Timothy Barrett

List of Publications by Citations

100 papers	4,792 citations	34 h-index	68 g-index
105 ext. papers	5,500 ext. citations	7.2 avg, IF	5.04 L-index

#	Paper	IF	Citations
100	EIF2AK3, encoding translation initiation factor 2-alpha kinase 3, is mutated in patients with Wolcott-Rallison syndrome. *Nature Genetics*, **2000**, 25, 406-9	36.3	635
99	Association between mutations in a thyroid hormone transporter and severe X-linked psychomotor retardation. *Lancet, The*, **2004**, 364, 1435-7	40	517
98	Macrosomia and hyperinsulinaemic hypoglycaemia in patients with heterozygous mutations in the HNF4A gene. *PLoS Medicine*, **2007**, 4, e118	11.6	279
97	Mutations in SLC19A2 cause thiamine-responsive megaloblastic anaemia associated with diabetes mellitus and deafness. *Nature Genetics*, **1999**, 22, 300-4	36.3	216
96	Rising incidence of type 2 diabetes in children in the U.K. *Diabetes Care*, **2007**, 30, 1097-101	14.6	174
95	Clinical and molecular genetic analysis of 19 Wolfram syndrome kindreds demonstrating a wide spectrum of mutations in WFS1. *American Journal of Human Genetics*, **1999**, 65, 1279-90	11	164
94	Wolcott-Rallison Syndrome: clinical, genetic, and functional study of EIF2AK3 mutations and suggestion of genetic heterogeneity. *Diabetes*, **2004**, 53, 1876-83	0.9	143
93	ISPAD Clinical Practice Consensus Guidelines 2014. Type 2 diabetes in the child and adolescent. *Pediatric Diabetes*, **2014**, 15 Suppl 20, 26-46	3.6	128
92	Ethnic differences in insulin resistance and body composition in United Kingdom adolescents. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 3963-9	5.6	120
91	WFS1/wolframin mutations, Wolfram syndrome, and associated diseases. *Human Mutation*, **2001**, 17, 357-67	4.7	119
90	Dysregulation of autophagy as a common mechanism in lysosomal storage diseases. *Essays in Biochemistry*, **2017**, 61, 733-749	7.6	107
89	Liraglutide in Children and Adolescents with Type 2 Diabetes. *New England Journal of Medicine*, **2019**, 381, 637-646	59.2	105
88	Premature adrenarche: novel lessons from early onset androgen excess. *European Journal of Endocrinology*, **2011**, 165, 189-207	6.5	94
87	Effect of Oral Insulin on Prevention of Diabetes in Relatives of Patients With Type 1 Diabetes: A Randomized Clinical Trial. *JAMA - Journal of the American Medical Association*, **2017**, 318, 1891-1902	27.4	88
86	Association studies of genetic variation in the WFS1 gene and type 2 diabetes in U.K. populations. *Diabetes*, **2002**, 51, 1287-90	0.9	79
85	Localization of the gene for thiamine-responsive megaloblastic anemia syndrome, on the long arm of chromosome 1, by homozygosity mapping. *American Journal of Human Genetics*, **1997**, 61, 1335-41	11	78
84	Prevalence of abnormal lipid profiles and the relationship with the development of microalbuminuria in adolescents with type 1 diabetes. *Diabetes Care*, **2009**, 32, 658-63	14.6	74

