Johannes A Mayr

List of Publications by Year
in descending order

157	7,903	34105 52	64796 79
papers	citations	h-index	g-index

168	168	168	10632
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
2	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
3	Lipoic acid biosynthesis defects. Journal of Inherited Metabolic Disease, 2014, 37, 553-563.	3.6	191
4	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	21.4	183
5	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
6	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	12.8	178
7	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain, 2016, 139, 31-38.	7.6	174
8	Loss of Complex I due to Mitochondrial DNA Mutations in Renal Oncocytoma. Clinical Cancer Research, 2008, 14, 2270-2275.	7.0	154
9	Decrease of mitochondrial DNA content and energy metabolism in renal cell carcinoma. Carcinogenesis, 2004, 25, 1005-1010.	2.8	144
10	Mitochondrial Phosphateâ€“Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. American Journal of Human Genetics, 2007, 80, 478-484.	6.2	142
11	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
12	Mitochondrial ATP synthase deficiency due to a mutation in the ATP5E gene for the F1 Â subunit. Human Molecular Genetics, 2010, 19, 3430-3439.	2.9	133
13	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	6.2	127
14	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	6.2	123
15	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
16	Serine Catabolism Feeds NADH when Respiration Is Impaired. Cell Metabolism, 2020, 31, 809-821.e6.	16.2	118
17	Thiamine Pyrophosphokinase Deficiency in Encephalopathic Children with Defects in the Pyruvate Oxidation Pathway. American Journal of Human Genetics, 2011, 89, 806-812.	6.2	107
18	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	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. Journal of Medical Genetics, 2016, 53, 270-278.	3.2	105
20	Lipoic Acid Synthetase Deficiency Causes Neonatal-Onset Epilepsy, Defective Mitochondrial Energy Metabolism, and Glycine Elevation. American Journal of Human Genetics, 2011, 89, 792-797.	6.2	104
21	Spectrum of combined respiratory chain defects. Journal of Inherited Metabolic Disease, 2015, 38, 629-640.	3.6	102
22	Mitochondrial DNA haplogroup T is associated with coronary artery disease and diabetic retinopathy: a case control study. BMC Medical Genetics, 2009, 10, 35.	2.1	93
23	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
24	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043.	1.4	90
25	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
26	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	6.2	86
27	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
28	Homozygous missense mutation in BOLA3 causes multiple mitochondrial dysfunctions syndrome in two siblings. Journal of Inherited Metabolic Disease, 2013, 36, 55-62.	3.6	83
29	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	2.3	81
30	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
31	Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. Orphanet Journal of Rare Diseases, 2014, 9, 119.	2.7	77
32	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
33	Identification of a novel, Ca2+-dependent phospholipase D with preference for phosphatidylserine and phosphatidylethanolamine inSaccharomyces cerevisiae. FEBS Letters, 1996, 393, 236-240.	2.8	73
34	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	6.2	73
35	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
36	Severe depletion of mitochondrial DNA in spinal muscular atrophy. Acta Neuropathologica, 2003, 105, 245-251.	7.7	72

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37	Low aerobic mitochondrial energy metabolism in poorly- or undifferentiated neuroblastoma. BMC Cancer, 2010, 10, 149.	2.6	72
38	Mitochondrial encephalocardio-myopathy with early neonatal onset due to TMEM70 mutation. Archives of Disease in Childhood, 2010, 95, 296-301.	1.9	72
39	Impaired riboflavin transport due to missense mutations in SLC52A2 causes Brownâ€Vialettoâ€Van Laere syndrome. Journal of Inherited Metabolic Disease, 2012, 35, 943-948.	3.6	72
40	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
41	The Mitochondrial T16189C Polymorphism Is Associated with Coronary Artery Disease in Middle European Populations. PLoS ONE, 2011, 6, e16455.	2.5	70
42	Deficiency of the mitochondrial phosphate carrier presenting as myopathy and cardiomyopathy in a family with three affected children. Neuromuscular Disorders, 2011, 21, 803-808.	0.6	65
43	Neonatal onset of mitochondrial disorders in 129 patients: clinical and laboratory characteristics and a new approach to diagnosis. Journal of Inherited Metabolic Disease, 2012, 35, 749-759.	3.6	65
44	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
45	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
46	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	6.2	61
47	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
48	Mutation or knock-down of 17Î²-hydroxysteroid dehydrogenase type 10 cause loss of MRPP1 and impaired processing of mitochondrial heavy strand transcripts. Human Molecular Genetics, 2014, 23, 3618-3628.	2.9	60
49	Lipid metabolism in mitochondrial membranes. Journal of Inherited Metabolic Disease, 2015, 38, 137-144.	3.6	60
50	A Guideline for the Diagnosis of Pediatric Mitochondrial Disease: The Value of Muscle and Skin Biopsies in the Genetics Era. Neuropediatrics, 2017, 48, 309-314.	0.6	60
51	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
52	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	6.2	58
53	GAL ₃ receptor KO mice exhibit an anxiety-like phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 7138-7143.	7.1	57
54	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	6.2	57

