John Christodoulou

List of Publications by Year in Descending Order

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	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children.. *Molecular Genetics and Metabolism*, **2022**, 135, 63-71	3.7	1
273	Genetic Metabolic Disease **2022**, 267-289		
272	Biallelic Variants in Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function.. *International Journal of Molecular Sciences*, **2022**, 23,	6.3	1
271	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
270	Reply: Niacin therapy improves outcome and normalizes metabolic abnormalities in a NAXD-deficient patient.. *Brain*, **2022**,	11.2	0
269	Distinct diagnostic trajectories in NBAS-associated acute liver failure highlights the need for timely functional studies.. *JIMD Reports*, **2022**, 63, 240-249	1.9	
268	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
267	TAT-MeCP2 protein variants rescue disease phenotypes in human and mouse models of Rett syndrome.. *International Journal of Biological Macromolecules*, **2022**, 209, 972-983	7.9	0
266	Expression, Purification, Characterization and Cellular Uptake of MeCP2 Variants.. *Protein Journal*, **2022**, 1	3.9	0
265	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants.. *Genetics in Medicine*, **2021**,	8.1	3
264	Co-therapy with S-adenosylmethionine and nicotinamide riboside improves t-cell survival and function in Arts Syndrome (PRPS1 deficiency). *Molecular Genetics and Metabolism Reports*, **2021**, 26, 100709	1.8	0
263	ALG13 X-linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. *Journal of Inherited Metabolic Disease*, **2021**, 44, 1001-1012	5.4	3
262	Preferences and values for rapid genomic testing in critically ill infants and children: a discrete choice experiment. *European Journal of Human Genetics*, **2021**, 29, 1645-1653	5.3	3
261	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. *Genes*, **2021**, 12,	4.2	3
260	Genotype and defects in microtubule-based motility correlate with clinical severity in -associated neurological disorder. *Human Genetics and Genomics Advances*, **2021**, 2,	0.8	9
259	Ethically utilising COVID-19 host-genomic data. *Npj Genomic Medicine*, **2021**, 6, 31	6.2	2
258	Genomic sequencing for the diagnosis of childhood mitochondrial disorders: a health economic evaluation. *European Journal of Human Genetics*, **2021**,	5.3	1

257	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. *Genetics in Medicine*, **2021**, 23, 1705-1714	8.1	5
256	Abnormalities of mitochondrial dynamics and bioenergetics in neuronal cells from CDKL5 deficiency disorder. *Neurobiology of Disease*, **2021**, 155, 105370	7.5	3
255	Multiomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. *Human Mutation*, **2021**, 42, 19-24	4.7	9
254	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. *Med*, **2021**, 2, 49-73	31.7	15
253	The value of genomic sequencing in complex pediatric neurological disorders: a discrete choice experiment. *Genetics in Medicine*, **2021**, 23, 155-162	8.1	6
252	The long and winding road: perspectives of people and parents of children with mitochondrial conditions negotiating management after diagnosis. *Orphanet Journal of Rare Diseases*, **2021**, 16, 310	4.2	1
251	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. *American Journal of Human Genetics*, **2021**, 108, 1692-1709	11	1
250	Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations. *Internal Medicine Journal*, **2021**,	1.6	1
249	A novel cause of DKC1-related bone marrow failure: Partial deletion of the 3? untranslated region. *EJHaem*, **2021**, 2, 157-166	0.9	0
248	Newborn bloodspot screening in the time of COVID-19. *Genetics in Medicine*, **2021**, 23, 1143-1150	8.1	3
247	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. *Genetics in Medicine*, **2020**, 22, 1254-1261	8.1	22
246	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7
245	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). *Human Genetics*, **2020**, 139, 1325-1343	6.3	8
244	Reply: NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness. *Brain*, **2020**, 143, e55	11.2	0
243	An Electrochemiluminescence-Based Assay for MeCP2 Protein Variants. *Journal of Visualized Experiments*, **2020**,	1.6	2
242	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
241	Activating variants in PDGFRB result in a spectrum of disorders responsive to imatinib monotherapy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1576-1591	2.5	11
240	TRAPPing a neurological disorder: from yeast to humans. *Autophagy*, **2020**, 16, 965-966	10.2	6