83	Mutation analysis of the WFS1 gene in seven Danish Wolfram syndrome families; four new mutations identified. *European Journal of Human Genetics*, **2005**, 13, 1275-84	5.3	68
82	Functional analysis of monocarboxylate transporter 8 mutations identified in patients with X-linked psychomotor retardation and elevated serum triiodothyronine. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 2378-81	5.6	66
81	Sodium-potassium ATPase 1 subunit is a molecular partner of Wolframin, an endoplasmic reticulum protein involved in ER stress. *Human Molecular Genetics*, **2008**, 17, 190-200	5.6	63
80	Optic atrophy in Wolfram (DIDMOAD) syndrome. *Eye*, **1997**, 11 (Pt 6), 882-8	4.4	60
79	Heritability of body mass index in pre-adolescence, young adulthood and late adulthood. *European Journal of Epidemiology*, **2012**, 27, 247-53	12.1	58
78	Thiamine-responsive megaloblastic anaemia syndrome: long-term follow-up and mutation analysis of seven families. *Acta Paediatrica, International Journal of Paediatrics*, **2006**, 95, 99-104	3.1	51
77	Coach autonomy support predicts autonomous motivation and daily moderate-to-vigorous physical activity and sedentary time in youth sport participants. *Psychology of Sport and Exercise*, **2014**, 15, 453-463	4.2	47
76	The emergence of type 2 diabetes in childhood. *Annals of Clinical Biochemistry*, **2004**, 41, 10-6	2.2	44
75	Risk Factors for Severe Renal Disease in Bardet-Biedl Syndrome. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 963-970	12.7	41
74	Sartorial eloquence: does it exist in the paediatrician-patient relationship?. *BMJ: British Medical Journal*, **1994**, 309, 1710-2		41
73	Differential diagnosis of type 1 diabetes: which genetic syndromes need to be considered?. *Pediatric Diabetes*, **2007**, 8 Suppl 6, 15-23	3.6	40
72	Thyroid dyshormonogenesis is mainly caused by TPO mutations in consanguineous community. *Clinical Endocrinology*, **2013**, 79, 275-81	3.4	38
71	Vacuolar-type H+-ATPase V1A subunit is a molecular partner of Wolfram syndrome 1 (WFS1) protein, which regulates its expression and stability. *Human Molecular Genetics*, **2013**, 22, 203-17	5.6	37
70	Microstructural abnormalities in white and gray matter in obese adolescents with and without type 2 diabetes. *NeuroImage: Clinical*, **2017**, 16, 43-51	5.3	37
69	EURO-WABB: an EU rare diseases registry for Wolfram syndrome, Alstrm syndrome and Bardet-Biedl syndrome. *BMC Pediatrics*, **2013**, 13, 130	2.6	36
68	Wolfram syndrome. *Reviews in Endocrine and Metabolic Disorders*, **2003**, 4, 53-9	10.5	35
67	Physical activity and blood pressure in primary school children: a longitudinal study. *Hypertension*, **2013**, 61, 70-5	8.5	34
66	Sarco(endo)plasmic reticulum ATPase is a molecular partner of Wolfram syndrome 1 protein, which negatively regulates its expression. *Human Molecular Genetics*, **2015**, 24, 814-27	5.6	32

65	Gastrointestinal symptoms in children with type 1 diabetes screened for celiac disease. *Pediatrics*, **2009**, 124, e489-95	7.4	31
64	Monogenic diabetes syndromes: Locus-specific databases for Alstrm, Wolfram, and Thiamine-responsive megaloblastic anemia. *Human Mutation*, **2017**, 38, 764-777	4.7	30
63	Bipolar disorder and variation at a common polymorphism (A1832G) within exon 8 of the Wolfram gene. *American Journal of Medical Genetics Part A*, **2000**, 96, 154-7		27
62	A cluster-randomised controlled trial to assess the effectiveness and cost-effectiveness of a childhood obesity prevention programme delivered through schools, targeting 6-7 year old children: the WAVES study protocol. *BMC Public Health*, **2015**, 15, 488	4.1	26
61	A novel albumin gene mutation (R222I) in familial dysalbuminemic hyperthyroxinemia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1381-6	5.6	26
60	Assessment of childhood obesity in secondary care: OSCA consensus statement. *Archives of Disease in Childhood: Education and Practice Edition*, **2012**, 97, 98-105	0.5	26
59	TSHR is the main causative locus in autosomal recessively inherited thyroid dysgenesis. *Journal of Pediatric Endocrinology and Metabolism*, **2012**, 25, 419-26	1.6	26
58	Novel TSHR mutations in consanguineous families with congenital nongoitrous hypothyroidism. *Clinical Endocrinology*, **2010**, 73, 671-7	3.4	25
57	Homozygous loss-of-function mutations in SLC26A7 cause goitrous congenital hypothyroidism. *JCI Insight*, **2018**, 3,	9.9	25
56	Steroid Sulfatase Deficiency and Androgen Activation Before and After Puberty. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2545-53	5.6	25
55	Empowering youth sport environments: Implications for daily moderate-to-vigorous physical activity and adiposity. *Journal of Sport and Health Science*, **2017**, 6, 423-433	8.2	23
54	Optimising physical activity engagement during youth sport: a self-determination theory approach. *Journal of Sports Sciences*, **2016**, 34, 1874-84	3.6	23
53	High quality, patient centred and coordinated care for Alstrom syndrome: a model of care for an ultra-rare disease. *Orphanet Journal of Rare Diseases*, **2015**, 10, 149	4.2	23
52	Novel truncating thyroglobulin gene mutations associated with congenital hypothyroidism. *Endocrine*, **2014**, 45, 206-12	4	22
51	Home urine C-peptide creatinine ratio (UCPCR) testing can identify type 2 and MODY in pediatric diabetes. *Pediatric Diabetes*, **2013**, 14, 181-8	3.6	22
50	Preventing childhood obesity, phase II feasibility study focusing on South Asians: BEACHeS. *BMJ Open*, **2014**, 4, e004579	3	21
49	Clinical utility gene card for: Alstrm Syndrome - update 2013. *European Journal of Human Genetics*, **2013**, 21,	5.3	21
48	IGFALS gene dosage effects on serum IGF-I and glucose metabolism, body composition, bone growth in length and width, and the pharmacokinetics of recombinant human IGF-I administration. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E703-12	5.6	20

