Patrick Scott

List of Publications by Year
in descending order

23 papers	397 citations	840776 11 h-index	752698 20 g-index
23 all docs	23 docs citations	23 times ranked	659 citing authors

#	Article	IF	Citations
1	A novel POC1A variant in an alternatively spliced exon causes classic SOFT syndrome: clinical presentation of seven patients. Journal of Human Genetics, 2020, 65, 193-197.	2.3	7
2	Underdiagnoses resulting from variant misinterpretation: Time for systematic reanalysis of whole exome data?. European Journal of Medical Genetics, 2019, 62, 39-43.	1.3	22
3	Inborn errors of metabolism in a cohort of pregnancies with non-immune hydrops fetalis: a single center experience. Journal of Perinatal Medicine, 2018, 46, 968-974.	1.4	8
4	Exome Sequencing Identifies a Novel Sorting Nexin 14 Gene Mutation Causing Cerebellar Atrophy and Intellectual Disability. Case Reports in Genetics, 2018, 2018, 1-3.	0.2	3
5	Co-inheritance of the membrane frizzled-related protein ocular phenotype and glycogen storage disease type Ib. Ophthalmic Genetics, 2017, 38, 544-548.	1.2	7
6	Segmental Spinal Muscular Atrophy Localised to the Lower Limbs: First case from Oman. Sultan Qaboos University Medical Journal, 2017, 17, e355-357.	1.0	2
7	Occurrence of Optic Neuritis and Cervical Cord Schwannoma with Charcot-Marie-Tooth Type 4B1 Disease. Oman Medical Journal, 2016, 31, 227-230.	1.0	16
8	Familial amyotrophic lateral sclerosis in Alberta, Canada. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 273-277.	1.7	10
9	Report on the p.Ser489X (p.Ser489*) CFTR mutation, a variant with severe associated phenotype and high prevalence in a Quebec French-Canadian cystic fibrosis patient population. Genetics in Medicine, 2012, 14, 883-886.	2.4	5
10	Toward Optimal Detection of the Common Prenatal Aneuploidies by Quantitative Fluorescentâ€“Polymerase Chain Reaction: Comparison of Two Commercial Assays. Genetic Testing and Molecular Biomarkers, 2012, 16, 943-947.	0.7	1
11	Defining the Role of Laboratory Genetic Counselor. Journal of Genetic Counseling, 2012, 21, 605-611.	1.6	26
12	Endothelial ultrastructural alterations of intramuscular capillaries in infantile mitochondrial cytopathies: â€œMitochondrial angiopathyâ€• Neuropathology, 2012, 32, 617-627.	1.2	20
13	A second ALS patient having an L67P mutation in exon 3 of the Cu/Zn superoxide dismutase gene. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 466-467.	2.1	2
14	Newborn screening for cystic fibrosis in Alberta: Two years of experience. Paediatrics and Child Health, 2010, 15, 590-594.	0.6	15
15	An intronic mutation in DKC1 in an infant with HÃ¸yraalâ€“Hreidarsson syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2159-2161.	1.2	21
16	Frequency and phenotypic consequences of the 3199del6 CFTR mutation in French Canadians. Genetics in Medicine, 2005, 7, 210-211.	2.4	1
17	Prenatal diagnosis for severe methylenetetrahydrofolate reductase deficiency by linkage analysis and enzymatic assay. Molecular Genetics and Metabolism, 2005, 85, 115-120.	1.1	12
18	Prenatal diagnosis for methylmalonic acidemia and inborn errors of vitamin B12 metabolism and transport. Molecular Genetics and Metabolism, 2005, 86, 160-171.	1.1	41

#	Article	IF	Citations
19	Neonatal hyperphenylalaninemia, perinatal hemochromatosis, and renal tubulopathy: A unique patient or a novel metabolic disorder?. Molecular Genetics and Metabolism, 2005, 86, 148-152.	1.1	4
20	Evaluation of the calcium-sensing receptor gene in idiopathic hypercalciuria and calcium nephrolithiasis. Kidney International, 2000, 58, 38-42.	5.2	51
21	Suggestive Evidence for a Susceptibility Gene Near the Vitamin D Receptor Locus in Idiopathic Calcium Stone Formation. Journal of the American Society of Nephrology: JASN, 1999, 10, 1007-1013.	6.1	92
22	Cytogenetic Analysis of a Parachordoma. Cancer Genetics and Cytogenetics, 1998, 105, 14-19.	1.0	25
23	DNA replication asynchrony between the paternal and maternal alleles of imprinted genes does not straddle the R/G transition. Chromosoma, 1997, 106, 405-411.	2.2	6