John Christodoulou

List of Publications by Citations

274 papers	11,045 citations	56 h-index	93 g-index
302 ext. papers	12,830 ext. citations	5.5 avg, IF	5.83 L-index

#	Paper	IF	Citations
274	Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, **2010**, 68, 944-50	9.4	804
273	Leigh syndrome: clinical features and biochemical and DNA abnormalities. *Annals of Neurology*, **1996**, 39, 343-51	9.4	614
272	Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing. *Science Translational Medicine*, **2012**, 4, 118ra10	17.5	362
271	Mutations of CDKL5 cause a severe neurodevelopmental disorder with infantile spasms and mental retardation. *American Journal of Human Genetics*, **2004**, 75, 1079-93	11	352
270	Rett syndrome in Australia: a review of the epidemiology. *Journal of Pediatrics*, **2006**, 148, 347-52	3.6	241
269	Mutation of the mitochondrial tyrosyl-tRNA synthetase gene, YARS2, causes myopathy, lactic acidosis, and sideroblastic anemia--MLASA syndrome. *American Journal of Human Genetics*, **2010**, 87, 52-9	11	190
268	Mecp2 deficiency is associated with learning and cognitive deficits and altered gene activity in the hippocampal region of mice. *Brain*, **2006**, 129, 887-98	11.2	172
267	The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. *European Journal of Human Genetics*, **2013**, 21, 266-73	5.3	161
266	Environmental enrichment ameliorates a motor coordination deficit in a mouse model of Rett syndrome--Mecp2 gene dosage effects and BDNF expression. *European Journal of Neuroscience*, **2008**, 27, 3342-50	3.5	151
265	Mutation of C20orf7 disrupts complex I assembly and causes lethal neonatal mitochondrial disease. *American Journal of Human Genetics*, **2008**, 83, 468-78	11	150
264	Early onset seizures and Rett-like features associated with mutations in CDKL5. *European Journal of Human Genetics*, **2005**, 13, 1113-20	5.3	143
263	RettBASE: The IRSA MECP2 variation database-a new mutation database in evolution. *Human Mutation*, **2003**, 21, 466-72	4.7	130
262	Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial translation. *Cell Metabolism*, **2011**, 14, 428-34	24.6	123
261	Rett syndrome: clinical review and genetic update. *Journal of Medical Genetics*, **2005**, 42, 1-7	5.8	121
260	Guidelines for reporting clinical features in cases with MECP2 mutations. *Brain and Development*, **2001**, 23, 208-11	2.2	121
259	Expanded newborn screening: outcome in screened and unscreened patients at age 6 years. *Pediatrics*, **2009**, 124, e241-8	7.4	117
258	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. *Brain*, **2014**, 137, 44-56	11.2	115

257	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genetics in Medicine*, **2017**, 19,	8.1	113
256	Interpreting missense variants: comparing computational methods in human disease genes CDKN2A, MLH1, MSH2, MECP2, and tyrosinase (TYR). *Human Mutation*, **2007**, 28, 683-93	4.7	111
255	Pathophysiological mechanisms of dominant and recessive GLRA1 mutations in hyperekplexia. *Journal of Neuroscience*, **2010**, 30, 9612-20	6.6	102
254	Early treatment of Menkes disease with parenteral Cooper-Histidine: Long-term follow-up of four treated patients **1998**, 76, 154-164		101
253	Rett syndrome: new clinical and molecular insights. *European Journal of Human Genetics*, **2006**, 14, 896-903	3.3	94
252	The effects of large neutral amino acid supplements in PKU: an MRS and neuropsychological study. *Molecular Genetics and Metabolism*, **2007**, 91, 48-54	3.7	88
251	Describing the phenotype in Rett syndrome using a population database. *Archives of Disease in Childhood*, **2003**, 88, 38-43	2.2	88
250	Early copper-histidine treatment for Menkes disease. *Nature Genetics*, **1996**, 12, 11-3	36.3	88