239	Bi-allelic LoF NRROS Variants Impairing Active TGF-β Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. *American Journal of Human Genetics*, **2020**, 106, 559-569	11	7
238	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
237	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (KIF1A). *Human Mutation*, **2020**, 41, 1761-1774	4.7	10
236	Reply: Recurrent bi-allelic splicing variant c.454+3A>G in TRAPPC4 is associated with progressive encephalopathy and muscle involvement. *Brain*, **2020**, 143, e30	11.2	
235	Clinical genomic testing: what matters to key stakeholders?. *European Journal of Human Genetics*, **2020**, 28, 866-873	5.3	8
234	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. *Molecular Genetics and Metabolism*, **2020**, 130, 49-57	3.7	11
233	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. *American Journal of Human Genetics*, **2020**, 106, 467-483	11	12
232	Parental health spillover effects of paediatric rare genetic conditions. *Quality of Life Research*, **2020**, 29, 2445-2454	3.7	6
231	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
230	The effect of emerging nutraceutical interventions for clinical and biological outcomes in multiple sclerosis: A systematic review. *Multiple Sclerosis and Related Disorders*, **2020**, 37, 101486	4	8
229	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. *Brain*, **2020**, 143, 112-130	11.2	19
228	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. *Human Mutation*, **2020**, 41, 1884-1891	4.7	4
227	Reply: Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. *Brain*, **2020**, 143, e84	11.2	
226	Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1333-1348	5.4	10
225	The Genetic Landscape and Epidemiology of Phenylketonuria. *American Journal of Human Genetics*, **2020**, 107, 234-250	11	44
224	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A (KAT6A). *Journal of Genetics and Genomics*, **2020**, 47, 650-654	4	1
223	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. *Journal of Experimental Medicine*, **2020**, 217,	16.6	4
222	The expanding LARS2 phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. *Human Mutation*, **2020**, 41, 1425-1434	4.7	7

221	Diagnosis of possible mitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
220	Disorders of riboflavin metabolism. *Journal of Inherited Metabolic Disease*, **2019**, 42, 608-619	5.4	42
219	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. *Genetics in Medicine*, **2019**, 21, 2823-2826	8.1	23
218	An electrochemiluminescence based assay for quantitative detection of endogenous and exogenously applied MeCP2 protein variants. *Scientific Reports*, **2019**, 9, 7929	4.9	2
217	Leigh syndrome caused by mutations in is associated with a better prognosis. *Annals of Clinical and Translational Neurology*, **2019**, 6, 515-524	5.3	9
216	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic protein-truncating variant. *Human Mutation*, **2019**, 40, 893-898	4.7	7
215	Biallelic variants in and cause deafness and (ovario)leukodystrophy. *Neurology*, **2019**, 92, e1225-e1237	6.5	21
214	Genome-wide transcriptomic and proteomic studies of Rett syndrome mouse models identify common signaling pathways and cellular functions as potential therapeutic targets. *Human Mutation*, **2019**, 40, 2184-2196	4.7	4
213	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. *American Journal of Human Genetics*, **2019**, 105, 7-14	11	39
212	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. *European Journal of Human Genetics*, **2019**, 27, 1821-1826	5.3	12
211	Whole exome sequencing reveals a de novo missense variant in in a Rett syndrome-like patient. *Clinical Case Reports (discontinued)*, **2019**, 7, 2476-2482	0.7	4
210	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic Variants. *Journal of Clinical Medicine*, **2019**, 8,	5.1	10
209	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. *Molecular Genetics and Metabolism*, **2019**, 126, 77-82	3.7	11
208	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
207	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. *Clinical Genetics*, **2019**, 95, 403-408	4	5
206	Paroxysmal dyskinesias with drowsiness and thalamic lesions in GABA transaminase deficiency. *Neurology*, **2019**, 92, 94-97	6.5	6
205	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. *Genetics in Medicine*, **2019**, 21, 1021-1026	8.1	17
204	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. *Mitochondrion*, **2019**, 44, 58-64	4.9	13

