

Johannes A Mayr

List of Publications by Year in descending order

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157
papers

7,903
citations

34076

52
h-index

64755

79
g-index

168
all docs

168
docs citations

168
times ranked

10632
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. <i>Nature Communications</i> , 2017, 8, 15824.	5.8	432
2	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 314-320.	2.6	192
3	Lipoic acid biosynthesis defects. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 553-563.	1.7	191
4	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalomyopathy. <i>Nature Genetics</i> , 2008, 40, 1288-1290.	9.4	183
5	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. <i>Journal of Medical Genetics</i> , 2012, 49, 277-283.	1.5	182
6	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. <i>Nature Communications</i> , 2016, 7, 12039.	5.8	178
7	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. <i>Brain</i> , 2016, 139, 31-38.	3.7	174
8	Loss of Complex I due to Mitochondrial DNA Mutations in Renal Oncocytoma. <i>Clinical Cancer Research</i> , 2008, 14, 2270-2275.	3.2	154
9	Decrease of mitochondrial DNA content and energy metabolism in renal cell carcinoma. <i>Carcinogenesis</i> , 2004, 25, 1005-1010.	1.3	144
10	Mitochondrial Phosphate Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. <i>American Journal of Human Genetics</i> , 2007, 80, 478-484.	2.6	142
11	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 482-495.	2.6	138
12	Mitochondrial ATP synthase deficiency due to a mutation in the ATP5E gene for the F1 subunit. <i>Human Molecular Genetics</i> , 2010, 19, 3430-3439.	1.4	133
13	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 211-223.	2.6	127
14	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. <i>American Journal of Human Genetics</i> , 2014, 95, 708-720.	2.6	123
15	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. <i>American Journal of Human Genetics</i> , 2016, 98, 1130-1145.	2.6	118
16	Serine Catabolism Feeds NADH when Respiration Is Impaired. <i>Cell Metabolism</i> , 2020, 31, 809-821.e6.	7.2	118
17	Thiamine Pyrophosphokinase Deficiency in Encephalopathic Children with Defects in the Pyruvate Oxidation Pathway. <i>American Journal of Human Genetics</i> , 2011, 89, 806-812.	2.6	107
18	CAD mutations and uridine-responsive epileptic encephalopathy. <i>Brain</i> , 2017, 140, 279-286.	3.7	106

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19	Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. <i>Journal of Medical Genetics</i> , 2016, 53, 270-278.	1.5	105
20	Lipoic Acid Synthetase Deficiency Causes Neonatal-Onset Epilepsy, Defective Mitochondrial Energy Metabolism, and Glycine Elevation. <i>American Journal of Human Genetics</i> , 2011, 89, 792-797.	2.6	104
21	Spectrum of combined respiratory chain defects. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 629-640.	1.7	102
22	Mitochondrial DNA haplogroup T is associated with coronary artery disease and diabetic retinopathy: a case control study. <i>BMC Medical Genetics</i> , 2009, 10, 35.	2.1	93
23	Deficiency of <sc>ECHS</sc>1 causes mitochondrial encephalopathy with cardiac involvement. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 492-509.	1.7	90
24	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. <i>Blood</i> , 2020, 136, 1033-1043.	0.6	90
25	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	89
26	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. <i>American Journal of Human Genetics</i> , 2015, 96, 309-317.	2.6	86
27	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	3.6	85
28	Homozygous missense mutation in <i>BOLA3</i> causes multiple mitochondrial dysfunctions syndrome in two siblings. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 55-62.	1.7	83
29	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. <i>Frontiers in Genetics</i> , 2015, 06, 123.	1.1	81
30	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes including <i>NDUFB9</i>. <i>Journal of Medical Genetics</i> , 2012, 49, 83-89.	1.5	78
31	Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 119.	1.2	77
32	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. <i>American Journal of Human Genetics</i> , 2016, 99, 894-902.	2.6	75
33	Identification of a novel, Ca ²⁺ -dependent phospholipase D with preference for phosphatidylserine and phosphatidylethanolamine in <i>Saccharomyces cerevisiae</i> . <i>FEBS Letters</i> , 1996, 393, 236-240.	1.3	73
34	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 414-422.	2.6	73
35	Delineating <i>MT-ATP6</i>-associated disease. <i>Neurology: Genetics</i> , 2020, 6, e393.	0.9	73
36	Severe depletion of mitochondrial DNA in spinal muscular atrophy. <i>Acta Neuropathologica</i> , 2003, 105, 245-251.	3.9	72

