Irenaeus F De Coo

List of Publications by Year
in descending order

97 papers	5,506 citations	71061 41 h-index	88593 70 g-index
99 all docs	99 docs citations	99 times ranked	8695 citing authors

#	Article	IF	Citations
1	Phenotypic spectrum and clinical course of single large-scale mitochondrial DNA deletion disease in the paediatric population: a multicentre study. Journal of Medical Genetics, 2023, 60, 65-73.	1.5	2
2	Neurodegenerative and functional signatures of the cerebellar cortex in m.3243Aâ€‰>â€‰G patients. Brain Communications, 2022, 4, fcac024.	1.5	2
3	Whole exome sequencing reveals a homozygous C1QBP deletion as the cause of progressive external ophthalmoplegia and multiple mtDNA deletions. Neuromuscular Disorders, 2021, 31, 859-864.	0.3	2
4	Merosin deficient congenital muscular dystrophy type 1A: An international workshop on the road to therapy 15-17 November 2019, Maastricht, the Netherlands. Neuromuscular Disorders, 2021, 31, 673-680.	0.3	2
5	Pathogenic SLIRP variants as a novel cause of autosomal recessive mitochondrial encephalomyopathy with complex I and IV deficiency. European Journal of Human Genetics, 2021, 29, 1789-1795.	1.4	7
6	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	5.5	50
7	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	1.7	11
8	Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with MCT8 deficiency: an international, single-arm, open-label, phase 2 trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 695-706.	5.5	77
9	A novel mitochondrial m.4414T>C MT-TM gene variant causing progressive external ophthalmoplegia and myopathy. Neuromuscular Disorders, 2019, 29, 693-697.	0.3	2
10	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	1.7	17
11	Autosomal recessive limbâ€girdle and Miyoshi muscular dystrophies in the Netherlands: The clinical and molecular spectrum of 244 patients. Clinical Genetics, 2019, 96, 126-133.	1.0	41
12	Healthy, mtDNA-mutationÂfree mesoangioblasts from mtDNA patients qualify for autologous therapy. Stem Cell Research and Therapy, 2019, 10, 405.	2.4	8
13	Preclinical Efficacy and Safety Evaluation of Hematopoietic Stem Cell Gene Therapy in a Mouse Model of MNGIE. Molecular Therapy - Methods and Clinical Development, 2018, 8, 152-165.	1.8	18
14	Anatomic & metabolic brain markers of the m.3243A>G mutation: A multi-parametric 7T MRI study. NeuroImage: Clinical, 2018, 18, 231-244.	1.4	15
15	Genetic defects in mtDNA-encoded protein translation cause pediatric, mitochondrial cardiomyopathy with early-onset brain disease. European Journal of Human Genetics, 2018, 26, 537-551.	1.4	23
16	Phenotype-genotype correlations in Leigh syndrome: new insights from a multicentre study of 96 patients. Journal of Medical Genetics, 2018, 55, 21-27.	1.5	54
17	Mutated zinc finger protein of the cerebellum 1 leads to microcephaly, cortical malformation, callosal agenesis, cerebellar dysplasia, tethered cord and scoliosis. European Journal of Medical Genetics, 2018, 61, 783-789.	0.7	10
18	Transplantation, gene therapy and intestinal pathology in MNGIE patients and mice. BMC Gastroenterology, 2018, 18, 149.	0.8	16

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19	Whole Exome Sequencing Is the Preferred Strategy to Identify the Genetic Defect in Patients With a Probable or Possible Mitochondrial Cause. Frontiers in Genetics, 2018, 9, 400.	1.1	66
20	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
21	Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing. Journal of Pediatrics, 2017, 182, 371-374.e2.	0.9	10
22	Dietary nitrate does not reduce oxygen cost of exercise or improve muscle mitochondrial function in patients with mitochondrial myopathy. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2017, 312, R689-R701.	0.9	8
23	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	1.5	54
24	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	2.8	65
25	Peripheral Neuropathy, Episodic Rhabdomyolysis, and Hypoparathyroidism in a Patient with Mitochondrial Trifunctional Protein Deficiency. JIMD Reports, 2017, 38, 101-105.	0.7	11
26	Mitochondrial Neurogastrointestinal Encephalomyopathy Caused by Thymidine Phosphorylase Enzyme Deficiency: From Pathogenesis to Emerging Therapeutic Options. Frontiers in Cellular Neuroscience, 2017, 11, 31.	1.8	42
27	Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect. Frontiers in Molecular Neuroscience, 2017, 10, 336.	1.4	11
28	Human mutations in integrator complex subunits link transcriptome integrity to brain development. PLoS Genetics, 2017, 13, e1006809.	1.5	66
29	International Consensus Statement on the Clinical and Therapeutic Management of Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2017, 37, 371-381.	0.4	156
30	Specific MRI Abnormalities Reveal Severe Perrault Syndrome due to CLPP Defects. Frontiers in Neurology, 2016, 7, 203.	1.1	25
31	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	4.2	224
32	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	1.7	79
33	Pathogenic CWF19L1 variants as a novel cause of autosomal recessive cerebellar ataxia and atrophy. European Journal of Human Genetics, 2016, 24, 619-622.	1.4	19
34	Lack of robust satellite cell activation and muscle regeneration during the progression of Pompe disease. Acta Neuropathologica Communications, 2015, 3, 65.	2.4	32
35	<scp>RYR</scp>1â€related myopathies: a wide spectrum of phenotypes throughout life. European Journal of Neurology, 2015, 22, 1094-1112.	1.7	111
36	Preventing the transmission of mitochondrial DNA disorders using prenatal or preimplantation genetic diagnosis. Annals of the New York Academy of Sciences, 2015, 1350, 29-36.	1.8	52