203	Tread carefully: A functional variant in the human NADPH oxidase 4 (NOX4) is not disease causing. *Molecular Genetics and Metabolism*, **2018**, 123, 382-387	3.7	
202	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
201	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
200	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
199	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
198	A Third Case of Glycogen Storage Disease IB and Giant Cell Tumour of the Mandible: A Disease Association or Iatrogenic Complication of Therapy. *JIMD Reports*, **2018**, 42, 5-8	1.9	5
197	Rett Syndrome: A Genetic Update and Clinical Review Focusing on Comorbidities. *ACS Chemical Neuroscience*, **2018**, 9, 167-176	5.7	44
196	A simple and efficient toolset for analysing mitochondrial trafficking in neuronal cells. *Acta Histochemica*, **2018**, 120, 797-805	2	4
195	Impact of Gastrostomy Placement on Nutritional Status, Physical Health, and Parental Well-Being of Females with Rett Syndrome: A Longitudinal Study of an Australian Population. *Journal of Pediatrics*, **2018**, 200, 188-195.e1	3.6	5
194	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
193	RettBASE: Rett syndrome database update. *Human Mutation*, **2017**, 38, 922-931	4.7	65
192	A novel mutation in GMPPA in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2246-2250	2.5	6
191	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. *Journal of Inherited Metabolic Disease*, **2017**, 40, 261-269	5.4	70
190	Whole Exome Sequencing Identifies the Genetic Basis of Late-Onset Leigh Syndrome in a Patient with MRI but Little Biochemical Evidence of a Mitochondrial Disorder. *JIMD Reports*, **2017**, 32, 117-124	1.9	7
189	Mutations in RARS cause a hypomyelination disorder akin to Pelizaeus-Merzbacher disease. *European Journal of Human Genetics*, **2017**, 25, 1134-1141	5.3	20
188	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genetics in Medicine*, **2017**, 19,	8.1	113
187	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
186	Response to Newman et al. *Genetics in Medicine*, **2017**, 19,	8.1	2

185	Progressive deafness-dystonia due to SERAC1 mutations: A study of 67 cases. *Annals of Neurology*, **2017**, 82, 1004-1015	9.4	36
184	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
183	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. *PLoS ONE*, **2017**, 12, e0178125	3.7	13
182	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. *JIMD Reports*, **2016**, 28, 49-57	1.9	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	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. *Mitochondrion*, **2016**, 30, 162-7	4.9	11
179	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. *Neurology*, **2016**, 87, 2206-2213	6.5	51
178	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
177	Extensive Variation in the Mutation Rate Between and Within Human Genes Associated with Mendelian Disease. *Human Mutation*, **2016**, 37, 488-94	4.7	11
176	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. *Molecular Genetics and Metabolism*, **2016**, 118, 178-184	3.7	40
175	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
174	Utility of next-generation sequencing technologies for the efficient genetic resolution of haematological disorders. *Clinical Genetics*, **2016**, 89, 163-72	4	13
173	The Natural History of Scoliosis in Females With Rett Syndrome. *Spine*, **2016**, 41, 856-63	3.3	40
172	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
171	Family satisfaction following spinal fusion in Rett syndrome. *Developmental Neurorehabilitation*, **2016**, 19, 31-7	1.8	6
170	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
169	Mitochondrial Aminoacyl-tRNA Synthetase Disorders Not Generally Affecting Brain **2016**, 243-249		
168	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

167	Sporadic and Familial Congenital Cataracts: Mutational Spectrum and New Diagnoses Using Next-Generation Sequencing. *Human Mutation*, **2016**, 37, 371-84	4.7	87
166	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
165	There is variability in the attainment of developmental milestones in the CDKL5 disorder. *Journal of Neurodevelopmental Disorders*, **2015**, 7, 2	4.6	47
164	Deletion of protein tyrosine phosphatase, non-receptor type 4 (PTPN4) in twins with a Rett syndrome-like phenotype. *European Journal of Human Genetics*, **2015**, 23, 1171-5	5.3	8
163	Pathogenicity of C-terminal mutations in CDKL5. *Journal of Pediatric Epilepsy*, **2015**, 01, 185-186	0.1	2
162	Genetic Metabolic Disease **2015**, 275-298		1
161	Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. *Human Molecular Genetics*, **2015**, 24, 6146-59	5.6	56
160	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
159	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
158	The Utility of Next-Generation Sequencing in Gene Discovery for Mutation-Negative Patients with Rett Syndrome. *Frontiers in Cellular Neuroscience*, **2015**, 9, 266	6.1	10
157	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
156	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. *Neuromuscular Disorders*, **2015**, 25, 257-61	2.9	9
155	Phenylketonuria: a review of current and future treatments. *Translational Pediatrics*, **2015**, 4, 304-17	4.2	85
154	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
153	Update on transcobalamin deficiency: clinical presentation, treatment and outcome. *Journal of Inherited Metabolic Disease*, **2014**, 37, 461-73	5.4	42
152	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
151	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
150	Inherited bone marrow failure associated with germline mutation of ACD, the gene encoding telomere protein TPP1. *Blood*, **2014**, 124, 2767-74	2.2	75