249	Sporadic and Familial Congenital Cataracts: Mutational Spectrum and New Diagnoses Using Next-Generation Sequencing. *Human Mutation*, **2016**, 37, 371-84	4.7	87
248	Efficacy and safety of cyclic pyranopterin monophosphate substitution in severe molybdenum cofactor deficiency type A: a prospective cohort study. *Lancet, The*, **2015**, 386, 1955-1963	40	86
247	Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria Type 2, is caused by a microdeletion in the riboflavin transporter gene GPR172B. *Human Mutation*, **2011**, 32, E1976-84	4.7	85
246	Phenylketonuria: a review of current and future treatments. *Translational Pediatrics*, **2015**, 4, 304-17	4.2	85
245	Effects of MECP2 mutation type, location and X-inactivation in modulating Rett syndrome phenotype. *American Journal of Medical Genetics Part A*, **2003**, 118A, 103-14		84
244	Mutations in LYRM4, encoding iron-sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. *Human Molecular Genetics*, **2013**, 22, 4460-73	5.6	81
243	Mutations in the UQCC1-interacting protein, UQCC2, cause human complex III deficiency associated with perturbed cytochrome b protein expression. *PLoS Genetics*, **2013**, 9, e1004034	6	79
242	Predictors of seizure onset in Rett syndrome. *Journal of Pediatrics*, **2006**, 149, 542-7	3.6	79
241	Inherited bone marrow failure associated with germline mutation of ACD, the gene encoding telomere protein TPP1. *Blood*, **2014**, 124, 2767-74	2.2	75
240	Early progressive encephalopathy in boys and MECP2 mutations. *Neurology*, **2006**, 67, 164-6	6.5	75

239	PRPS1 mutations: four distinct syndromes and potential treatment. *American Journal of Human Genetics*, **2010**, 86, 506-18	11	74
238	Correlation between clinical severity in patients with Rett syndrome with a p.R168X or p.T158M MECP2 mutation, and the direction and degree of skewing of X-chromosome inactivation. *Journal of Medical Genetics*, **2007**, 44, 148-52	5.8	72
237	Downstream targets of methyl CpG binding protein 2 and their abnormal expression in the frontal cortex of the human Rett syndrome brain. *BMC Neuroscience*, **2010**, 11, 53	3.2	71
236	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. *Journal of Inherited Metabolic Disease*, **2017**, 40, 261-269	5.4	70
235	Comprehensive mutation analysis of GLDC, AMT, and GCSH in nonketotic hyperglycinemia. *Human Mutation*, **2006**, 27, 343-52	4.7	70
234	Mutations in SLC33A1 cause a lethal autosomal-recessive disorder with congenital cataracts, hearing loss, and low serum copper and ceruloplasmin. *American Journal of Human Genetics*, **2012**, 90, 61-8	11	69
233	Mitochondrial myopathy with tRNA(Leu(UUR)) mutation and complex I deficiency responsive to riboflavin. *Journal of Pediatrics*, **1997**, 130, 138-45	3.6	69
232	Seizures in Rett syndrome: an overview from a one-year calendar study. *European Journal of Paediatric Neurology*, **2007**, 11, 310-7	3.8	68
231	Potential of AAV vectors in the treatment of metabolic disease. *Gene Therapy*, **2008**, 15, 831-9	4	67
230	Patients with the R133C mutation: is their phenotype different from patients with Rett syndrome with other mutations?. *Journal of Medical Genetics*, **2003**, 40, e52	5.8	67
229	Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet Journal of Rare Diseases*, **2017**, 12, 68	4.2	66
228	RettBASE: Rett syndrome database update. *Human Mutation*, **2017**, 38, 922-931	4.7	65
227	Arts syndrome is caused by loss-of-function mutations in PRPS1. *American Journal of Human Genetics*, **2007**, 81, 507-18	11	65
226	Refining the phenotype of common mutations in Rett syndrome. *Journal of Medical Genetics*, **2004**, 41, 25-30	5.8	65