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37	The expanding phenotype of COL4A1 and COL4A2 mutations: clinical data on 13 newly identified families and a review of the literature. Genetics in Medicine, 2015, 17, 843-853.	1.1	204
38	Germline activating AKT3 mutation associated with megalencephaly, polymicrogyria, epilepsy and hypoglycemia. Molecular Genetics and Metabolism, 2015, 114, 467-473.	0.5	42
39	Selecting the Right Embryo in Mitochondrial Disorders. , 2015, , 231-246.		0
40	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	3.7	128
41	Compound heterozygous or homozygous truncating MYBPC3 mutations cause lethal cardiomyopathy with features of noncompaction and septal defects. European Journal of Human Genetics, 2015, 23, 922-928.	1.4	70
42	Leberâ€™s hereditary optic neuropathy with late disease onset: clinical and molecular characteristics of 20 patients. Orphanet Journal of Rare Diseases, 2014, 9, 158.	1.2	58
43	Severe presentation of WDR62 mutation: Is there a role for modifying genetic factors?. American Journal of Medical Genetics, Part A, 2014, 164, 2161-2171.	0.7	30
44	Reply: Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain, 2014, 137, e296-e296.	3.7	5
45	A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet Journal of Rare Diseases, 2014, 9, 52.	1.2	182
46	Mutations in DARS Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. American Journal of Human Genetics, 2013, 92, 774-780.	2.6	151
47	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	1.6	23
48	Clinical and mutational characteristics of spinal muscular atrophy with respiratory distress type 1 in the Netherlands. Neuromuscular Disorders, 2013, 23, 461-468.	0.3	15
49	Exome sequencing reveals a novel Moroccan founder mutation inSLC19A3as a new cause of early-childhood fatal Leigh syndrome. Brain, 2013, 136, 882-890.	3.7	81
50	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
51	COX19 mediates the transduction of a mitochondrial redox signal from SCO1 that regulates ATP7A-mediated cellular copper efflux. Molecular Biology of the Cell, 2013, 24, 683-691.	0.9	58
52	Novel noâ€stop FLNA mutation causes multiâ€organ involvement in males. American Journal of Medical Genetics, Part A, 2013, 161, 2376-2384.	0.7	33
53	ACTA2 mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. American Journal of Medical Genetics, Part A, 2013, 161, 1376-1380.	0.7	36
54	PRRT2 mutation causes benign familial infantile convulsions. Neurology, 2012, 79, 2154-2155.	1.5	22

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55	RTTN Mutations Link Primary Cilia Function to Organization of the Human Cerebral Cortex. American Journal of Human Genetics, 2012, 91, 533-540.	2.6	60
56	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. Brain, 2011, 134, 210-219.	3.7	113
57	Long-term follow-up of type 1 lissencephaly: survival is related to neuroimaging abnormalities. Developmental Medicine and Child Neurology, 2011, 53, 417-421.	1.1	15
58	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	2.6	166
59	Combined cardiological and neurological abnormalities due to filamin A gene mutation. Clinical Research in Cardiology, 2011, 100, 45-50.	1.5	43
60	Germline Mitochondrial DNA Mutations As a Novel First Event in Childhood Myelodysplastic Syndrome. Blood, 2011, 118, 1711-1711.	0.6	0
61	Patients with Leber hereditary optic neuropathy fail to compensate impaired oxidative phosphorylation. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 197-203.	0.5	19
62	Periventricular nodular heterotopia and distal limb deficiency: A recurrent association. American Journal of Medical Genetics, Part A, 2010, 152A, 954-959.	0.7	2
63	Unbalanced der(5)t(5;20) translocation associated with megalencephaly, perisylvian polymicrogyria, polydactyly and hydrocephalus. American Journal of Medical Genetics, Part A, 2010, 152A, 1488-1497.	0.7	6
64	The EEG response to pyridoxineâ€IV neither identifies nor excludes pyridoxineâ€dependent epilepsy. Epilepsia, 2010, 51, 2406-2411.	2.6	53
65	KBG syndrome associated with periventricular nodular heterotopia. Clinical Dysmorphology, 2010, 19, 164-165.	0.1	8
66	Nonsense mutations in CABC1/ADCK3 cause progressive cerebellar ataxia and atrophy. Mitochondrion, 2010, 10, 510-515.	1.6	66
67	The unfolding clinical spectrum of POLG mutations. Journal of Medical Genetics, 2009, 46, 776-785.	1.5	151
68	Mutation in the AP4M1 Gene Provides a Model for Neuroaxonal Injury in Cerebral Palsy. American Journal of Human Genetics, 2009, 85, 40-52.	2.6	156
69	Phenotypic consequences of a novel SCO2 gene mutation. American Journal of Medical Genetics, Part A, 2008, 146A, 2822-2827.	0.7	25
70	Extensive cerebral infarction in the newborn due to incontinentia pigmenti. European Journal of Paediatric Neurology, 2008, 12, 284-289.	0.7	39
71	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€“Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
72	Cardiac Involvement in Adults With m.3243A>G MELAS Gene Mutation. American Journal of Cardiology, 2007, 99, 264-269.	0.7	72