149	Experience of gastrostomy using a quality care framework: the example of rett syndrome. *Medicine (United States)*, **2014**, 93, e328	1.8	19
148	Rapid identification of a novel complex I MT-ND3 m.10134C>A mutation in a Leigh syndrome patient. *PLoS ONE*, **2014**, 9, e104879	3.7	5
147	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. *Brain*, **2014**, 137, 44-56	11.2	115
146	The Molecular Bases of Phenylketonuria (PKU) in New South Wales, Australia: Mutation Profile and Correlation with Tetrahydrobiopterin (BH4) Responsiveness. *JIMD Reports*, **2014**, 14, 55-65	1.9	10
145	Phenylketonuria: translating research into novel therapies. *Translational Pediatrics*, **2014**, 3, 49-62	4.2	14
144	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
143	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
142	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
141	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
140	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
139	A pilot study of the effect of (e, e)-2, 4-undecadienal on the offensive odour of trimethylamine. *JIMD Reports*, **2013**, 8, 11-5	1.9	3
138	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
137	Severe subacute necrotizing encephalopathy (Leigh-like syndrome) in American Staffordshire bull terrier dogs. *Journal of Comparative Pathology*, **2013**, 148, 345-53	1	7
136	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
135	Hearing loss and PRPS1 mutations: Wide spectrum of phenotypes and potential therapy. *International Journal of Audiology*, **2013**, 52, 23-8	2.6	22
134	Trimethylaminuria: an under-recognised and socially debilitating metabolic disorder. *Journal of Paediatrics and Child Health*, **2012**, 48, E153-5	1.3	22
133	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
132	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

131	Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing. *Science Translational Medicine*, **2012**, 4, 118ra10	17.5	362
130	The spectrum of mutations identified in Cypriot patients with phenylalanine hydroxylase deficiency detected through neonatal screening. *Clinical Biochemistry*, **2012**, 45, 588-92	3.5	4
129	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
128	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
127	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
126	Mitochondrial respiratory chain hepatopathies: role of liver transplantation. A case series of five patients. *JIMD Reports*, **2012**, 4, 5-11	1.9	15
125	Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial translation. *Cell Metabolism*, **2011**, 14, 428-34	24.6	123
124	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
123	The PRPP synthetase spectrum: what does it demonstrate about nucleotide syndromes?. *Nucleosides, Nucleotides and Nucleic Acids*, **2011**, 30, 1129-39	1.4	27
122	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
121	Genome-wide linkage of obstructive sleep apnoea and high-density lipoprotein cholesterol in a Filipino family: bivariate linkage analysis of obstructive sleep apnoea. *Journal of Sleep Research*, **2010**, 19, 349-57	5.8	11
120	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
119	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
118	Updating the profile of C-terminal MECP2 deletions in Rett syndrome. *Journal of Medical Genetics*, **2010**, 47, 242-8	5.8	51
117	Alleviating transcriptional inhibition of the norepinephrine slc6a2 transporter gene in depolarized neurons. *Journal of Neuroscience*, **2010**, 30, 1494-501	6.6	23
116	Pathophysiological mechanisms of dominant and recessive GLRA1 mutations in hyperekplexia. *Journal of Neuroscience*, **2010**, 30, 9612-20	6.6	102
115	PRPS1 mutations: four distinct syndromes and potential treatment. *American Journal of Human Genetics*, **2010**, 86, 506-18	11	74
114	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

113	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
112	Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, **2010**, 68, 944-50	9.4	804
111	The common BDNF polymorphism may be a modifier of disease severity in Rett syndrome. *Neurology*, **2009**, 72, 1242-7	6.5	59
110	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
109	Combined enzyme replacement and haematopoietic stem cell transplantation in Hurler syndrome. *Journal of Paediatrics and Child Health*, **2009**, 45, 469-72	1.3	11
108	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
107	Expanded newborn screening: outcome in screened and unscreened patients at age 6 years. *Pediatrics*, **2009**, 124, e241-8	7.4	117
106	Potential of AAV vectors in the treatment of metabolic disease. *Gene Therapy*, **2008**, 15, 831-9	4	67
105	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
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