

Robert K Semple

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

164 papers	11,045 citations	53 h-index	103 g-index
202 ext. papers	12,807 ext. citations	9 avg, IF	5.74 L-index

#	Paper	IF	Citations
164	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015 , 526, 82-90	50.4	776
163	TAC3 and TACR3 mutations in familial hypogonadotropic hypogonadism reveal a key role for Neurokinin B in the central control of reproduction. <i>Nature Genetics</i> , 2009 , 41, 354-358	36.3	685
162	PPAR gamma and human metabolic disease. <i>Journal of Clinical Investigation</i> , 2006 , 116, 581-9	15.9	612
161	Modeling inherited metabolic disorders of the liver using human induced pluripotent stem cells. <i>Journal of Clinical Investigation</i> , 2010 , 120, 3127-36	15.9	457
160	Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45,891 individuals. <i>PLoS Genetics</i> , 2012 , 8, e1002607	6	326
159	Mutations in the pericentrin (PCNT) gene cause primordial dwarfism. <i>Science</i> , 2008 , 319, 816-9	33.3	325
158	Integrative genomic analysis implicates limited peripheral adipose storage capacity in the pathogenesis of human insulin resistance. <i>Nature Genetics</i> , 2017 , 49, 17-26	36.3	312
157	Association of a homozygous nonsense caveolin-1 mutation with Berardinelli-Seip congenital lipodystrophy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008 , 93, 1129-34	5.6	302
156	PIK3CA-related overgrowth spectrum (PROS): diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 287-95	2.5	275
155	Genetic variation near IRS1 associates with reduced adiposity and an impaired metabolic profile. <i>Nature Genetics</i> , 2011 , 43, 753-60	36.3	237
154	Two novel missense mutations in g protein-coupled receptor 54 in a patient with hypogonadotropic hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005 , 90, 1849-55	5.6	236
153	Mutations in the selenocysteine insertion sequence-binding protein 2 gene lead to a multisystem selenoprotein deficiency disorder in humans. <i>Journal of Clinical Investigation</i> , 2010 , 120, 4220-35	15.9	229
152	Mosaic overgrowth with fibroadipose hyperplasia is caused by somatic activating mutations in PIK3CA. <i>Nature Genetics</i> , 2012 , 44, 928-33	36.3	221
151	Genetic syndromes of severe insulin resistance. <i>Endocrine Reviews</i> , 2011 , 32, 498-514	27.2	212
150	Postreceptor insulin resistance contributes to human dyslipidemia and hepatic steatosis. <i>Journal of Clinical Investigation</i> , 2009 , 119, 315-22	15.9	210
149	Perilipin deficiency and autosomal dominant partial lipodystrophy. <i>New England Journal of Medicine</i> , 2011 , 364, 740-8	59.2	201
148	Clinical delineation and natural history of the PIK3CA-related overgrowth spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1713-33	2.5	198