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73	Novel mutations in three families confirm a major role of COL4A1 in hereditary porencephaly. Journal of Medical Genetics, 2006, 43, 490-495.	1.5	145
74	Chip-based mtDNA mutation screening enables fast and reliable genetic diagnosis of OXPHOS patients. Genetics in Medicine, 2006, 8, 620-627.	1.1	32
75	Additional mitochondrial DNA mutations may explain extra-ocular involvement in LHON. American Journal of Medical Genetics, Part A, 2006, 140A, 1478-1481.	0.7	2
76	Re: Polymicrogyria versus pachygyria in 22q11 microdeletion. American Journal of Medical Genetics, Part A, 2005, 136A, 419-419.	0.7	1
77	Regional absence of mitochondria causing energy depletion in the myocardium of muscle LIM protein knockout mice. Cardiovascular Research, 2005, 65, 411-418.	1.8	43
78	Significance of Respirasomes for the Assembly/Stability of Human Respiratory Chain Complex I. Journal of Biological Chemistry, 2004, 279, 36349-36353.	1.6	287
79	Changes in Globus Pallidus With (Pre)Term Kernicterus. Pediatrics, 2003, 112, 1256-1263.	1.0	143
80	Benign familial infantile convulsions: a clinical study of seven Dutch families. European Journal of Paediatric Neurology, 2002, 6, 269-283.	0.7	34
81	An out-of-frame cytochromeb gene deletion from a patient with parkinsonism is associated with impaired complex III assembly and an increase in free radical production. Annals of Neurology, 2000, 48, 774-781.	2.8	126
82	Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population. European Journal of Human Genetics, 2000, 8, 557-560.	1.4	34
83	Human NDUFS3 gene coding for the 30-kDa subunit of mitochondrial Complex I: genomic organization and expression. Mammalian Genome, 2000, 11, 808-810.	1.0	9
84	Mutations in noncoding regions of the proteolipid protein gene in Pelizaeusâ€“Merzbacher disease. Neurology, 2000, 55, 1089-1096.	1.5	57
85	Childhood-onset cerebral X-linked adrenoleukodystrophy. Lancet, The, 2000, 356, 1608-1609.	6.3	2
86	The structure of the human NDUFV1 gene encoding the 51-kDa subunit of mitochondrial complex I 61, A305, 1997.-->. Mammalian Genome, 1999, 10, 49-53.	1.0	4
87	A 4-base pair deletion in the mitochondrial cytochromeb gene associated with parkinsonism/MELAS overlap syndrome. Annals of Neurology, 1999, 45, 130-133.	2.8	141
88	Nuclear DNA origin of mitochondrial complex I deficiency in fatal infantile lactic acidosis evidenced by transnuclear complementation of cultured fibroblasts. Journal of Clinical Investigation, 1999, 104, 83-92.	3.9	33
89	Ileus in mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes. Netherlands Journal of Medicine, 1998, 53, 27-31.	0.6	18
90	A PCR test for progressive external ophthalmoplegia and Kearns-Sayre syndrome on DNA from blood samples. Journal of the Neurological Sciences, 1997, 149, 37-40.	0.3	14

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91	Clinical heterogeneity in respiratory chain complex III deficiency in childhood. Journal of the Neurological Sciences, 1997, 149, 111-117.	0.3	62
92	Molecular Cloning and Characterization of the Human Mitochondrial NADH:Oxidoreductase 10-kDa Gene (NDUFV3). Genomics, 1997, 45, 434-437.	1.3	33
93	Magnetic resonance imaging in classification of congenital muscular dystrophies with brain abnormalities. Annals of Neurology, 1997, 42, 50-59.	2.8	141
94	Leber's hereditary optic neuropathy with the 11 778 mtDNA mutation and white matter disease resembling multiple sclerosis: clinical, MRI and MRS findings. Journal of the Neurological Sciences, 1996, 135, 176-180.	0.3	53
95	Molecular cloning and characterization of the active human mitochondrial NADH:ubiquinone oxidoreductase 24-kDa gene (NDUFV2) and its pseudogene. Genomics, 1995, 26, 461-466.	1.3	42
96	Recessively inherited â€˜pureâ€™ spastic paraplegia: Case study. Clinical Neurology and Neurosurgery, 1982, 84, 247-253.	0.6	3
97	Gastroenterology and Hepatology. , 0, , 163-177.		0