Charles M LourenÃ§o

List of Publications by Year
in descending order

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121 papers	7,593 citations	109137 35 h-index	58464 82 g-index
133 all docs	133 docs citations	133 times ranked	11890 citing authors

#	Article	IF	Citations
1	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	9.4	712
2	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	610
3	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	9.4	490
4	Assessment of interferon-related biomarkers in Aicardi-GoutiÃ¨res syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. Lancet Neurology, The, 2013, 12, 1159-1169.	4.9	473
5	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	0.7	447
6	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	13.9	390
7	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
8	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	1.5	262
9	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
10	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. Journal of Medical Genetics, 2015, 52, 514-522.	1.5	219
11	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	3.7	189
12	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
13	Mutations in the Mitochondrial Methionyl-tRNA Synthetase Cause a Neurodegenerative Phenotype in Flies and a Recessive Ataxia (ARSAL) in Humans. PLoS Biology, 2012, 10, e1001288.	2.6	147
14	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	0.8	119
15	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	2.8	116
16	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	1.4	115
17	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
18	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	2.6	104

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19	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
20	Twelve novel HGD gene variants identified in 99 alkaptonuria patients: focus on â€˜black bone diseaseâ€™ in Italy. European Journal of Human Genetics, 2016, 24, 66-72.	1.4	87
21	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
22	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
23	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
24	Plasma Lysosphingomyelin Demonstrates Great Potential as a Diagnostic Biomarker for Niemann-Pick Disease Type C in a Retrospective Study. PLoS ONE, 2014, 9, e114669.	1.1	75
25	Natural History of Vanishing White Matter. Annals of Neurology, 2018, 84, 274-288.	2.8	69
26	Expanding the genotypic spectrum of Perrault syndrome. Clinical Genetics, 2017, 91, 302-312.	1.0	68
27	Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction. American Journal of Human Genetics, 2015, 96, 765-774.	2.6	67
28	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
29	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	1.1	64
30	KIAA0586is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	1.1	62
31	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	2.6	55
32	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	13.7	53
33	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	5.5	50
34	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
35	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.	1.7	45
36	Musculoskeletal Disease in MDA5â€Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. Arthritis and Rheumatology, 2017, 69, 2081-2091.	2.9	44

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37	Determinants of white matter hyperintensity burden in patients with Fabry disease. Neurology, 2016, 86, 1880-1886.	1.5	42
38	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	1.1	39
39	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
40	Chromosomal microarray analysis in the genetic evaluation of 279 patients with syndromic obesity. Molecular Cytogenetics, 2018, 11, 14.	0.4	35
41	Detection of copy number variations in epilepsy using exome data. Clinical Genetics, 2018, 93, 577-587.	1.0	35
42	SPG11 mutations cause widespread white matter and basal ganglia abnormalities, but restricted cortical damage. NeuroImage: Clinical, 2018, 19, 848-857.	1.4	33
43	Epilepsy in mucopolysaccharidosis disorders. Molecular Genetics and Metabolism, 2017, 122, 55-61.	0.5	29
44	Clinical and neuroimaging features of autosomal recessive spastic paraplegia 35 (SPG35): case reports, new mutations, and brief literature review. Neurogenetics, 2018, 19, 123-130.	0.7	29
45	Clinical, Biomarker, and Molecular Delineations and Genotype-Phenotype Correlations of Ataxia With Oculomotor Apraxia Type 1. JAMA Neurology, 2018, 75, 495.	4.5	28
46	Mucolipidosis II and III alpha/beta in Brazil: Analysis of the GNPTAB gene. Gene, 2013, 524, 59-64.	1.0	27
47	Biomolecules damage and redox status abnormalities in Fabry patients before and during enzyme replacement therapy. Clinica Chimica Acta, 2016, 461, 41-46.	0.5	27
48	De novo <scp>DNM1</scp> mutations in two cases of epileptic encephalopathy. Epilepsia, 2016, 57, e18-23.	2.6	27
49	Neurodevelopmental disorders associated with dosage imbalance ofZBTB20correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	1.5	26
50	A Suspicion Index to aid screening of early-onset Niemann-Pick disease Type C (NP-C). BMC Pediatrics, 2016, 16, 107.	0.7	26
51	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
52	Analyses of disease-related GNPTAB mutations define a novel GlcNAc-1-phosphotransferase interaction domain and an alternative site-1 protease cleavage site. Human Molecular Genetics, 2015, 24, 3497-3505.	1.4	25
53	Application of N-palmitoyl-O-phosphocholineserine for diagnosis and assessment of response to treatment in Niemann-Pick type C disease. Molecular Genetics and Metabolism, 2020, 129, 292-302.	0.5	24
54	Mutations, Clinical Findings and Survival Estimates in South American Patients with X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e34195.	1.1	24