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55	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	7.6	57
56	Mitochondrial DNA mutation analysis from exome sequencingâ€"A more holistic approach in diagnostics of suspected mitochondrial disease. Journal of Inherited Metabolic Disease, 2019, 42, 909-917.	3.6	57
57	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	6.2	55
58	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. Nature Communications, 2021, 12, 1929.	12.8	55
59	BOLA (BolA Family Member 3) Deficiency Controls Endothelial Metabolism and Glycine Homeostasis in Pulmonary Hypertension. Circulation, 2019, 139, 2238-2255.	1.6	54
60	Mitochondrial Haplogroups and Control Region Polymorphisms in Age-Related Macular Degeneration: A Case-Control Study. PLoS ONE, 2012, 7, e30874.	2.5	54
61	The problem of interlab variation in methods for mitochondrial disease diagnosis: enzymatic measurement of respiratory chain complexesâ˜†. Mitochondrion, 2004, 4, 427-439.	3.4	52
62	The mitochondrial phosphate carrier: Role in oxidative metabolism, calcium handling and mitochondrial disease. Biochemical and Biophysical Research Communications, 2015, 464, 369-375.	2.1	52
63	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion, 2015, 21, 12-18.	3.4	51
64	A novel mutation of the RRM2B gene in an infant with early fatal encephalomyopathy, central hypomyelination, and tubulopathy. Molecular Genetics and Metabolism, 2009, 98, 300-304.	1.1	50
65	Functional Differences between Mitochondrial Haplogroup T and Haplogroup H in HEK293 Cybrid Cells. PLoS ONE, 2012, 7, e52367.	2.5	50
66	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: A treatable neurological disorder caused by TPK1 mutations. Molecular Genetics and Metabolism, 2014, 113, 301-306.	1.1	50
67	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
68	Defining clinical subgroups and genotypeâ€"phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
69	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
70	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	3.6	44
71	Rapid screening of the entire mitochondrial DNA for low-level heteroplasmic mutations. Mitochondrion, 2005, 5, 282-296.	3.4	43
72	Mitochondrial Haplogroups, Control Region Polymorphisms and Malignant Melanoma: A Study in Middle European Caucasians. PLoS ONE, 2011, 6, e27192.	2.5	43

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73	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
74	Cholineâ€relatedâ€inherited metabolic diseasesâ€"A mini review. Journal of Inherited Metabolic Disease, 2019, 42, 237-242.	3.6	42
75	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
76	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	6.2	41
77	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	6.2	40
78	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
79	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	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. Pediatric Research, 2004, 55, 988-994.	2.3	34
81	Respiratory chain complex I is a mitochondrial tumor suppressor of oncocytic tumors. Frontiers in Bioscience - Elite, 2011, E3, 315-325.	1.8	34
82	Combined Respiratory Chain Deficiency and UQCC2 Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-11.	4.0	33
83	Mutations in NDUFS1 Cause Metabolic Reprogramming and Disruption of the Electron Transfer. Cells, 2019, 8, 1149.	4.1	30
84	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30
85	Protein Sets Define Disease States and Predict In Vivo Effects of Drug Treatment. Molecular and Cellular Proteomics, 2013, 12, 1965-1979.	3.8	29
86	HIBCH deficiency in a patient with phenotypic characteristics of mitochondrial disorders. American Journal of Medical Genetics, Part A, 2014, 164, 3162-3169.	1.2	27
87	Mitochondrial Haplogroups and Control Region Polymorphisms Are Not Associated with Prostate Cancer in Middle European Caucasians. PLoS ONE, 2009, 4, e6370.	2.5	25
88	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	2.7	25
89	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
90	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24

