Anibh M Das

List of Publications by Year
in descending order

67 papers	1,642 citations	279798 23 h-index	315739 38 g-index
70 all docs	70 docs citations	70 times ranked	2504 citing authors

#	Article	IF	Citations
1	Treatment of Fabry Disease management with migalastatâ€"outcome from a prospective 24 months observational multicenter study (FAMOUS). European Heart Journal - Cardiovascular Pharmacotherapy, 2022, 8, 272-281.	3.0	33
2	Serum N-glycomics of a novel CDG-IIb patient reveals aberrant IgG glycosylation. Glycobiology, 2022, 32, 380-390.	2.5	5
3	A SUMO4 initiator codon variant in amyotrophic lateral sclerosis reduces SUMO4 expression and alters stress granule dynamics. Journal of Neurology, 2022, 269, 4863-4871.	3.6	3
4	Impact of interventional and nonâ€interventional variables on anthropometric longâ€term development in glutaric aciduria type 1: A national prospective multiâ€centre study. Journal of Inherited Metabolic Disease, 2021, 44, 629-638.	3.6	13
5	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
6	Hydrolytic biotransformation of the bumetanide ester prodrug DIMAEB to bumetanide by esterases in neonatal human and rat serum and neonatal rat brainâ€"A new treatment strategy for neonatal seizures?. Epilepsia, 2021, 62, 269-278.	5.1	5
7	Migalastat Tissue Distribution: Extrapolation From Mice to Humans Using Pharmacokinetic Modeling and Comparison With Agalsidase Beta Tissue Distribution in Mice. Clinical Pharmacology in Drug Development, 2021, 10, 1075-1088.	1.6	4
8	Analysis of Sirtuin 1 and Sirtuin 3 at Enzyme and Protein Levels in Human Breast Milk during the Neonatal Period. Metabolites, 2021, 11, 348.	2.9	2
9	Long-term safety and outcomes in hereditary tyrosinaemia type 1 with nitisinone treatment: a 15-year non-interventional, multicentre study. Lancet Diabetes and Endocrinology,the, 2021, 9, 427-435.	11.4	19
10	Cross-sectional analysis: clinical presentation of children with persistently low ALP levels. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1559-1566.	0.9	5
11	A Case Series on Genotype and Outcome of Liver Transplantation in Children with Niemann-Pick Disease Type C. Children, 2021, 8, 819.	1.5	1
12	The biochemical subtype is a predictor for cognitive function in glutaric aciduria type 1: a national prospective follow-up study. Scientific Reports, 2021, 11, 19300.	3.3	9
13	Hepatorenal Tyrosinaemia: Impact of a Simplified Diet on Metabolic Control and Clinical Outcome. Nutrients, 2021, 13, 134.	4.1	4
14	Impact of Dietary Modifications on Plasma Sirtuins 1, 3 and 5 in Older Overweight Individuals Undergoing 12-Weeks of Circuit Training. Nutrients, 2021, 13, 3824.	4.1	10
15	The Biochemical High Excreter Phenotype Is the Major Risk Factor for Cognitive Impairment in Early Diagnosed Individuals with Glutaric Aciduria Type 1. Neuropediatrics, 2021, 52, .	0.6	0
16	Defining clinical subgroups and genotypeâ€"phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
17	Mitochondrial PCK2 Missense Variant in Shetland Sheepdogs with Paroxysmal Exercise-Induced Dyskinesia (PED). Genes, 2020, 11, 774.	2.4	14
18	Mechanism of Action of Ketogenic Diet Treatment: Impact of Decanoic Acid and Betaâ€"Hydroxybutyrate on Sirtuins and Energy Metabolism in Hippocampal Murine Neurons. Nutrients, 2020, 12, 2379.	4.1	27

