Simon Olpin

List of Publications by Year in Descending Order

18 papers	2,157 citations	11 h-index	18 g-index
18 ext. papers	2,454 ext. citations	6.2 avg, IF	3.05 L-index

#	Paper	IF	Citations
18	Novel mutations associated with carnitine-acylcarnitine translocase and carnitine palmitoyl transferase 2 deficiencies in Malaysia. *Clinical Biochemistry*, **2021**, 98, 48-53	3.5	0
17	Biochemical assessment of patients following ketogenic diets for epilepsy: Current practice in the UK and Ireland. *Epilepsia Open*, **2020**, 5, 73-79	4	3
16	Fibroblast Fatty-Acid Oxidation Flux Assays Stratify Risk in Newborns with Presumptive-Positive Results on Screening for Very-Long Chain Acyl-CoA Dehydrogenase Deficiency. *International Journal of Neonatal Screening*, **2017**, 3, 2	2.6	5
15	Mitochondrial trifunctional protein deficiency in human cultured fibroblasts: effects of bezafibrate. *Journal of Inherited Metabolic Disease*, **2016**, 39, 47-58	5.4	24
14	Transient 5-oxoprolinuria: unusually high anion gap acidosis in an infant. *European Journal of Pediatrics*, **2015**, 174, 1685-8	4.1	1
13	Gene expression signatures in motor neurone disease fibroblasts reveal dysregulation of metabolism, hypoxia-response and RNA processing functions. *Neuropathology and Applied Neurobiology*, **2015**, 41, 201-26	5.2	45
12	Niemann-Pick type C: a potentially treatable disorder?. *Practical Neurology*, **2013**, 13, 382-5	2.4	10
11	MCAD deficiency in Denmark. *Molecular Genetics and Metabolism*, **2012**, 106, 175-88	3.7	26
10	Hypertrophic pyloric stenosis: predicting the resolution of biochemical abnormalities. *Pediatric Surgery International*, **2011**, 27, 695-8	2.1	8
9	Dysregulation of hypoxia pathways in fumarate hydratase-deficient cells is independent of defective mitochondrial metabolism. *Human Molecular Genetics*, **2010**, 19, 3844-51	5.6	84
8	Investigation and functional characterization of rare genetic variants in the adipose triglyceride lipase in a large healthy working population. *PLoS Genetics*, **2010**, 6, e1001239	6	34
7	Failure to repair the c.338C>T mutation in carnitine palmitoyl transferase 2 deficient skin fibroblasts using chimeraplasty. *Molecular Genetics and Metabolism*, **2008**, 93, 347-9	3.7	1
6	Mitochondrial function and morphology are impaired in parkin-mutant fibroblasts. *Annals of Neurology*, **2008**, 64, 555-65	9.4	280
5	Fumarase deficiency caused by homozygous P131R mutation and paternal partial isodisomy of chromosome 1. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1004-9	2.5	17
4	Missense mutations in fumarate hydratase in multiple cutaneous and uterine leiomyomatosis and renal cell cancer. *Journal of Molecular Diagnostics*, **2005**, 7, 437-43	5.1	42
3	Clinical features of multiple cutaneous and uterine leiomyomatosis: an underdiagnosed tumor syndrome. *Archives of Dermatology*, **2005**, 141, 199-206		138
2	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. *Nature Genetics*, **2002**, 30, 406-10	36.3	1191

1	Clear correlation of genotype with disease phenotype in very-long-chain acyl-CoA dehydrogenase deficiency. *American Journal of Human Genetics*, **1999**, 64, 479-94	11	248