225	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. *Journal of Medical Genetics*, **2010**, 47, 211-6	5.8	64
224	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. *JAMA - Journal of the American Medical Association*, **2020**, 323, 2503-2511	27.4	63
223	Rett syndrome: clinical characteristics and recent genetic advances. *Disability and Rehabilitation*, **2001**, 23, 98-106	2.4	61
222	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. *American Journal of Human Genetics*, **2016**, 99, 1229-1244	11	59

221	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. *American Journal of Human Genetics*, **2017**, 101, 239-254	11	59
220	The common BDNF polymorphism may be a modifier of disease severity in Rett syndrome. *Neurology*, **2009**, 72, 1242-7	6.5	59
219	Ornithine transcarbamylase deficiency presenting with strokelike episodes. *Journal of Pediatrics*, **1993**, 122, 423-5	3.6	59
218	Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. *Human Molecular Genetics*, **2015**, 24, 6146-59	5.6	56
217	Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. *Molecular Genetics and Metabolism*, **2015**, 114, 388-96	3.7	56
216	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. *Neurology*, **2016**, 87, 2206-2213	6.5	51
215	Updating the profile of C-terminal MECP2 deletions in Rett syndrome. *Journal of Medical Genetics*, **2010**, 47, 242-8	5.8	51
214	Predictors of scoliosis in Rett syndrome. *Journal of Child Neurology*, **2006**, 21, 809-13	2.5	51
213	A founder mutation in PET100 causes isolated complex IV deficiency in Lebanese individuals with Leigh syndrome. *American Journal of Human Genetics*, **2014**, 94, 209-22	11	49
212	Prolonged QT interval in Rett syndrome. *Archives of Disease in Childhood*, **1999**, 80, 470-2	2.2	49
211	Pathology of hepatic peroxisomes and mitochondria in patients with peroxisomal disorders. *Virchows Archiv A, Pathological Anatomy and Histopathology*, **1990**, 416, 255-64		49
210	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. *Human Molecular Genetics*, **2015**, 24, 2297-307	5.6	48
209	Mutations in CYC1, encoding cytochrome c1 subunit of respiratory chain complex III, cause insulin-responsive hyperglycemia. *American Journal of Human Genetics*, **2013**, 93, 384-9	11	48
208	There is variability in the attainment of developmental milestones in the CDKL5 disorder. *Journal of Neurodevelopmental Disorders*, **2015**, 7, 2	4.6	47
207	MECP2 genomic structure and function: insights from ENCODE. *Nucleic Acids Research*, **2008**, 36, 6035-47	20.1	47
206	Glutaric aciduria type I: outcome following detection by newborn screening. *Journal of Inherited Metabolic Disease*, **2008**, 31, 503-7	5.4	45
205	The molecular basis of malonyl-CoA decarboxylase deficiency. *American Journal of Human Genetics*, **1999**, 65, 318-26	11	45
204	Peroxisomal assembly defects: clinical, pathologic, and biochemical findings in two patients in a newly identified complementation group. *Journal of Pediatrics*, **1995**, 127, 596-9	3.6	45

203	Mitochondrial dysfunction in the skeletal muscle of a mouse model of Rett syndrome (RTT): implications for the disease phenotype. *Mitochondrion*, **2014**, 15, 10-7	4.9	44
202	Mutation screening of the mitochondrial genome using denaturing high-performance liquid chromatography. *Molecular Genetics and Metabolism*, **2005**, 84, 61-74	3.7	44
201	The Genetic Landscape and Epidemiology of Phenylketonuria. *American Journal of Human Genetics*, **2020**, 107, 234-250	11	44
200	Rett Syndrome: A Genetic Update and Clinical Review Focusing on Comorbidities. *ACS Chemical Neuroscience*, **2018**, 9, 167-176	5.7	44
199	Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. *Orphanet Journal of Rare Diseases*, **2013**, 8, 193	4.2	43