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37	Low aerobic mitochondrial energy metabolism in poorly- or undifferentiated neuroblastoma. <i>BMC Cancer</i> , 2010, 10, 149.	1.1	72
38	Mitochondrial encephalocardio-myopathy with early neonatal onset due to TMEM70 mutation. <i>Archives of Disease in Childhood</i> , 2010, 95, 296-301.	1.0	72
39	Impaired riboflavin transport due to missense mutations in <i>SLC52A2</i> causes Brown-Vialetto-Van Laere syndrome. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 943-948.	1.7	72
40	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
41	The Mitochondrial T16189C Polymorphism Is Associated with Coronary Artery Disease in Middle European Populations. <i>PLoS ONE</i> , 2011, 6, e16455.	1.1	70
42	Deficiency of the mitochondrial phosphate carrier presenting as myopathy and cardiomyopathy in a family with three affected children. <i>Neuromuscular Disorders</i> , 2011, 21, 803-808.	0.3	65
43	Neonatal onset of mitochondrial disorders in 129 patients: clinical and laboratory characteristics and a new approach to diagnosis. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 749-759.	1.7	65
44	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 342-352.	0.5	65
45	Progressive deafness-dystonia due to <i>SERAC1</i> mutations: A study of 67 cases. <i>Annals of Neurology</i> , 2017, 82, 1004-1015.	2.8	63
46	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. <i>American Journal of Human Genetics</i> , 2018, 102, 685-695.	2.6	61
47	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	1.2	61
48	Mutation or knock-down of 17 β -hydroxysteroid dehydrogenase type 10 cause loss of MRPP1 and impaired processing of mitochondrial heavy strand transcripts. <i>Human Molecular Genetics</i> , 2014, 23, 3618-3628.	1.4	60
49	Lipid metabolism in mitochondrial membranes. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 137-144.	1.7	60
50	A Guideline for the Diagnosis of Pediatric Mitochondrial Disease: The Value of Muscle and Skin Biopsies in the Genetics Era. <i>Neuropediatrics</i> , 2017, 48, 309-314.	0.3	60
51	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
52	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2017, 101, 525-538.	2.6	58
53	<i>GAL3</i> receptor KO mice exhibit an anxiety-like phenotype. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 7138-7143.	3.3	57
54	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57

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55	Treatable mitochondrial diseases: cofactor metabolism and beyond. <i>Brain</i> , 2017, 140, e11-e11.	3.7	57
56	Mitochondrial DNA mutation analysis from exome sequencingâ€”A more holistic approach in diagnostics of suspected mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 909-917.	1.7	57
57	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 101, 283-290.	2.6	55
58	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. <i>Nature Communications</i> , 2021, 12, 1929.	5.8	55
59	BOLA (BoLA Family Member 3) Deficiency Controls Endothelial Metabolism and Glycine Homeostasis in Pulmonary Hypertension. <i>Circulation</i> , 2019, 139, 2238-2255.	1.6	54
60	Mitochondrial Haplogroups and Control Region Polymorphisms in Age-Related Macular Degeneration: A Case-Control Study. <i>PLoS ONE</i> , 2012, 7, e30874.	1.1	54
61	The problem of interlab variation in methods for mitochondrial disease diagnosis: enzymatic measurement of respiratory chain complexesâ†. <i>Mitochondrion</i> , 2004, 4, 427-439.	1.6	52
62	The mitochondrial phosphate carrier: Role in oxidative metabolism, calcium handling and mitochondrial disease. <i>Biochemical and Biophysical Research Communications</i> , 2015, 464, 369-375.	1.0	52
63	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. <i>Mitochondrion</i> , 2015, 21, 12-18.	1.6	51
64	A novel mutation of the RRM2B gene in an infant with early fatal encephalomyopathy, central hypomyelination, and tubulopathy. <i>Molecular Genetics and Metabolism</i> , 2009, 98, 300-304.	0.5	50
65	Functional Differences between Mitochondrial Haplogroup T and Haplogroup H in HEK293 Cybrid Cells. <i>PLoS ONE</i> , 2012, 7, e52367.	1.1	50
66	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: A treatable neurological disorder caused by TPK1 mutations. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 301-306.	0.5	50
67	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. <i>American Journal of Human Genetics</i> , 2016, 99, 674-682.	2.6	48
68	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. <i>Genetics in Medicine</i> , 2020, 22, 610-621.	1.1	46
69	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to <i>FBXL4</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 905-914.	1.7	45
70	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 391-403.	1.7	44
71	Rapid screening of the entire mitochondrial DNA for low-level heteroplasmic mutations. <i>Mitochondrion</i> , 2005, 5, 282-296.	1.6	43
72	Mitochondrial Haplogroups, Control Region Polymorphisms and Malignant Melanoma: A Study in Middle European Caucasians. <i>PLoS ONE</i> , 2011, 6, e27192.	1.1	43