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55	Teratogenic effect of retinoic acid in swiss mice. Acta Cirurgica Brasileira, 2007, 22, 451-456.	0.3	23
56	New mutations in the ATM gene and clinical data of 25 AT patients. Neurogenetics, 2011, 12, 273-282.	0.7	23
57	Mutational screening of 320 Brazilian patients with autosomal dominant spinocerebellar ataxia. Journal of the Neurological Sciences, 2014, 347, 375-379.	0.3	23
58	New mutations in the GLA gene in Brazilian families with Fabry disease. Journal of Human Genetics, 2012, 57, 347-351.	1.1	22
59	SPG11â€related parkinsonism: Clinical profile, molecular imaging and <scp>l</scp>â€dopa response. Movement Disorders, 2018, 33, 1650-1656.	2.2	22
60	Analysis of the caregiver burden associated with Sanfilippo syndrome type B: panel recommendations based on qualitative and quantitative data. Orphanet Journal of Rare Diseases, 2019, 14, 168.	1.2	22
61	Characterizing the phenotypic manifestations of MFN2 R104W mutation in Charcotâ€“Marieâ€“Tooth type 2. Neuromuscular Disorders, 2011, 21, 428-432.	0.3	21
62	Clinical, ophthalmological, imaging and genetic features in Brazilian patients with ARSACS. Parkinsonism and Related Disorders, 2019, 62, 148-155.	1.1	21
63	New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592.	0.6	20
64	Enzyme replacement therapy for Mucopolysaccharidosis Type I among patients followed within the MPS Brazil Network. Genetics and Molecular Biology, 2014, 37, 23-29.	0.6	19
65	Biotinidase deficiency: Genotype-biochemical phenotype association in Brazilian patients. PLoS ONE, 2017, 12, e0177503.	1.1	19
66	Clinical and biochemical study of 29 Brazilian patients with metachromatic leukodystrophy. Journal of Inherited Metabolic Disease, 2010, 33, 257-262.	1.7	18
67	Mutations in the transloconâ€associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2019, 42, 993-997.	1.7	18
68	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. Orphanet Journal of Rare Diseases, 2021, 16, 185.	1.2	17
69	Coexistence of two chronic neuropathies in a young child: Charcotâ€“marieâ€“tooth disease type 1A and chronic inflammatory demyelinating polyneuropathy. Muscle and Nerve, 2010, 42, 598-600.	1.0	16
70	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. BMC Medical Genetics, 2014, 15, 96.	2.1	16
71	Revealing the clinical phenotype of atypical neuronal ceroid lipofuscinosis type 2 disease: Insights from the largest cohort in the world. Journal of Paediatrics and Child Health, 2021, 57, 519-525.	0.4	15
72	X-linked adrenoleukodystrophy in heterozygous female patients: women are not just carriers. Arquivos De Neuro-Psiquiatria, 2012, 70, 487-491.	0.3	13