198	Patient with a 22q11.2 deletion with no overlap of the minimal DiGeorge syndrome critical region (MDGCR) **1999**, 86, 27-33		43
197	Disorders of riboflavin metabolism. *Journal of Inherited Metabolic Disease*, **2019**, 42, 608-619	5.4	42
196	Update on transcobalamin deficiency: clinical presentation, treatment and outcome. *Journal of Inherited Metabolic Disease*, **2014**, 37, 461-73	5.4	42
195	Enlarged temporal lobes in Turner syndrome: an X-chromosome effect?. *Cerebral Cortex*, **2004**, 14, 156-64	5.1	42
194	Acylcarnitine profiles in fibroblasts from patients with respiratory chain defects can resemble those from patients with mitochondrial fatty acid beta-oxidation disorders. *Metabolism: Clinical and Experimental*, **2002**, 51, 366-71	12.7	41
193	Rett syndrome: randomized controlled trial of L-carnitine. *Journal of Child Neurology*, **1999**, 14, 162-7	2.5	41
192	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. *Molecular Genetics and Metabolism*, **2016**, 118, 178-184	3.7	40
191	The Natural History of Scoliosis in Females With Rett Syndrome. *Spine*, **2016**, 41, 856-63	3.3	40
190	Delineation of large deletions of the MECP2 gene in Rett syndrome patients, including a familial case with a male proband. *European Journal of Human Genetics*, **2007**, 15, 1218-29	5.3	40
189	Neurodevelopmental Outcome and Treatment Efficacy of Benzoate and Dextromethorphan in Siblings with Attenuated Nonketotic Hyperglycinemia. *Journal of Pediatrics*, **2016**, 170, 234-9	3.6	39
188	Diagnostic yield of targeted massively parallel sequencing in children with epileptic encephalopathy. *Seizure: the Journal of the British Epilepsy Association*, **2018**, 59, 132-140	3.2	39
187	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. *American Journal of Human Genetics*, **2019**, 105, 7-14	11	39
186	A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. *Human Genetics*, **2012**, 131, 187-200	6.3	38

185	A population-based approach to the investigation of osteopenia in Rett syndrome. *Developmental Medicine and Child Neurology*, **1999**, 41, 323-8	3.3	38
184	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. *JIMD Reports*, **2016**, 28, 49-57	1.9	37
183	NTNG1 mutations are a rare cause of Rett syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 691-4	2.5	37
182	Transient multiple acyl-CoA dehydrogenation deficiency in a newborn female caused by maternal riboflavin deficiency. *Molecular Genetics and Metabolism*, **2007**, 92, 109-14	3.7	37
181	Functional abilities in children and adults with the CDKL5 disorder. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2860-2869	2.5	36
180	Progressive deafness-dystonia due to SERAC1 mutations: A study of 67 cases. *Annals of Neurology*, **2017**, 82, 1004-1015	9.4	36
179	New DGK gene mutations in the hepatocerebral form of mitochondrial DNA depletion syndrome. *Archives of Neurology*, **2005**, 62, 745-7		36
178	Allogeneic bone marrow transplantation: cure for familial Mediterranean fever. *Blood*, **2002**, 100, 774-7	2.2	36
177	MeCP2 deficiency is associated with reduced levels of tubulin acetylation and can be restored using HDAC6 inhibitors. *Journal of Molecular Medicine*, **2015**, 93, 63-72	5.5	35
176	Distinct expression profiles of Mecp2 transcripts with different lengths of 3ℕTR in the brain and visceral organs during mouse development. *Genomics*, **2005**, 85, 441-52	4.3	33
175	Sleep dysfunction in Rett syndrome: lack of age related decrease in sleep duration. *Brain and Development*, **2001**, 23 Suppl 1, S101-3	2.2	33
174	Medium-term open label trial of L-carnitine in Rett syndrome. *Brain and Development*, **2001**, 23 Suppl 1, S85-9	2.2	31
173	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
