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	Desafios do diagnÃ³stico da hipofosfatasia em adultos. Medicina, 2022, 55, .	0.0	0
2	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
3	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
4	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	13.7	53
5	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
6	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. Orphanet Journal of Rare Diseases, 2021, 16, 185.	1.2	17
7	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
8	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
9	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
10	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
11	GDAP1 mutations are frequent among Brazilian patients with autosomal recessive axonal Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2021, 31, 505-511.	0.3	5
12	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
13	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
14	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
15	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
16	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
17	Acute hepatic porphyria: when to perform liver transplantation?. Medicina, 2021, 54, .	0.0	1
18	Paraparesia espÃ¡stica complicada como fenÃ³tipo neurolÃ³gico em OPA1. Medicina, 2021, 54, .	0.0	0

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19	Novel clinical and genetic insight into CXorf56-associated intellectual disability. European Journal of Human Genetics, 2020, 28, 367-372.	1.4	8
20	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	5.5	50
21	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
22	Maple syrup urine disease in Brazilian patients: variants and clinical phenotype heterogeneity. Orphanet Journal of Rare Diseases, 2020, 15, 309.	1.2	7
23	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
24	High glucose level as a modifier factor in CMT1A patients. Journal of the Peripheral Nervous System, 2020, 25, 132-137.	1.4	1
25	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
26	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
27	Uso de canabidiol como terapia adjuvante em paciente com sÃndrome de Zellweger: relato de caso. Medicina, 2020, 53, 321-326.	0.0	0
28	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. JIMD Reports, 2019, 49, 96-106.	0.7	10
29	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
30	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
31	Pitfalls and potential clues in diagnosis in attenuated form of Hunter syndrome. Molecular Genetics and Metabolism, 2019, 126, S62-S63.	0.5	0
32	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
33	Clinical and biochemical study of Brazilian patients with metachromatic leukodystrophy. Molecular Genetics and Metabolism, 2019, 126, S42-S43.	0.5	0
34	Hurler syndrome: Severe sleep apnea as initial presentation in a 10-month-old child. Molecular Genetics and Metabolism, 2019, 126, S61.	0.5	0
35	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
36	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	1.1	39

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37	Clinical Characterization of Mucolipidoses II and III: A Multicenter Study. Journal of Pediatric Genetics, 2019, 08, 198-204.	0.3	8
38	Clinical, ophthalmological, imaging and genetic features in Brazilian patients with ARSACS. Parkinsonism and Related Disorders, 2019, 62, 148-155.	1.1	21
39	SÃNDROME DE CANTÃš: O PRIMEIRO BRASILEIRO COM MUTAÃ‡ÃƒO CONFIRMADA DO GENE ABCC9. , 2019, , .		0
40	A INFLUÃŠNCIA DO HIV EM INFECÃ‡Ã•ES OPORTUNISTAS DO SISTEMA NERVOSO CENTRAL. , 2019, , .		0
41	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
42	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
43	Clinical, Biomarker, and Molecular Delineations and Genotype-Phenotype Correlations of Ataxia With Oculomotor Apraxia Type 1. JAMA Neurology, 2018, 75, 495.	4.5	28
44	Chromosomal microarray analysis in the genetic evaluation of 279 patients with syndromic obesity. Molecular Cytogenetics, 2018, 11, 14.	0.4	35
45	Detection of copy number variations in epilepsy using exome data. Clinical Genetics, 2018, 93, 577-587.	1.0	35
46	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	1.1	64
47	SPG11â€related parkinsonism: Clinical profile, molecular imaging and <scp>l</scp>â€dopa response. Movement Disorders, 2018, 33, 1650-1656.	2.2	22
48	SPG11 mutations cause widespread white matter and basal ganglia abnormalities, but restricted cortical damage. NeuroImage: Clinical, 2018, 19, 848-857.	1.4	33
49	Diagnosis and Management of Classical Homocystinuria in Brazil. FIRE Forum for International Research in Education, 2018, 6, 232640981878890.	0.7	2
50	Natural History of Vanishing White Matter. Annals of Neurology, 2018, 84, 274-288.	2.8	69
51	Expanding the genotypic spectrum of Perrault syndrome. Clinical Genetics, 2017, 91, 302-312.	1.0	68
52	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
53	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
54	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