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73	Respiratory dysfunction in Charcotâ€“Marieâ€“Tooth disease type 1A. Journal of Neurology, 2015, 262, 1164-1171.	1.8	13
74	GNPTAB missense mutations cause loss of GlcNAc-1-phosphotransferase activity in mucolipidosis type II through distinct mechanisms. International Journal of Biochemistry and Cell Biology, 2017, 92, 90-94.	1.2	11
75	Application of a glycinated bile acid biomarker for diagnosis and assessment of response to treatment in Niemann-pick disease type C1. Molecular Genetics and Metabolism, 2020, 131, 405-417.	0.5	11
76	Investigation of selected genomic deletions and duplications in a cohort of 338 patients presenting with syndromic obesity by multiplex ligation-dependent probe amplification using synthetic probes. Molecular Cytogenetics, 2014, 7, 75.	0.4	10
77	Psychiatric disorders, spinocerebellar ataxia type 3 and CAG expansion. Journal of Neurology, 2015, 262, 1777-1779.	1.8	10
78	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. JIMD Reports, 2019, 49, 96-106.	0.7	10
79	Enzyme replacement therapy interruption in patients with Mucopolysaccharidoses: Recommendations for distinct scenarios in Latin America. Molecular Genetics and Metabolism Reports, 2020, 23, 100572.	0.4	10
80	Expanding the differential diagnosis of inherited neuropathies with nonâ€uniform conduction: Andermann syndrome. Journal of the Peripheral Nervous System, 2012, 17, 123-127.	1.4	9
81	Quality of life in patients with Charcot-Marie-Tooth disease type 1A. Arquivos De Neuro-Psiquiatria, 2013, 71, 392-396.	0.3	9
82	TRAPÎ³-CDG shows asymmetric glycosylation and an effect on processing of proteins required in higher organisms. Journal of Medical Genetics, 2021, 58, 213-216.	1.5	9
83	Clinical Characterization of Mucolipidoses II and III: A Multicenter Study. Journal of Pediatric Genetics, 2019, 08, 198-204.	0.3	8
84	Novel clinical and genetic insight into CXorf56-associated intellectual disability. European Journal of Human Genetics, 2020, 28, 367-372.	1.4	8
85	Extension of the molecular analysis to the promoter region of the iduronate 2-sulfatase gene reveals genomic alterations in mucopolysaccharidosis type II patients with normal coding sequence. Gene, 2013, 526, 150-154.	1.0	7
86	Recommendations for Evaluation and Management of Pain in Patients With Mucopolysaccharidosis in Latin America. Journal of Pain and Symptom Management, 2018, 56, 146-152.	0.6	7
87	Maple syrup urine disease in Brazilian patients: variants and clinical phenotype heterogeneity. Orphanet Journal of Rare Diseases, 2020, 15, 309.	1.2	7
88	Retinal Architecture in Autosomal Recessive Spastic Ataxia of Charlevoixâ€Saguenay <scp>(ARSACS)</scp>: Insights into Disease Pathogenesis and Biomarkers. Movement Disorders, 2021, 36, 2027-2035.	2.2	7
89	Long-term impact of early initiation of enzyme replacement therapy in 34 MPS VI patients: A resurvey study. Molecular Genetics and Metabolism, 2021, 133, 94-99.	0.5	7
90	MECP2-related conditions in males: A systematic literature review and 8 additional cases. European Journal of Paediatric Neurology, 2021, 34, 7-13.	0.7	7