172	InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 709-13	2.5	30
171	X chromosome inactivation patterns in brain in Rett syndrome: implications for the disease phenotype. *Brain and Development*, **2005**, 27, 266-70	2.2	30
170	Genotype and early development in Rett syndrome: the value of international data. *Brain and Development*, **2005**, 27 Suppl 1, S59-S68	2.2	30
169	14q12 microdeletions excluding FOXG1 give rise to a congenital variant Rett syndrome-like phenotype. *European Journal of Human Genetics*, **2013**, 21, 522-7	5.3	29
168	Cyclin-dependent kinase-like 5 (CDKL5) mutation screening in Rett syndrome and related disorders. *Twin Research and Human Genetics*, **2010**, 13, 168-78	2.2	29

167	Inherited surfactant deficiency caused by uniparental disomy of rare mutations in the surfactant protein-B and ATP binding cassette, subfamily a, member 3 genes. *Journal of Pediatrics*, **2009**, 155, 854-859.e1	3.6	29
166	Tetrahydrobiopterin-responsive phenylketonuria: the New South Wales experience. *Molecular Genetics and Metabolism*, **2005**, 86 Suppl 1, S81-5	3.7	28
165	p.R270X MECP2 mutation and mortality in Rett syndrome. *European Journal of Human Genetics*, **2005**, 13, 1235-8	5.3	28
164	Flow cytometry in the study of mitochondrial respiratory chain disorders. *Mitochondrion*, **2002**, 1, 437-45	4.9	28
163	Malonic aciduria. *Brain and Development*, **1994**, 16 Suppl, 7-11	2.2	28
162	Diagnosis of possible mitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
161	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. *Human Mutation*, **2012**, 33, 1207-15	4.7	27
160	The PRPP synthetase spectrum: what does it demonstrate about nucleotide syndromes?. *Nucleosides, Nucleotides and Nucleic Acids*, **2011**, 30, 1129-39	1.4	27
159	MECP2 and beyond: phenotype-genotype correlations in Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 669-74	2.5	27
158	A novel system for assigning the mode of inheritance in mitochondrial disorders using cybrids and rhodamine 6G. *Human Molecular Genetics*, **1999**, 8, 1691-7	5.6	27
157	Atypical pyroglutamic aciduria: possible role of paracetamol. *Journal of Inherited Metabolic Disease*, **1990**, 13, 755-6	5.4	27
156	Mitochondrial respiratory chain disorders in childhood: insights into diagnosis and management in the new era of genomic medicine. *Biochimica Et Biophysica Acta - General Subjects*, **2014**, 1840, 1368-79	4	26
155	Coexistent MEFV and CIAS1 mutations manifesting as familial Mediterranean fever plus deafness. *Annals of the Rheumatic Diseases*, **2007**, 66, 1541	2.4	25
154	Surfactant protein B deficiency: clinical, histological and molecular evaluation. *Journal of Paediatrics and Child Health*, **1999**, 35, 214-20	1.3	25
153	Clinical approach to inborn errors of metabolism presenting in the newborn period. *Journal of Paediatrics and Child Health*, **2002**, 38, 511-7	1.3	24
152	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. *Genetics in Medicine*, **2019**, 21, 2823-2826	8.1	23
151	Alleviating transcriptional inhibition of the norepinephrine slc6a2 transporter gene in depolarized neurons. *Journal of Neuroscience*, **2010**, 30, 1494-501	6.6	23
150	Automated analysis of mitochondrial enzymes in cultured skin fibroblasts. *Analytical Biochemistry*, **1998**, 259, 176-80	3.1	23

149	Lost in translation: translational interference from a recurrent mutation in exon 1 of MECP2. *Journal of Medical Genetics*, **2006**, 43, 470-7	5.8	23
148	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. *Genetics in Medicine*, **2020**, 22, 1254-1261	8.1	22
147	Trimethylaminuria: an under-recognised and socially debilitating metabolic disorder. *Journal of Paediatrics and Child Health*, **2012**, 48, E153-5	1.3	22