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91	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
92	Cellular rescue-assay aids verification of causative DNA-variants in mitochondrial complex I deficiency. Molecular Genetics and Metabolism, 2011, 103, 161-166.	1.1	23
93	Mitochondrial DNA mutation â€œm.3243A>Gâ€â€"Heterogeneous clinical picture for cardiologists (â€œm.3243A>Gâ€• A phenotypic chameleon). Congenital Heart Disease, 2018, 13, 671-677.	0.2	22
94	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
95	Multiplex primer extension analysis for rapid detection of major European mitochondrial haplogroups. Electrophoresis, 2006, 27, 3864-3868.	2.4	21
96	Melanoma tumors exhibit a variable but distinct metabolic signature. Experimental Dermatology, 2018, 27, 204-207.	2.9	21
97	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsyâ€"Report of 11 Patients. Neuropediatrics, 2018, 49, 373-378.	0.6	21
98	Heterozygous mutation in the X chromosomal NDUFA1 gene in a girl with complex I deficiency. Molecular Genetics and Metabolism, 2011, 103, 358-361.	1.1	20
99	Loss of mitochondria in ganglioneuromas. Frontiers in Bioscience - Elite, 2011, E3, 179-186.	1.8	20
100	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion, 2017, 37, 55-61.	3.4	20
101	Oxidative Phosphorylation System in Gastric Carcinomas and Gastritis. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-14.	4.0	20
102	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	3.6	20
103	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. Genetics in Medicine, 2020, 22, 1589-1597.	2.4	19
104	Facial asymmetry correction with moulded helmet therapy in infants with deformational skull base plagiocephaly. Journal of Cranio-Maxillo-Facial Surgery, 2018, 46, 28-34.	1.7	17
105	Reduced Levels of ATP Synthase Subunit ATP5F1A Correlate with Earlier-Onset Prostate Cancer. Oxidative Medicine and Cellular Longevity, 2018, 2018, 1-10.	4.0	17
106	Changes in the expression of oxidative phosphorylation complexes in the aging intestinal mucosa. Experimental Gerontology, 2020, 135, 110924.	2.8	17
107	Heterogeneity of mitochondrial energy metabolism in classical triphasic Wilms tumor. Frontiers in Bioscience - Elite, 2011, E3, 187-193.	1.8	16
108	Bi-allelic Variants in TKFC Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease. American Journal of Human Genetics, 2020, 106, 256-263.	6.2	16

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109	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	2.4	16
110	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	8.2	16
111	Infantile peripheral neuropathy, deafness, and proximal tubulopathy associated with a novel mutation of the RRM2B gene. Croatian Medical Journal, 2013, 54, 579-584.	0.7	15
112	A novel sporadic mutation G14739A of the mitochondrial tRNAGlu in a girl with exercise intolerance. Neuromuscular Disorders, 2006, 16, 874-877.	0.6	13
113	MELAS Syndrome and Kidney Disease Without Fanconi Syndrome or Proteinuria: A Case Report. American Journal of Kidney Diseases, 2016, 68, 949-953.	1.9	13
114	Alterations of oxidative phosphorylation in meningiomas and peripheral nerve sheath tumors. Neuro-Oncology, 2016, 18, 184-194.	1.2	13
115	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	2.4	13
116	Severe Deoxyguanosine Kinase Deficiency in Austria. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e1-e6.	1.8	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. BMC Medical Genetics, 2020, 21, 153.	2.1	13
118	De novo variants in <scp>TCF7L2</scp> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	1.2	13
119	A spoonful of Lâ€fucoseâ€"an efficient therapy for GFUSâ€CDG, a new glycosylation disorder. EMBO Molecular Medicine, 2021, 13, e14332.	6.9	13
120	Alterations of respiratory chain complexes in sporadic pheochromocytoma. Frontiers in Bioscience - Elite, 2011, E3, 194-200.	1.8	12
121	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
122	Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches. European Journal of Medical Genetics, 2020, 63, 104046.	1.3	12
123	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
124	Mitochondrial complex deficiency by novel compound heterozygous <scp>TMEM</scp>70 variants and correlation with developmental delay, undescended testicle, and left ventricular noncompaction in a Japanese patient: A case report. Clinical Case Reports (discontinued), 2019, 7, 553-557.	0.5	11
125	Age-Related Deterioration of Mitochondrial Function in the Intestine. Oxidative Medicine and Cellular Longevity, 2020, 2020, 1-12.	4.0	11
126	Biallelic BUB1 mutations cause microcephaly, developmental delay, and variable effects on cohesion and chromosome segregation. Science Advances, 2022, 8, eabk0114.	10.3	11