47	Comparison of the clinical scoring systems in Silver-Russell syndrome and development of modified diagnostic criteria to guide molecular genetic testing. *Journal of Medical Genetics*, **2013**, 50, 635-9	5.8	19
46	Consensus clinical management guidelines for Alstrm syndrome. *Orphanet Journal of Rare Diseases*, **2020**, 15, 253	4.2	19
45	Common variations in the ALMS1 gene do not contribute to susceptibility to type 2 diabetes in a large white UK population. *Diabetologia*, **2006**, 49, 1209-13	10.3	17
44	A truncating DUOX2 mutation (R434X) causes severe congenital hypothyroidism. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 323-7	1.6	15
43	Maternal but not paternal association of ambulatory blood pressure with albumin excretion in young offspring with type 1 diabetes. *Diabetes Care*, **2010**, 33, 366-71	14.6	15
42	Hearing impairment in genotyped Wolfram syndrome patients. *Annals of Otology, Rhinology and Laryngology*, **2008**, 117, 494-500	2.1	15
41	Design and validation of a metabolic disorder resequencing microarray (BRUM1). *Human Mutation*, **2010**, 31, 858-65	4.7	14
40	The changing face and implications of childhood obesity. *New England Journal of Medicine*, **2004**, 350, 2414-6; author reply 2414-6	59.2	14
39	Rapid progression of type 2 diabetes and related complications in children and young people-A literature review. *Pediatric Diabetes*, **2020**, 21, 158-172	3.6	14
38	Refining genotype-phenotype correlation in Alstrm syndrome through study of primary human fibroblasts. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 390-404	2.3	12
37	Clinical utility gene card for: Alstrm syndrome. *European Journal of Human Genetics*, **2011**, 19,	5.3	11
36	Evaluation of human dermal fibroblasts directly reprogrammed to adipocyte-like cells as a metabolic disease model. *DMM Disease Models and Mechanisms*, **2017**, 10, 1411-1420	4.1	11
35	Causes, patterns and severity of androgen excess in 487 consecutively recruited pre- and post-pubertal children. *European Journal of Endocrinology*, **2019**, 180, 213-221	6.5	11
34	Targets and teamwork: Understanding differences in pediatric diabetes centers treatment outcomes. *Pediatric Diabetes*, **2018**, 19, 559-565	3.6	11
33	Inter-participant variability in daily physical activity and sedentary time among male youth sport footballers: independent associations with indicators of adiposity and cardiorespiratory fitness. *Journal of Sports Sciences*, **2016**, 34, 239-51	3.6	10
32	Autophagy in Rare (NonLysosomal) Neurodegenerative Diseases. *Journal of Molecular Biology*, **2020**, 432, 2735-2753	6.5	10
31	Trajectories and predictors of developmental skills in healthy twins up to 24 months of age. *Research in Social and Administrative Pharmacy*, **2013**, 36, 670-8	2.9	10
30	Duration of Diabetes Predicts Aortic Pulse Wave Velocity and Vascular Events in Alstrm Syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1116-24	5.6	10