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55	Epilepsy in mucopolysaccharidosis disorders. Molecular Genetics and Metabolism, 2017, 122, 55-61.	0.5	29
56	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
57	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	0.8	119
58	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
59	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
60	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
61	Biotinidase deficiency: Genotype-biochemical phenotype association in Brazilian patients. PLoS ONE, 2017, 12, e0177503.	1.1	19
62	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	2.8	116
63	A Suspicion Index to aid screening of early-onset Niemann-Pick disease Type C (NP-C). BMC Pediatrics, 2016, 16, 107.	0.7	26
64	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
65	Determinants of white matter hyperintensity burden in patients with Fabry disease. Neurology, 2016, 86, 1880-1886.	1.5	42
66	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	13.9	390
67	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
68	De novo <scp>DNM1</scp> mutations in two cases of epileptic encephalopathy. Epilepsia, 2016, 57, e18-23.	2.6	27
69	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
70	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
71	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
72	KIAA0586is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	1.1	62

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73	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
74	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
75	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
76	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. Journal of Medical Genetics, 2015, 52, 514-522.	1.5	219
77	Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4.	0.5	2
78	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
79	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
80	Respiratory dysfunction in Charcotâ€“Marieâ€“Tooth disease type 1A. Journal of Neurology, 2015, 262, 1164-1171.	1.8	13
81	Psychiatric disorders, spinocerebellar ataxia type 3 and CAG expansion. Journal of Neurology, 2015, 262, 1777-1779.	1.8	10
82	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
83	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	1.4	115
84	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
85	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
86	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
87	Mutational screening of 320 Brazilian patients with autosomal dominant spinocerebellar ataxia. Journal of the Neurological Sciences, 2014, 347, 375-379.	0.3	23
88	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
89	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
90	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

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91	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	3.7	189
92	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
93	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. BMC Medical Genetics, 2014, 15, 96.	2.1	16
94	New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592.	0.6	20
95	Evaluation of galsulfase for the treatment of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Expert Opinion on Orphan Drugs, 2014, 2, 407-417.	0.5	4
96	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
97	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
98	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
99	Mucolipidosis II and III alpha/beta in Brazil: Analysis of the GNPTAB gene. Gene, 2013, 524, 59-64.	1.0	27
100	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
101	Quality of life in patients with Charcot-Marie-Tooth disease type 1A. Arquivos De Neuro-Psiquiatria, 2013, 71, 392-396.	0.3	9
102	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
103	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	9.4	712
104	X-linked adrenoleukodystrophy in heterozygous female patients: women are not just carriers. Arquivos De Neuro-Psiquiatria, 2012, 70, 487-491.	0.3	13
105	New mutations in the GLA gene in Brazilian families with Fabry disease. Journal of Human Genetics, 2012, 57, 347-351.	1.1	22
106	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
107	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
108	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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109	Characterizing the phenotypic manifestations of MFN2 R104W mutation in Charcotâ€“Marieâ€“Tooth type 2. Neuromuscular Disorders, 2011, 21, 428-432.	0.3	21
110	New mutations in the ATM gene and clinical data of 25 AT patients. Neurogenetics, 2011, 12, 273-282.	0.7	23
111	Compound Charcot-Marie-Tooth disease may determine unusual and milder phenotypes. Neurogenetics, 2010, 11, 135-138.	0.7	6
112	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
113	Clinical and biochemical study of 29 Brazilian patients with metachromatic leukodystrophy. Journal of Inherited Metabolic Disease, 2010, 33, 257-262.	1.7	18
114	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
115	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
116	Teratogenic effect of retinoic acid in swiss mice. Acta Cirurgica Brasileira, 2007, 22, 451-456.	0.3	23
117	ATAXIA DE FRIEDREICH: RELATO DE CASO DE IRMÃƒOS COM FENÃ“TIPOS CLÃNICOS DISCORDANTES. , 0, , 362-372.		0
118	Chudleyâ€“McCullough Syndrome: Case Report and the Role of Neuroimaging to Suggest the Diagnosis. Journal of Pediatric Neurology, 0, , .	0.0	0
119	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
120	Perthes-Like Disease Masquerading Non-Classical MPS. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	0
121	Sanfilippo Syndrome: The Tale of a Challenging Diagnosis. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	2