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19	Ketogenic Diet: Impact on Cellular Lipids in Hippocampal Murine Neurons. Nutrients, 2020, 12, 3870.	4.1	8
20	Treatment of Fabry's Disease With Migalastat: Outcome From a Prospective Observational Multicenter Study (FAMOUS). Clinical Pharmacology and Therapeutics, 2020, 108, 326-337.	4.7	41
21	Different Trafficking Phenotypes of Niemann-Pick C1 Gene Mutations Correlate with Various Alterations in Lipid Storage, Membrane Composition and Miglustat Amenability. International Journal of Molecular Sciences, 2020, 21, 2101.	4.1	10
22	Impact of Nutrition on Short-Term Exercise-Induced Sirtuin Regulation: Vegans Differ from Omnivores and Lacto-Ovo Vegetarians. Nutrients, 2020, 12, 1004.	4.1	19
23	Influences of Hypoxia Exercise on Whole-Body Insulin Sensitivity and Oxidative Metabolism in Older Individuals. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5238-5248.	3.6	14
24	Different Niemann-Pick C1 Genotypes Generate Protein Phenotypes that Vary in their Intracellular Processing, Trafficking and Localization. Scientific Reports, 2019, 9, 5292.	3.3	31
25	Development and validation of GCâ€“MS methods for the comprehensive analysis of amino acids in plasma and urine and applications to the HELLP syndrome and pediatric kidney transplantation: evidence of altered methylation, transamidination, and arginase activity. Amino Acids, 2019, 51, 529-547.	2.7	44
26	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	5.3	65
27	An overview of combined Dâ€2â€•and Lâ€2â€hydroxyglutaric aciduria: functional analysis of CIC variants. Journal of Inherited Metabolic Disease, 2018, 41, 169-180.	3.6	24
28	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
29	FV 754. Newborn Screening, a Disease-Modifying Intervention for Glutaric Aciduria Type 1. , 2018, 49, .		0
30	Analytical challenges in the assessment of NO synthesis from L-arginine in the MELAS syndrome. International Journal of Cardiology, 2017, 234, 141-142.	1.7	4
31	3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency: Clinical presentation and outcome in a series of 37 patients. Molecular Genetics and Metabolism, 2017, 121, 206-215.	1.1	32
32	Case study on the pathophysiology of Fabry disease: abnormalities of cellular membranes can be reversed by substrate reduction in vitro. Bioscience Reports, 2017, 37, .	2.4	16
33	Rare ADAR and RNASEH2B variants and a type I interferon signature in glioma and prostate carcinoma risk and tumorigenesis. Acta Neuropathologica, 2017, 134, 905-922.	7.7	12
34	Dietary Considerations in Tyrosinemia Type I. Advances in Experimental Medicine and Biology, 2017, 959, 197-204.	1.6	18
35	Diagnosing Hepatorenal Tyrosinaemia in Europe: Newborn Mass Screening Versus Selective Screening. Advances in Experimental Medicine and Biology, 2017, 959, 125-132.	1.6	0
36	Congenital Transcobalamin II Deficiency: A Rare Entity with a Broad Differential. Klinische Padiatrie, 2017, 229, 355-357.	0.6	6