29	A nonsense thyrotropin receptor gene mutation (R609X) is associated with congenital hypothyroidism and heart defects. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 1101-5	1.6	10
28	One Base Deletion (c.2422delT) in the TPO Gene Causes Severe Congenital Hypothyroidism. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2014**, 6, 169-73	1.9	10
27	DIDMOAD syndrome; further studies and muscle biochemistry. *Journal of Inherited Metabolic Disease*, **1995**, 18, 218-20	5.4	10
26	The West Midlands ActiVe lifestyle and healthy Eating in School children (WAVES) study: a cluster randomised controlled trial testing the clinical effectiveness and cost-effectiveness of a multifaceted obesity prevention intervention programme targeted at children aged 6-7 years. *Health Technology Assessment*, **2018**, 22, 1-608	4.4	10
25	A deletion including exon 2 of the TSHR gene is associated with thyroid dysgenesis and severe congenital hypothyroidism. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 731-5	1.6	9
24	Wolcott-Rallison syndrome: a clinical and genetic study of three children, novel mutation in EIF2AK3 and a review of the literature **2004**, 93, 1195		9
23	Defining renal phenotype in Alstrm syndrome. *Nephrology Dialysis Transplantation*, **2020**, 35, 994-1001	4.3	9
22	Is arterial stiffening in Alstrm syndrome linked to the development of cardiomyopathy?. *European Journal of Clinical Investigation*, **2007**, 37, 99-105	4.6	8
21	Development of an assessment tool for screening children for glucose intolerance by oral glucose tolerance test. *Diabetes Care*, **2004**, 27, 280-1	14.6	8
20	Bladder dysfunction in Wolfram syndrome is highly prevalent and progresses to megacystis. *Journal of Pediatric Surgery*, **2018**, 53, 321-325	2.6	7
19	Human Induced Pluripotent Stem Cell Models of Neurodegenerative Disorders for Studying the Biomedical Implications of Autophagy. *Journal of Molecular Biology*, **2020**, 432, 2754-2798	6.5	7
18	The Contribution of Youth Sport Football to Weekend Physical Activity for Males Aged 9 to 16 Years: Variability Related to Age and Playing Position. *Pediatric Exercise Science*, **2015**, 27, 208-18	2	6
17	A truncating TPO mutation (Y55X) in patients with hypothyroidism and total iodide organification defect. *Endocrine Research*, **2015**, 40, 146-50	1.9	6
16	An essential splice site mutation (c.317+1G>A) in the TSHR gene leads to severe thyroid dysgenesis. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 1021-5	1.6	6
15	Identification of homozygous WFS1 mutations (p.Asp211Asn, p.Gln486*) causing severe Wolfram syndrome and first report of male fertility. *European Journal of Human Genetics*, **2013**, 21, 347-51	5.3	6
14	Thiamine-responsive megaloblastic anaemia syndrome: Long-term follow-up and mutation analysis of seven families. *Acta Paediatrica, International Journal of Paediatrics*, **2007**, 95, 99-104	3.1	6
13	Relationship between Parental Feeding Practices and Neural Responses to Food Cues in Adolescents. *PLoS ONE*, **2016**, 11, e0157037	3.7	6
12	Treatment adherence and BMI reduction are key predictors of HbA1c 1 year after diagnosis of childhood type 2 diabetes in the United Kingdom. *Pediatric Diabetes*, **2018**, 19, 1393-1399	3.6	6

List of Publications

11	The characterisation of the human Wolfram syndrome gene promoter demonstrating regulation by Sp1 and Sp3 transcription factors. *Biochimica Et Biophysica Acta Gene Regulatory Mechanisms*, **2006**, 1759, 367-77		5
10	A cost of illness study evaluating the burden of Wolfram syndrome in the United Kingdom. *Orphanet Journal of Rare Diseases*, **2019**, 14, 185	4.2	3
9	Adjuvant rituximab, a potential treatment for the young patient with Graves Phyperthyroidism (RiGD): study protocol for a single-arm, single-stage, phase II trial. *BMJ Open*, **2019**, 9, e024705	3	3
8	Adiposity and response to an obesity prevention intervention in Pakistani and Bangladeshi primary school boys and girls: a secondary analysis using the BEACHeS feasibility study. *BMJ Open*, **2016**, 6, e007907	3	3
7	WFS1 protein expression correlates with clinical progression of optic atrophy in patients with Wolfram syndrome. *Journal of Medical Genetics*, **2021**,	5.8	3
6	DYNAMIC: Dynamic glucose management strategies delivered through a structured education program improves time in range in a socioeconomically deprived cohort of children and young people with type 1 diabetes with a history of hypoglycemia. *Pediatric Diabetes*, **2021**, 22, 249-260	3.6	2
5	The Birmingham Registry for Twin and Heritability Studies (BiRTHS). *Twin Research and Human Genetics*, **2006**, 9, 907-912	2.2	1
4	General population screening for childhood type 1 diabetes: is it time for a UK strategy?. *Archives of Disease in Childhood*, **2021**,	2.2	1
3	Clinical characteristics and treatment requirements of children with autosomal recessive pseudohypoaldosteronism. *European Journal of Endocrinology*, **2021**, 184, K15-K20	6.5	1
2	WFS1/wolframin mutations, Wolfram syndrome, and associated diseases **2001**, 17, 357		1
1	Advanced course in paediatric and adolescent diabetes. *Practical Diabetes International: the International Journal for Diabetes Care Teams Worldwide*, **2003**, 20, 256-256		