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73	<i>WDR73</i> Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. <i>Human Mutation</i> , 2015, 36, 1021-1028.	1.1	42
74	Choline-related inherited metabolic diseases – A mini review. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 237-242.	1.7	42
75	Molecular and clinical spectra of FBXL4 deficiency. <i>Human Mutation</i> , 2017, 38, 1649-1659.	1.1	41
76	Bi-allelic Mutations in <i>NDUFA6</i> Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 592-601.	2.6	41
77	<i>NDUFB8</i> Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 460-467.	2.6	40
78	Bi-allelic <i>ADPRHL2</i> Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. <i>American Journal of Human Genetics</i> , 2018, 103, 817-825.	2.6	40
79	Bi-Allelic <i>UQCERS1</i> Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. <i>American Journal of Human Genetics</i> , 2020, 106, 102-111.	2.6	36
80	Reduced Respiratory Control with ADP and Changed Pattern of Respiratory Chain Enzymes as a Result of Selective Deficiency of the Mitochondrial ATP Synthase. <i>Pediatric Research</i> , 2004, 55, 988-994.	1.1	34
81	Respiratory chain complex I is a mitochondrial tumor suppressor of oncocytic tumors. <i>Frontiers in Bioscience - Elite</i> , 2011, E3, 315-325.	0.9	34
82	Combined Respiratory Chain Deficiency and <i>UQCERS2</i> Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. <i>Oxidative Medicine and Cellular Longevity</i> , 2017, 2017, 1-11.	1.9	33
83	Mutations in <i>NDUFS1</i> Cause Metabolic Reprogramming and Disruption of the Electron Transfer. <i>Cells</i> , 2019, 8, 1149.	1.8	30
84	Bi-allelic <i>HPDL</i> Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2020, 107, 364-373.	2.6	30
85	Protein Sets Define Disease States and Predict In Vivo Effects of Drug Treatment. <i>Molecular and Cellular Proteomics</i> , 2013, 12, 1965-1979.	2.5	29
86	<i>HIBCH</i> deficiency in a patient with phenotypic characteristics of mitochondrial disorders. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3162-3169.	0.7	27
87	Mitochondrial Haplogroups and Control Region Polymorphisms Are Not Associated with Prostate Cancer in Middle European Caucasians. <i>PLoS ONE</i> , 2009, 4, e6370.	1.1	25
88	Mutations in <i>TTC19</i> : expanding the molecular, clinical and biochemical phenotype. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 40.	1.2	25
89	Biallelic variants in the transcription factor <i>PAX7</i> are a new genetic cause of myopathy. <i>Genetics in Medicine</i> , 2019, 21, 2521-2531.	1.1	25
90	Biallelic variants in <i>WARS2</i> encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. <i>Human Mutation</i> , 2017, 38, 1786-1795.	1.1	24

