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	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
2	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
3	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
4	Biallelic BUB1 mutations cause microcephaly, developmental delay, and variable effects on cohesion and chromosome segregation. Science Advances, 2022, 8, eabk0114.	10.3	11
5	Mitochondrial Disease and Hearing Loss in Children: A Systematic Review. Laryngoscope, 2022, 132, 2459-2472.	2.0	3
6	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
7	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
8	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
9	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	2.4	16
10	Differences between Two Methods to Stabilize Supramalleolar Osteotomies in Childrenâ€”A Retrospective Case Series. Children, 2021, 8, 86.	1.5	1
11	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
12	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
13	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
14	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	8.2	16
15	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
16	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	2.2	4
17	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
18	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	5.3	3

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19	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
20	Three Novel EPCAM Variants Causing Tufting Enteropathy in Three Families. Children, 2021, 8, 503.	1.5	2
21	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. Molecular Genetics and Metabolism, 2021, 133, 362-371.	1.1	5
22	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
23	Congenital disorders of glycosylation with defective fucosylation. Journal of Inherited Metabolic Disease, 2021, 44, 1441-1452.	3.6	8
24	A spoonful of Lâ€fucoseâ€"an efficient therapy for GFUSâ€CDG, a new glycosylation disorder. EMBO Molecular Medicine, 2021, 13, e14332.	6.9	13
25	A novel cryptic splice site mutation in COL1A2 as a cause of osteogenesis imperfecta. Bone Reports, 2021, 15, 101110.	0.4	3
26	Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK1 Variants. Neuropediatrics, 2021, 52, 123-125.	0.6	4
27	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	6.9	5
28	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
29	Mitochondrial Transporter Defects: Successful Treatment with Ketogenic Diet Therapy. Neuropediatrics, 2021, 52, .	0.6	0
30	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
31	Defining clinical subgroups and genotypeâ€"phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
32	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
33	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
34	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
35	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
36	Age-Related Deterioration of Mitochondrial Function in the Intestine. Oxidative Medicine and Cellular Longevity, 2020, 2020, 1-12.	4.0	11

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37	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
38	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043.	1.4	90
39	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
40	Serine Catabolism Feeds NADH when Respiration Is Impaired. Cell Metabolism, 2020, 31, 809-821.e6.	16.2	118
41	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
42	Changes in the expression of oxidative phosphorylation complexes in the aging intestinal mucosa. Experimental Gerontology, 2020, 135, 110924.	2.8	17
43	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
44	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	2.4	13
45	Mutations in NDUFS1 Cause Metabolic Reprogramming and Disruption of the Electron Transfer. Cells, 2019, 8, 1149.	4.1	30
46	Severe Deoxyguanosine Kinase Deficiency in Austria. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e1-e6.	1.8	13
47	Cholineâ€relatedâ€inherited metabolic diseasesâ€"A mini review. Journal of Inherited Metabolic Disease, 2019, 42, 237-242.	3.6	42
48	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
49	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
50	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
51	BOLA (BolA Family Member 3) Deficiency Controls Endothelial Metabolism and Glycine Homeostasis in Pulmonary Hypertension. Circulation, 2019, 139, 2238-2255.	1.6	54
52	Damage control orthopedics applied in an 8-year-old child with life-threatening multiple injuries. Medicine (United States), 2019, 98, e15294.	1.0	5
53	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
54	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

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55	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
56	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
57	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
58	Melanoma tumors exhibit a variable but distinct metabolic signature. Experimental Dermatology, 2018, 27, 204-207.	2.9	21
59	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
60	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
61	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
62	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
63	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
64	Alterations of Oxidative Phosphorylation Complexes in Papillary Thyroid Carcinoma. Cells, 2018, 7, 40.	4.1	4
65	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
66	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
67	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
68	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
69	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
70	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
71	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	7.6	57
72	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

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73	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
74	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
75	Sternal Fracture in Children: Diagnosis by Ultrasonography. European Journal of Pediatric Surgery Reports, 2017, 05, e39-e42.	0.5	10
76	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
77	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
78	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion, 2017, 37, 55-61.	3.4	20
79	Effects of alpha-melanocyte-stimulating hormone on mitochondrial energy metabolism in rats of different age-groups. Neuropeptides, 2017, 64, 123-130.	2.2	9
80	Oxidative Phosphorylation System in Gastric Carcinomas and Gastritis. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-14.	4.0	20
81	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
82	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
83	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
84	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
85	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
86	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
87	Disorders of Oxidative Phosphorylation. , 2016, , 223-242.		6
88	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	12.8	178
89	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
90	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