146	Phosphoribosylpyrophosphate synthetase superactivity and recurrent infections is caused by a p.Val142Leu mutation in PRS-I. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 455-60	2.5	22
145	Hearing loss and PRPS1 mutations: Wide spectrum of phenotypes and potential therapy. *International Journal of Audiology*, **2013**, 52, 23-8	2.6	22
144	Rett syndrome: clinical update and review of recent genetic advances. *Journal of Paediatrics and Child Health*, **1999**, 35, 419-26	1.3	22
143	Atypical nonketotic hyperglycinemia confirmed by assay of the glycine cleavage system in lymphoblasts. *Journal of Pediatrics*, **1993**, 123, 100-2	3.6	22
142	Carnitine palmitoyltransferase II deficiency: a new cause of recurrent pancreatitis. *Journal of Pediatrics*, **1994**, 124, 938-40	3.6	22
141	Biallelic variants in and cause deafness and (ovario)leukodystrophy. *Neurology*, **2019**, 92, e1225-e1237	6.5	21
140	Whole-exome sequencing identifies novel variants in PNPT1 causing oxidative phosphorylation defects and severe multisystem disease. *European Journal of Human Genetics*, **2016**, 25, 79-84	5.3	21
139	Congenital disorder of glycosylation type Ia: heterogeneity in the clinical presentation from multivisceral failure to hyperinsulinaemic hypoglycaemia as leading symptoms in three infants with phosphomannomutase deficiency. *Journal of Inherited Metabolic Disease*, **2009**, 32 Suppl 1, S241-51	5.4	21
138	First prenatal diagnosis of the carnitine transporter defect. *American Journal of Medical Genetics Part A*, **1996**, 66, 21-4		21
137	Mutations in RARS cause a hypomyelination disorder akin to Pelizaeus-Merzbacher disease. *European Journal of Human Genetics*, **2017**, 25, 1134-1141	5.3	20
136	Transcription factor 4 and myocyte enhancer factor 2C mutations are not common causes of Rett syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 713-9	2.5	20
135	An improved ultra performance liquid chromatography-tandem mass spectrometry method for the determination of alloisoleucine and branched chain amino acids in dried blood samples. *Annals of Clinical Biochemistry*, **2011**, 48, 468-70	2.2	20
134	Origin of the prevalent SFTPB indel g.1549C > GAA (121ins2) mutation causing surfactant protein B (SP-B) deficiency. *American Journal of Medical Genetics, Part A*, **2006**, 140, 62-9	2.5	20
133	Quantitative fibroblast acylcarnitine profiles in mitochondrial fatty acid beta-oxidation defects: phenotype/metabolite correlations. *Molecular Genetics and Metabolism*, **2002**, 76, 327-34	3.7	20
132	Experience of gastrostomy using a quality care framework: the example of rett syndrome. *Medicine (United States)*, **2014**, 93, e328	1.8	19

131	Costeff optic atrophy syndrome: new clinical case and novel molecular findings. *Journal of Inherited Metabolic Disease*, **2008**, 31 Suppl 2, S419-23	5.4	19
130	Autopsy findings in two siblings with infantile Refsum disease. *Acta Neuropathologica*, **1992**, 83, 190-5	14.3	19
129	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. *Brain*, **2020**, 143, 112-130	11.2	19
128	Surgical fusion of early onset severe scoliosis increases survival in Rett syndrome: a cohort study. *Developmental Medicine and Child Neurology*, **2016**, 58, 632-8	3.3	19
127	A novel mutation in NDUFB11 unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. *Clinical Genetics*, **2017**, 91, 441-447	4	18
126	Affective dysfunction in a mouse model of Rett syndrome: Therapeutic effects of environmental stimulation and physical activity. *Developmental Neurobiology*, **2016**, 76, 209-24	3.2	18
125	Intragenic complementation at the human argininosuccinate lyase locus. Identification of the major complementing alleles. *Journal of Biological Chemistry*, **1997**, 272, 6777-83	5.4	18