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91	The genotypic and phenotypic spectrum of MTO1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 28-42.	0.5	24
92	Cellular rescue-assay aids verification of causative DNA-variants in mitochondrial complex I deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 161-166.	0.5	23
93	Mitochondrial DNA mutation "m.3243A>G" Heterogeneous clinical picture for cardiologists (m.3243A>G: A phenotypic chameleon). <i>Congenital Heart Disease</i> , 2018, 13, 671-677.	0.0	22
94	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. <i>Genetics in Medicine</i> , 2021, 23, 1705-1714.	1.1	22
95	Multiplex primer extension analysis for rapid detection of major European mitochondrial haplogroups. <i>Electrophoresis</i> , 2006, 27, 3864-3868.	1.3	21
96	Melanoma tumors exhibit a variable but distinct metabolic signature. <i>Experimental Dermatology</i> , 2018, 27, 204-207.	1.4	21
97	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsy"Report of 11 Patients. <i>Neuropediatrics</i> , 2018, 49, 373-378.	0.3	21
98	Heterozygous mutation in the X chromosomal NDUFA1 gene in a girl with complex I deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 358-361.	0.5	20
99	Loss of mitochondria in ganglioneuromas. <i>Frontiers in Bioscience - Elite</i> , 2011, E3, 179-186.	0.9	20
100	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. <i>Mitochondrion</i> , 2017, 37, 55-61.	1.6	20
101	Oxidative Phosphorylation System in Gastric Carcinomas and Gastritis. <i>Oxidative Medicine and Cellular Longevity</i> , 2017, 2017, 1-14.	1.9	20
102	How to proceed after "negative" exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 663-681.	1.7	20
103	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. <i>Genetics in Medicine</i> , 2020, 22, 1589-1597.	1.1	19
104	Facial asymmetry correction with moulded helmet therapy in infants with deformational skull base plagiocephaly. <i>Journal of Cranio-Maxillo-Facial Surgery</i> , 2018, 46, 28-34.	0.7	17
105	Reduced Levels of ATP Synthase Subunit ATP5F1A Correlate with Earlier-Onset Prostate Cancer. <i>Oxidative Medicine and Cellular Longevity</i> , 2018, 2018, 1-10.	1.9	17
106	Changes in the expression of oxidative phosphorylation complexes in the aging intestinal mucosa. <i>Experimental Gerontology</i> , 2020, 135, 110924.	1.2	17
107	Heterogeneity of mitochondrial energy metabolism in classical triphasic Wilms tumor. <i>Frontiers in Bioscience - Elite</i> , 2011, E3, 187-193.	0.9	16
108	Bi-allelic Variants in TKFC Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease. <i>American Journal of Human Genetics</i> , 2020, 106, 256-263.	2.6	16

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109	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. <i>Genetics in Medicine</i> , 2021, 23, 341-351.	1.1	16
110	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. <i>Genome Medicine</i> , 2021, 13, 55.	3.6	16
111	Infantile peripheral neuropathy, deafness, and proximal tubulopathy associated with a novel mutation of the RRM2B gene. <i>Croatian Medical Journal</i> , 2013, 54, 579-584.	0.2	15
112	A novel sporadic mutation G14739A of the mitochondrial tRNA ^{Glu} in a girl with exercise intolerance. <i>Neuromuscular Disorders</i> , 2006, 16, 874-877.	0.3	13
113	MELAS Syndrome and Kidney Disease Without Fanconi Syndrome or Proteinuria: A Case Report. <i>American Journal of Kidney Diseases</i> , 2016, 68, 949-953.	2.1	13
114	Alterations of oxidative phosphorylation in meningiomas and peripheral nerve sheath tumors. <i>Neuro-Oncology</i> , 2016, 18, 184-194.	0.6	13
115	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. <i>Journal of Clinical Medicine</i> , 2019, 8, 991.	1.0	13
116	Severe Deoxyguanosine Kinase Deficiency in Austria. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2019, 68, e1-e6.	0.9	13
117	Severe syndromic ID and skewed X-inactivation in a girl with NAA10 dysfunction and a novel heterozygous de novo NAA10 p.(His16Pro) variant - a case report. <i>BMC Medical Genetics</i> , 2020, 21, 153.	2.1	13
118	De novo variants in <i>TCF7L2</i> are associated with a syndromic neurodevelopmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2384-2390.	0.7	13
119	A spoonful of "L-fucose" an efficient therapy for GFUS/CDG, a new glycosylation disorder. <i>EMBO Molecular Medicine</i> , 2021, 13, e14332.	3.3	13
120	Alterations of respiratory chain complexes in sporadic pheochromocytoma. <i>Frontiers in Bioscience - Elite</i> , 2011, E3, 194-200.	0.9	12
121	Mitochondrial myopathy associated with a novel 5522G>A mutation in the mitochondrial tRNA ^{Trp} gene. <i>European Journal of Human Genetics</i> , 2013, 21, 871-875.	1.4	12
122	Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches. <i>European Journal of Medical Genetics</i> , 2020, 63, 104046.	0.7	12
123	Variants in Mitochondrial <i>ATP</i> Synthase Cause Variable Neurologic Phenotypes. <i>Annals of Neurology</i> , 2022, 91, 225-237.	2.8	12
124	Mitochondrial complex deficiency by novel compound heterozygous <i>TMEM70</i> variants and correlation with developmental delay, undescended testicle, and left ventricular noncompaction in a Japanese patient: A case report. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 553-557.	0.2	11
125	Age-Related Deterioration of Mitochondrial Function in the Intestine. <i>Oxidative Medicine and Cellular Longevity</i> , 2020, 2020, 1-12.	1.9	11
126	Biallelic <i>BUB1</i> mutations cause microcephaly, developmental delay, and variable effects on cohesion and chromosome segregation. <i>Science Advances</i> , 2022, 8, eabk0114.	4.7	11