147	Partial lipodystrophy and insulin resistant diabetes in a patient with a homozygous nonsense mutation in CIDEA. <i>EMBO Molecular Medicine</i> , 2009 , 1, 280-7	12	195
146	Serotonin 2C receptor agonists improve type 2 diabetes via melanocortin-4 receptor signaling pathways. <i>Cell Metabolism</i> , 2007 , 6, 398-405	24.6	170
145	The human lipodystrophy gene BSCL2/seipin may be essential for normal adipocyte differentiation. <i>Diabetes</i> , 2008 , 57, 2055-60	0.9	161
144	Expression of the thermogenic nuclear hormone receptor coactivator PGC-1alpha is reduced in the adipose tissue of morbidly obese subjects. <i>International Journal of Obesity</i> , 2004 , 28, 176-9	5.5	152
143	Non-DNA binding, dominant-negative, human PPARGgamma mutations cause lipodystrophic insulin resistance. <i>Cell Metabolism</i> , 2006 , 4, 303-11	24.6	143
142	Genetic evidence for a normal-weight "metabolically obese" phenotype linking insulin resistance, hypertension, coronary artery disease, and type 2 diabetes. <i>Diabetes</i> , 2014 , 63, 4369-77	0.9	131
141	A genome-wide association study reveals variants in ARL15 that influence adiponectin levels. <i>PLoS Genetics</i> , 2009 , 5, e1000768	6	129
140	An activating mutation of AKT2 and human hypoglycemia. <i>Science</i> , 2011 , 334, 474	33.3	129
139	Common genetic variants highlight the role of insulin resistance and body fat distribution in type 2 diabetes, independent of obesity. <i>Diabetes</i> , 2014 , 63, 4378-4387	0.9	127
138	Autoimmune forms of hypoglycemia. <i>Medicine (United States)</i> , 2009 , 88, 141-153	1.8	122
137	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. <i>Nature Genetics</i> , 2013 , 45, 947-50	36.3	120
136	Elevated plasma adiponectin in humans with genetically defective insulin receptors. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006 , 91, 3219-23	5.6	117
135	Cancer-Associated PIK3CA Mutations in Overgrowth Disorders. <i>Trends in Molecular Medicine</i> , 2018 , 24, 856-870	11.5	112
134	Hypogonadotropic hypogonadism due to a novel missense mutation in the first extracellular loop of the neurokinin B receptor. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009 , 94, 3633-3639	5.6	110
133	Mosaic RAS/MAPK variants cause sporadic vascular malformations which respond to targeted therapy. <i>Journal of Clinical Investigation</i> , 2018 , 128, 1496-1508	15.9	97
132	Evidence of a Causal Association Between Insulinemia and Endometrial Cancer: A Mendelian Randomization Analysis. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	96
131	Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and type 2 diabetes. <i>Diabetes</i> , 2013 , 62, 3589-98	0.9	95
130	Somatic activating mutations in Pik3ca cause sporadic venous malformations in mice and humans. <i>Science Translational Medicine</i> , 2016 , 8, 332ra43	17.5	92

129	Hypoadiponectinemia--cause or consequence of human "insulin resistance"? <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010 , 95, 1544-54	5.6	92
128	Mutations disrupting the Kennedy phosphatidylcholine pathway in humans with congenital lipodystrophy and fatty liver disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 8901-6	11.5	88
127	AKR1C3-Mediated Adipose Androgen Generation Drives Lipotoxicity in Women With Polycystic Ovary Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 3327-3339	5.6	87
126	The recent genetics of hypogonadotrophic hypogonadism - novel insights and new questions. <i>Clinical Endocrinology</i> , 2010 , 72, 427-35	3.4	85
125	Paradoxical elevation of high-molecular weight adiponectin in acquired extreme insulin resistance due to insulin receptor antibodies. <i>Diabetes</i> , 2007 , 56, 1712-7	0.9	82
124	Mitochondrial dysfunction in patients with primary congenital insulin resistance. <i>Journal of Clinical Investigation</i> , 2011 , 121, 2457-61	15.9	78
123	Recent insights into fatty liver, metabolic dyslipidaemia and their links to insulin resistance. <i>Current Opinion in Lipidology</i> , 2010 , 21, 329-36	4.4	75
122	Plasma adiponectin as a marker of insulin receptor dysfunction: clinical utility in severe insulin resistance. <i>Diabetes Care</i> , 2008 , 31, 977-9	14.6	75
121	A truncation mutation in TBC1D4 in a family with acanthosis nigricans and postprandial hyperinsulinemia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009 , 106, 9350-5	11.5	73
120	ETO/MTG8 is an inhibitor of C/EBPbeta activity and a regulator of early adipogenesis. <i>Molecular and Cellular Biology</i> , 2004 , 24, 9863-72	4.8	68
119	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. <i>Genetics in Medicine</i> , 2019 , 21, 1189-1198	8.1	67
118	Monoallelic and biallelic mutations in MAB21L2 cause a spectrum of major eye malformations. <i>American Journal of Human Genetics</i> , 2014 , 94, 915-23	11	64
117	Complement abnormalities in acquired lipodystrophy revisited. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009 , 94, 10-6	5.6	64
116	IGF-I treatment of insulin resistance. <i>European Journal of Endocrinology</i> , 2007 , 157 Suppl 1, S51-6	6.5	64
115	Treatment of type B insulin resistance: a novel approach to reduce insulin receptor autoantibodies. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010 , 95, 3641-7	5.6	63
114	Hypomorphism in human NSMCE2 linked to primordial dwarfism and insulin resistance. <i>Journal of Clinical Investigation</i> , 2014 , 124, 4028-38	15.9	63
113	Whole-genome sequence-based analysis of thyroid function. <i>Nature Communications</i> , 2015 , 6, 5681	17.4	56
112	The effects of neurokinin B upon gonadotrophin release in male rodents. <i>Journal of Neuroendocrinology</i> , 2010 , 22, 181-7	3.8	54