124	Perimortem laboratory investigation of genetic metabolic disorders. *Seminars in Fetal and Neonatal Medicine*, **2004**, 9, 275-80		18
123	Metacarpophalangeal pattern profile and bone age in Rett syndrome: further radiological clues to the diagnosis. *American Journal of Medical Genetics Part A*, **1999**, 83, 88-95		18
122	Respiratory chain complex III [correction of complex] in deficiency with pruritus: a novel vitamin responsive clinical feature. *Journal of Pediatrics*, **1999**, 134, 352-4	3.6	18
121	Metabolic stroke in methylmalonic acidemia. *Journal of Pediatrics*, **1989**, 115, 499-500	3.6	18
120	In vitro read-through of phenylalanine hydroxylase (PAH) nonsense mutations using aminoglycosides: a potential therapy for phenylketonuria. *Journal of Inherited Metabolic Disease*, **2013**, 36, 955-9	5.4	17
119	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. *Genetics in Medicine*, **2019**, 21, 1021-1026	8.1	17
118	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. *Orphanet Journal of Rare Diseases*, **2018**, 13, 86	4.2	16
117	Reduced proportion of Purkinje cells expressing paternally derived mutant Mecp2308 allele in female mouse cerebellum is not due to a skewed primary pattern of X-chromosome inactivation. *Human Molecular Genetics*, **2005**, 14, 1851-61	5.6	16
116	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. *American Journal of Human Genetics*, **2018**, 103, 125-130	11	15
115	Mitochondrial respiratory chain hepatopathies: role of liver transplantation. A case series of five patients. *JIMD Reports*, **2012**, 4, 5-11	1.9	15
114	"Reducing body"-like inclusions in skeletal muscle in childhood-onset acid maltase deficiency. *Acta Neuropathologica*, **1992**, 85, 111-5	14.3	15

113	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. *Med*, **2021**, 2, 49-73	31.7	15
112	The personal utility and uptake of genomic sequencing in pediatric and adult conditions: eliciting societal preferences with three discrete choice experiments. *Genetics in Medicine*, **2020**, 22, 1311-1319	8.1	14
111	The phenotypic spectrum of germline variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. *Haematologica*, **2018**, 103, 2008-2015	6.6	14
110	OPA3 mutation screening in patients with unexplained 3-methylglutaconic aciduria. *Journal of Inherited Metabolic Disease*, **2005**, 28, 525-32	5.4	14
109	Rett syndrome: significant clinical overlap with Angelman syndrome but not with methylation status. *Journal of Child Neurology*, **1998**, 13, 448-51	2.5	14
108	Phenylketonuria: translating research into novel therapies. *Translational Pediatrics*, **2014**, 3, 49-62	4.2	14
107	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. *American Journal of Human Genetics*, **2020**, 106, 779-792	11	14
106	Utility of next-generation sequencing technologies for the efficient genetic resolution of haematological disorders. *Clinical Genetics*, **2016**, 89, 163-72	4	13
105	Neonatal onset of medium-chain acyl-coenzyme A dehydrogenase deficiency with confusing biochemical features. *Journal of Pediatrics*, **1995**, 126, 65-8	3.6	13
104	Ring chromosome 22 karyotype in a patient with Opitz (BBBG) syndrome. *American Journal of Medical Genetics Part A*, **1990**, 37, 422-4		13
103	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. *PLoS ONE*, **2017**, 12, e0178125	3.7	13
102	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. *Mitochondrion*, **2019**, 44, 58-64	4.9	13
101	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. *American Journal of Human Genetics*, **2020**, 106, 467-483	11	12
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