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91	Alterations of oxidative phosphorylation in meningiomas and peripheral nerve sheath tumors. Neuro-Oncology, 2016, 18, 184-194.	1.2	13
92	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
93	Deficiency of respiratory chain complex I in Hashimoto thyroiditis. Mitochondrion, 2016, 26, 1-6.	3.4	8
94	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
95	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
96	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	2.3	81
97	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	2.7	25
98	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	1.5	8
99	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
100	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	3.6	44
101	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion, 2015, 21, 12-18.	3.4	51
102	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
103	Spectrum of combined respiratory chain defects. Journal of Inherited Metabolic Disease, 2015, 38, 629-640.	3.6	102
104	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
105	Lipid metabolism in mitochondrial membranes. Journal of Inherited Metabolic Disease, 2015, 38, 137-144.	3.6	60
106	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
107	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
108	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

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109	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
110	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
111	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
112	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
113	Lipoic acid biosynthesis defects. Journal of Inherited Metabolic Disease, 2014, 37, 553-563.	3.6	191
114	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
115	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	6.2	127
116	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
117	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
118	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
119	Protein Sets Define Disease States and Predict In Vivo Effects of Drug Treatment. Molecular and Cellular Proteomics, 2013, 12, 1965-1979.	3.8	29
120	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
121	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
122	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
123	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
124	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
125	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
126	Functional Differences between Mitochondrial Haplogroup T and Haplogroup H in HEK293 Cybrid Cells. PLoS ONE, 2012, 7, e52367.	2.5	50

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127	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
128	Mitochondrial Haplogroups and Control Region Polymorphisms in Age-Related Macular Degeneration: A Case-Control Study. PLoS ONE, 2012, 7, e30874.	2.5	54
129	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
130	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
131	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
132	Loss of mitochondria in ganglioneuromas. Frontiers in Bioscience - Elite, 2011, E3, 179-186.	1.8	20
133	Alterations of respiratory chain complexes in sporadic pheochromocytoma. Frontiers in Bioscience - Elite, 2011, E3, 194-200.	1.8	12
134	Heterogeneity of mitochondrial energy metabolism in classical triphasic Wilms tumor. Frontiers in Bioscience - Elite, 2011, E3, 187-193.	1.8	16
135	Respiratory chain complex I is a mitochondrial tumor suppressor of oncocytic tumors. Frontiers in Bioscience - Elite, 2011, E3, 315-325.	1.8	34
136	Mitochondrial Haplogroups, Control Region Polymorphisms and Malignant Melanoma: A Study in Middle European Caucasians. PLoS ONE, 2011, 6, e27192.	2.5	43
137	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
138	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
139	The Mitochondrial T16189C Polymorphism Is Associated with Coronary Artery Disease in Middle European Populations. PLoS ONE, 2011, 6, e16455.	2.5	70
140	Low aerobic mitochondrial energy metabolism in poorly- or undifferentiated neuroblastoma. BMC Cancer, 2010, 10, 149.	2.6	72
141	Mitochondrial encephalocardio-myopathy with early neonatal onset due to TMEM70 mutation. Archives of Disease in Childhood, 2010, 95, 296-301.	1.9	72
142	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
143	Mitochondrial Haplogroups and Control Region Polymorphisms Are Not Associated with Prostate Cancer in Middle European Caucasians. PLoS ONE, 2009, 4, e6370.	2.5	25
144	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

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145	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
146	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	21.4	183
147	Loss of Complex I due to Mitochondrial DNA Mutations in Renal Oncocytoma. Clinical Cancer Research, 2008, 14, 2270-2275.	7.0	154
148	Mitochondrial Phosphateâ€“Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. American Journal of Human Genetics, 2007, 80, 478-484.	6.2	142
149	Fractures and Dislocations of the Foot in Children. Clinics in Podiatric Medicine and Surgery, 2006, 23, 167-189.	0.6	9
150	A novel sporadic mutation G14739A of the mitochondrial tRNAGlu in a girl with exercise intolerance. Neuromuscular Disorders, 2006, 16, 874-877.	0.6	13
151	Multiplex primer extension analysis for rapid detection of major European mitochondrial haplogroups. Electrophoresis, 2006, 27, 3864-3868.	2.4	21
152	Rapid screening of the entire mitochondrial DNA for low-level heteroplasmic mutations. Mitochondrion, 2005, 5, 282-296.	3.4	43
153	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
154	Decrease of mitochondrial DNA content and energy metabolism in renal cell carcinoma. Carcinogenesis, 2004, 25, 1005-1010.	2.8	144
155	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
156	Severe depletion of mitochondrial DNA in spinal muscular atrophy. Acta Neuropathologica, 2003, 105, 245-251.	7.7	72
157	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