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127	Reduction of nuclear encoded enzymes of mitochondrial energy metabolism in cells devoid of mitochondrial DNA. Biochemical and Biophysical Research Communications, 2012, 417, 1052-1057.	2.1	10
128	Sternal Fracture in Children: Diagnosis by Ultrasonography. European Journal of Pediatric Surgery Reports, 2017, 05, e39-e42.	0.5	10
129	Fractures and Dislocations of the Foot in Children. Clinics in Podiatric Medicine and Surgery, 2006, 23, 167-189.	0.6	9
130	Effects of alpha-melanocyte-stimulating hormone on mitochondrial energy metabolism in rats of different age-groups. Neuropeptides, 2017, 64, 123-130.	2.2	9
131	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
132	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	1.5	8
133	Deficiency of respiratory chain complex I in Hashimoto thyroiditis. Mitochondrion, 2016, 26, 1-6.	3.4	8
134	Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. Methods in Molecular Biology, 2017, 1567, 379-390.	0.9	8
135	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	2.4	8
136	Congenital disorders of glycosylation with defective fucosylation. Journal of Inherited Metabolic Disease, 2021, 44, 1441-1452.	3.6	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.	2.0	6
139	Expression of Oxidative Phosphorylation Complexes and Mitochondrial Mass in Pediatric and Adult Inflammatory Bowel Disease. Oxidative Medicine and Cellular Longevity, 2022, 2022, 1-14.	4.0	6
140	Damage control orthopedics applied in an 8-year-old child with life-threatening multiple injuries. Medicine (United States), 2019, 98, e15294.	1.0	5
141	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. Molecular Genetics and Metabolism, 2021, 133, 362-371.	1.1	5
142	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	6.9	5
143	Alterations of Oxidative Phosphorylation Complexes in Papillary Thyroid Carcinoma. Cells, 2018, 7, 40.	4.1	4
144	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	2.2	4

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145	Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK1 Variants. Neuropediatrics, 2021, 52, 123-125.	0.6	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. Neuropediatrics, 2012, 43, 130-134.	0.6	3
147	Hip reconstruction in closed triradiate cartilage: long-term outcomes in patients with cerebral palsy. Archives of Orthopaedic and Trauma Surgery, 2021, , 1.	2.4	3
148	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	5.3	3
149	A novel cryptic splice site mutation in COL1A2 as a cause of osteogenesis imperfecta. Bone Reports, 2021, 15, 101110.	0.4	3
150	Mitochondrial Disease and Hearing Loss in Children: A Systematic Review. Laryngoscope, 2022, 132, 2459-2472.	2.0	3
151	Three Novel EPCAM Variants Causing Tufting Enteropathy in Three Families. Children, 2021, 8, 503.	1.5	2
152	Dimensions of the anterior cruciate ligament and thickness of the distal femoral growth plate in children: a MRI-based study. Archives of Orthopaedic and Trauma Surgery, 2022, , 1.	2.4	2
153	Pain during Cast Wedging of Forearm Shaft and Distal Forearm Fractures in Children Aged 3 to 12 Yearsâ€”A Prospective, Observational Study. Children, 2020, 7, 229.	1.5	1
154	Differences between Two Methods to Stabilize Supramalleolar Osteotomies in Childrenâ€”A Retrospective Case Series. Children, 2021, 8, 86.	1.5	1
155	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
156	Mitochondrial Transporter Defects: Successful Treatment with Ketogenic Diet Therapy. Neuropediatrics, 2021, 52, .	0.6	0
157	The switch in the diagnosis of mitochondrial diseases from the classical 'function first' to the NGS-based 'genetics first' diagnostic era. Medycyna Wieku Rozwojowego, 2020, 24, 47-52.	0.2	0