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127	Reduction of nuclear encoded enzymes of mitochondrial energy metabolism in cells devoid of mitochondrial DNA. <i>Biochemical and Biophysical Research Communications</i> , 2012, 417, 1052-1057.	1.0	10
128	Sternal Fracture in Children: Diagnosis by Ultrasonography. <i>European Journal of Pediatric Surgery Reports</i> , 2017, 05, e39-e42.	0.1	10
129	Fractures and Dislocations of the Foot in Children. <i>Clinics in Podiatric Medicine and Surgery</i> , 2006, 23, 167-189.	0.2	9
130	Effects of alpha-melanocyte-stimulating hormone on mitochondrial energy metabolism in rats of different age-groups. <i>Neuropeptides</i> , 2017, 64, 123-130.	0.9	9
131	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. <i>Genetics in Medicine</i> , 2022, 24, 179-191.	1.1	9
132	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. <i>JIMD Reports</i> , 2015, 29, 89-93.	0.7	8
133	Deficiency of respiratory chain complex I in Hashimoto thyroiditis. <i>Mitochondrion</i> , 2016, 26, 1-6.	1.6	8
134	Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. <i>Methods in Molecular Biology</i> , 2017, 1567, 379-390.	0.4	8
135	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , 2021, 23, 2415-2425.	1.1	8
136	Congenital disorders of glycosylation with defective fucosylation. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1441-1452.	1.7	8
137	Disorders of Oxidative Phosphorylation. , 2016, , 223-242.		6
138	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. Seizure: the Journal of the British Epilepsy Association, 2021, 87, 25-29.	0.9	6
139	Expression of Oxidative Phosphorylation Complexes and Mitochondrial Mass in Pediatric and Adult Inflammatory Bowel Disease. <i>Oxidative Medicine and Cellular Longevity</i> , 2022, 2022, 1-14.	1.9	6
140	Damage control orthopedics applied in an 8-year-old child with life-threatening multiple injuries. <i>Medicine (United States)</i> , 2019, 98, e15294.	0.4	5
141	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. <i>Molecular Genetics and Metabolism</i> , 2021, 133, 362-371.	0.5	5
142	Characterising a homozygous two-exon deletion in <i>UQCRC1</i> : comparing human and mouse phenotypes. <i>EMBO Molecular Medicine</i> , 2021, 13, e14397.	3.3	5
143	Alterations of Oxidative Phosphorylation Complexes in Papillary Thyroid Carcinoma. <i>Cells</i> , 2018, 7, 40.	1.8	4
144	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. <i>Mammalian Genome</i> , 2021, 32, 332-349.	1.0	4

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145	Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK1 Variants. <i>Neuropediatrics</i> , 2021, 52, 123-125.	0.3	4
146	A 1.1 Million Base Pair X-Chromosomal Deletion Covering the PDHA1 and CDKL5 Genes in a Female Patient with West Syndrome and Pyruvate Oxidation Deficiency. <i>Neuropediatrics</i> , 2012, 43, 130-134.	0.3	3
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149	A novel cryptic splice site mutation in COL1A2 as a cause of osteogenesis imperfecta. <i>Bone Reports</i> , 2021, 15, 101110.	0.2	3
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151	Three Novel EPCAM Variants Causing Tufting Enteropathy in Three Families. <i>Children</i> , 2021, 8, 503.	0.6	2
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