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91	Compound Charcot-Marie-Tooth disease may determine unusual and milder phenotypes. Neurogenetics, 2010, 11, 135-138.	0.7	6
92	Development of a Clinical Algorithm for the Early Diagnosis of Mucopolysaccharidosis III. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	6
93	GDAP1 mutations are frequent among Brazilian patients with autosomal recessive axonal Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2021, 31, 505-511.	0.3	5
94	Evaluation of galsulfase for the treatment of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Expert Opinion on Orphan Drugs, 2014, 2, 407-417.	0.5	4
95	Morquioâ€like dysostosis multiplex presenting with neuronopathic features is a distinct <scp>GLB1</scp>â€related phenotype. JIMD Reports, 2021, 60, 23-31.	0.7	4
96	New novel mutations in Brazilian families with Xâ€linked Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2019, 24, 207-212.	1.4	3
97	Evaluation of 3-O-methyldopa as a biomarker for aromatic L-amino acid decarboxylase deficiency in 7 Brazilian cases. Molecular Genetics and Metabolism Reports, 2021, 27, 100744.	0.4	3
98	Challenges in familial chylomicronemia syndrome diagnosis and management across Latin American countries: An expert panel discussion. Journal of Clinical Lipidology, 2021, 15, 620-624.	0.6	3
99	Transthyretin <scp>Asp38Tyr</scp>: a new mutation associated to a late onset neuropathy. Journal of the Peripheral Nervous System, 2015, 20, 60-62.	1.4	2
100	Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4.	0.5	2
101	Diagnosis and Management of Classical Homocystinuria in Brazil. FIRE Forum for International Research in Education, 2018, 6, 232640981878890.	0.7	2
102	Sanfilippo Syndrome: The Tale of a Challenging Diagnosis. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	2
103	Metachromatic leukodystrophy: pediatric presentation and the challenges of early diagnosis. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2020, 66, 1344-1350.	0.3	2
104	High glucose level as a modifier factor in CMT1A patients. Journal of the Peripheral Nervous System, 2020, 25, 132-137.	1.4	1
105	Sapropterin dihydrochloride therapy in dihydropteridine reductase deficiency: Insight from the first case with molecular diagnosis in Brazil. JIMD Reports, 2021, 61, 19-24.	0.7	1
106	MutaÃ§Ã£o missense de novo patogÃªnica c.2415C G (p.Asp805Glu) no gene ATP1A3 em paciente com hemiplegia alternante da infÃ¢ncia com resposta favorÃ¡vel ao cloridrato de biperideno. Medicina, 2021, 54, .	0.0	1
107	Acute hepatic porphyria: when to perform liver transplantation?. Medicina, 2021, 54, .	0.0	1
108	Disease duration and survival in Brazilian Niemann-Pick disease type C patients: Preliminary data on potential impact of miglustat. Molecular Genetics and Metabolism, 2016, 117, S50-S51.	0.5	0

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109	Pitfalls and potential clues in diagnosis in attenuated form of Hunter syndrome. Molecular Genetics and Metabolism, 2019, 126, S62-S63.	0.5	0
110	Against all odds: enzyme replacement therapy in non-ambulatory and ambulatory Morquio syndrome type A patients. Molecular Genetics and Metabolism, 2019, 126, S32.	0.5	0
111	Clinical and biochemical study of Brazilian patients with metachromatic leukodystrophy. Molecular Genetics and Metabolism, 2019, 126, S42-S43.	0.5	0
112	Hurler syndrome: Severe sleep apnea as initial presentation in a 10-month-old child. Molecular Genetics and Metabolism, 2019, 126, S61.	0.5	0
113	ATAXIA DE FRIEDREICH: RELATO DE CASO DE IRMÃƒOS COM FENÃ“TIPOS CLÃNICOS DISCORDANTES. , 0, , 362-372.		0
114	Chudleyâ€“McCullough Syndrome: Case Report and the Role of Neuroimaging to Suggest the Diagnosis. Journal of Pediatric Neurology, 0, , .	0.0	0
115	HipoventilaÃ§Ã£o relacionada ao sono de origem central secundÃ¡ria Ã deficiÃªncia de biotinidase: relato de caso. Medicina, 2021, 54, e166390.	0.0	0
116	SÃNDROME DE CANTÃš: O PRIMEIRO BRASILEIRO COM MUTAÃ‡ÃƒO CONFIRMADA DO GENE ABCC9. , 2019, , .		0
117	A INFLUÃŠNCIA DO HIV EM INFECÃ‡Ã•ES OPORTUNISTAS DO SISTEMA NERVOSO CENTRAL. , 2019, , .		0
118	Perthes-Like Disease Masquerading Non-Classical MPS. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	0
119	Uso de canabidiol como terapia adjuvante em paciente com sÃndrome de Zellweger: relato de caso. Medicina, 2020, 53, 321-326.	0.0	0
120	Paraparesia espÃ¡stica complicada como fenÃ³tipo neurolÃ³gico em OPA1. Medicina, 2021, 54, .	0.0	0
121	Desafios do diagnÃ³stico da hipofosfatasia em adultos. Medicina, 2022, 55, .	0.0	0