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37	Clinical presentation and outcome in a series of 32 patients with 2-methylacetoacetyl-coenzyme A thiolase (MAT) deficiency. Molecular Genetics and Metabolism, 2017, 122, 67-75.	1.1	24
38	The l-arginine/NO pathway in the MELAS syndrome: An insufficiently explored and controversial research area. International Journal of Cardiology, 2017, 229, 27.	1.7	1
39	HELLP Syndrome. Reproductive Sciences, 2017, 24, 568-574.	2.5	11
40	Clinical utility of nitisinone for the treatment of hereditary tyrosinemia type-1 (HT-1). The Application of Clinical Genetics, 2017, Volume 10, 43-48.	3.0	55
41	Non-invasive test using palmitate in patients with suspected fatty acid oxidation defects: disease-specific acylcarnitine patterns can help to establish the diagnosis. Orphanet Journal of Rare Diseases, 2017, 12, 187.	2.7	4
42	Correction: Congenital Transcobalamin II Deficiency: A Rare Entity with a Broad Differential. Klinische Padiatrie, 2017, 229, e1-e1.	0.6	0
43	Alterations of sirtuins in mitochondrial cytochrome c-oxidase deficiency. PLoS ONE, 2017, 12, e0186517.	2.5	17
44	Lipopolysaccharides and Prematurity: Effect on the Activity of Respiratory Chain Enzymes in Human Umbilical Venous Endothelial Cells. Journal of Pediatric Biochemistry, 2016, 05, 088-093.	0.2	0
45	Unaltered l-arginine/NO pathway in a MELAS patient: Is mitochondrial NO synthase involved in the MELAS syndrome?. International Journal of Cardiology, 2016, 223, 479-481.	1.7	8
46	Neurological Manifestations of Mycoplasma pneumoniae Infection in Hospitalized Children and Their Long-Term Follow-Up. Neuropediatrics, 2016, 47, 308-317.	0.6	21
47	Pharmacotherapy of inborn errors of metabolism illustrating challenges in orphan diseases. Journal of Pharmacological and Toxicological Methods, 2016, 81, 9-14.	0.7	9
48	Gestational diabetes induces alterations of sirtuins in fetal endothelial cells. Pediatric Research, 2016, 79, 788-798.	2.3	22
49	In vivo monitoring of urea cycle activity with 13C-acetate as a tracer of ureagenesis. Molecular Genetics and Metabolism, 2016, 117, 19-26.	1.1	5
50	L-Arginine/NO Pathway Is Altered in Children with Haemolytic-Uraemic Syndrome (HUS). Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-9.	4.0	9
51	Glycogen storage disease type III: modified Atkins diet improves myopathy. Orphanet Journal of Rare Diseases, 2014, 9, 196.	2.7	47
52	Treatment of arginase deficiency revisited: guanidinoacetate as a therapeutic target and biomarker for therapeutic monitoring. Developmental Medicine and Child Neurology, 2014, 56, 1021-1024.	2.1	28
53	Towards newborn screening for ornithine transcarbamylase deficiency: Fast non-chromatographic orotic acid quantification from dried blood spots by tandem mass spectrometry. Clinica Chimica Acta, 2014, 430, 28-32.	1.1	15
54	Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice. Orphanet Journal of Rare Diseases, 2014, 9, 107.	2.7	110

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55	Macro-AST: misleading finding in an adolescent with MCAD-deficiency. BMC Gastroenterology, 2012, 12, 119.	2.0	2
56	Effect of tacrolimus on energy metabolism in human umbilical endothelial cells. Annals of Transplantation, 2011, 16, 68-75.	0.9	17
57	Plasma and urine amino acid pattern in preterm infants on enteral nutrition: impact of gestational age. Amino Acids, 2010, 38, 959-972.	2.7	13
58	Inborn Errors of Energy Metabolism Associated with Myopathies. Journal of Biomedicine and Biotechnology, 2010, 2010, 1-19.	3.0	48
59	Glycogen Storage Disease Type 1: Impact of Medium-Chain Triglycerides on Metabolic Control and Growth. Annals of Nutrition and Metabolism, 2010, 56, 225-232.	1.9	27
60	One Liver for Four Children: First Clinical Series of Liver Cell Transplantation for Severe Neonatal Urea Cycle Defects. Transplantation, 2009, 87, 636-641.	1.0	142
61	Carnitineâ€palmitoyltransferase 2 deficiency: Novel mutations and relevance of newborn screening. American Journal of Medical Genetics, Part A, 2008, 146A, 2925-2928.	1.2	15
62	Relationship of Mitochondrial DNA Depletion and Respiratory Chain Activity in Preadipocytes treated with Nucleoside Reverse Transcriptase Inhibitors. Antiviral Therapy, 2007, 12, 205-216.	1.0	27
63	Isolated Mitochondrial Long-Chain Ketoacyl-CoA Thiolase Deficiency Resulting from Mutations in the HADHB Gene. Clinical Chemistry, 2006, 52, 530-534.	3.2	60
64	Neonatal screening for defects of the mitochondrial trifunctional protein. Molecular Genetics and Metabolism, 2005, 85, 108-114.	1.1	63
65	Fabry disease: reduced activities of respiratory chain enzymes with decreased levels of energy-rich phosphates in fibroblasts. Molecular Genetics and Metabolism, 2004, 82, 93-97.	1.1	90
66	Regulation of the mitochondrial ATP-synthase in health and disease. Molecular Genetics and Metabolism, 2003, 79, 71-82.	1.1	97
67	Altered levels of high-energy phosphate compoundsin fibroblasts from different forms of neuronal Paediatric Neurology, 2001, 5, 143-146.	1.6	17