abnormal Hypermethylation at Imprinting Control Regions in Patients with S-Adenosylhomocysteine Hydrolase (AHCY) Deficiency. PLoS ONE, 2016, 11, e0151261.	2.5	8
26	Glycine N-Methyltransferase Deficiency: A Member of Dysmethylating Liver Disorders?. JIMD Reports, 2016, 31, 101-106.	1.5	11
27	Impact of age at onset and newborn screening on outcome in organic acidurias. Journal of Inherited Metabolic Disease, 2016, 39, 341-353.	3.6	60
28	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. Molecular Genetics and Metabolism, 2016, 119, 44-49.	1.1	9
29	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. American Journal of Human Genetics, 2016, 99, 894-902.	6.2	75
30	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	3.6	92
31	Behavioural and emotional problems, intellectual impairment and healthâ€related quality of life in patients with organic acidurias and urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 231-241.	3.6	29
32	Phenylketonuria screening and management in southeastern Europe â€“ survey results from 11 countries. Orphanet Journal of Rare Diseases, 2015, 10, 68.	2.7	32
33	Review and evaluation of the methodological quality of the existing guidelines and recommendations for inherited neurometabolic disorders. Orphanet Journal of Rare Diseases, 2015, 10, 164.	2.7	19
34	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	1.9	23
35	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34
36	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	6.2	110

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37	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	3.6	186
38	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	3.6	175
39	A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. New England Journal of Medicine, 2015, 373, 1010-1020.	27.0	212
40	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. European Journal of Human Genetics, 2014, 22, 762-767.	2.8	39
41	Sulphur Amino Acids. , 2014, , 33-46.		4
42	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
43	Newborn screening in southeastern Europe. Molecular Genetics and Metabolism, 2014, 113, 42-45.	1.1	45
44	Exome sequencing reveals mutated SLC19A3 in patients withan early-infantile, lethal, encephalopathy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 63-63.	0.0	0
45	Mitochondrial myopathy associated with a novel 5522G>A mutation in the mitochondrial tRNATrp gene. European Journal of Human Genetics, 2013, 21, 871-875.	2.8	12
46	Commentary. Clinical Chemistry, 2013, 59, 1164-1164.	3.2	0
47	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
48	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	3.2	78
49	Glyceroluria and Neonatal Hemochromatosis. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, e126-8.	1.8	4
50	Clinical picture of S-adenosylhomocysteine hydrolase deficiency resembles phosphomannomutase 2 deficiency. Molecular Genetics and Metabolism, 2012, 107, 611-613.	1.1	30
51	Plasma biomarker identification in Sâ€adenosylhomocysteine hydrolase deficiency. Electrophoresis, 2011, 32, 1970-1975.	2.4	3
52	Sâ€adenosylhomocysteine hydrolase deficiency: two siblings with fetal hydrops and fatal outcomes. Journal of Inherited Metabolic Disease, 2010, 33, 705-713.	3.6	35
53	Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I. Brain, 2009, 132, 1764-1782.	7.6	160
54	Inherited disorders in the conversion of methionine to homocysteine. Journal of Inherited Metabolic Disease, 2009, 32, 459-471.	3.6	49

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55	Genotype-predicted tetrahydrobiopterin (BH4)-responsiveness and molecular genetics in Croatian patients with phenylalanine hydroxylase (PAH) deficiency. Molecular Genetics and Metabolism, 2009, 97, 165-171.	1.1	65
56	Functional analysis of human S-adenosylhomocysteine hydrolase isoforms SAHH-2 and SAHH-3. European Journal of Human Genetics, 2007, 15, 347-351.	2.8	9
57	A single mutation at Tyr143 of human S-adenosylhomocysteine hydrolase renders the enzyme thermosensitive and affects the oxidation state of bound cofactor nicotinamideâ€“adenine dinucleotide. Biochemical Journal, 2006, 400, 245-253.	3.7	18
58	Studies of S-adenosylhomocysteine-hydrolase polymorphism in a Croatian population. Journal of Human Genetics, 2006, 51, 21-24.	2.3	4
59	S-adenosylhomocysteine hydrolase deficiency in a human: A genetic disorder of methionine metabolism. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 4234-4239.	7.1	201
60	Haplotype distribution and mutations at the PAH locus in Croatia. Human Genetics, 1992, 90, 155-157.	3.8	12