111	Analysis of genetic variation in Akt2/PKB-beta in severe insulin resistance, lipodystrophy, type 2 diabetes, and related metabolic phenotypes. <i>Diabetes</i> , 2007 , 56, 714-9	0.9	50
110	Next generation sequencing--implications for clinical practice. <i>British Medical Bulletin</i> , 2011 , 99, 53-71	5.4	45
109	Genetic defects in human pericentrin are associated with severe insulin resistance and diabetes. <i>Diabetes</i> , 2011 , 60, 925-35	0.9	44
108	Treatment with recombinant human insulin-like growth factor (rhIGF)-I/rhIGF binding protein-3 complex improves metabolic control in subjects with severe insulin resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010 , 95, 2113-22	5.6	43
107	Human biallelic MFN2 mutations induce mitochondrial dysfunction, upper body adipose hyperplasia, and suppression of leptin expression. <i>ELife</i> , 2017 , 6,	8.9	42
106	Nephroblastomatosis or Wilms tumor in a fourth patient with a somatic PIK3CA mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2559-69	2.5	41
105	Evaluating the role of LPIN1 variation in insulin resistance, body weight, and human lipodystrophy in U.K. Populations. <i>Diabetes</i> , 2008 , 57, 2527-33	0.9	40
104	DNA Polymerase Epsilon Deficiency Causes IMAGE Syndrome with Variable Immunodeficiency. <i>American Journal of Human Genetics</i> , 2018 , 103, 1038-1044	11	39
103	Oncogenic PIK3CA induces centrosome amplification and tolerance to genome doubling. <i>Nature Communications</i> , 2017 , 8, 1773	17.4	38
102	Vps34 PI 3-kinase inactivation enhances insulin sensitivity through reprogramming of mitochondrial metabolism. <i>Nature Communications</i> , 2017 , 8, 1804	17.4	37
101	Mechanistic insights into insulin resistance in the genetic era. <i>Diabetic Medicine</i> , 2011 , 28, 1476-86	3.5	35
100	Monogenic polycystic ovary syndrome due to a mutation in the lamin A/C gene is sensitive to thiazolidinediones but not to metformin. <i>European Journal of Endocrinology</i> , 2008 , 159, 347-53	6.5	35
99	What is the best management strategy for patients with severe insulin resistance?. <i>Clinical Endocrinology</i> , 2010 , 73, 286-90	3.4	34
98	EJE PRIZE 2015: How does insulin resistance arise, and how does it cause disease? Human genetic lessons. <i>European Journal of Endocrinology</i> , 2016 , 174, R209-23	6.5	33
97	Managing clinically significant findings in research: the UK10K example. <i>European Journal of Human Genetics</i> , 2014 , 22, 1100-4	5.3	33
96	Severe insulin resistance due to anti-insulin antibodies: response to plasma exchange and immunosuppressive therapy. <i>Diabetic Medicine</i> , 2009 , 26, 79-82	3.5	32
95	Oncogenic promotes cellular stemness in an allele dose-dependent manner. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 , 116, 8380-8389	11.5	31
94	Genetics in endocrinology: genetic forms of severe insulin resistance: what endocrinologists should know. <i>European Journal of Endocrinology</i> , 2013 , 169, R71-80	6.5	31

93	Monogenic diabetes syndromes: Locus-specific databases for Alström, Wolfram, and Thiamine-responsive megaloblastic anemia. <i>Human Mutation</i> , 2017 , 38, 764-777	4.7	30
92	Neurokinin B signalling in the human reproductive axis. <i>Molecular and Cellular Endocrinology</i> , 2011 , 346, 57-64	4.4	30
91	Insulin resistance uncoupled from dyslipidemia due to C-terminal PIK3R1 mutations. <i>JCI Insight</i> , 2016 , 1, e88766	9.9	30
90	Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. <i>European Journal of Human Genetics</i> , 2020 , 28, 1387-1393	5.3	27
89	ENDOCRINOLOGY IN THE TIME OF COVID-19: Remodelling diabetes services and emerging innovation. <i>European Journal of Endocrinology</i> , 2020 , 183, G67-G77	6.5	27
88	Adiponectin receptor genes: mutation screening in syndromes of insulin resistance and association studies for type 2 diabetes and metabolic traits in UK populations. <i>Diabetologia</i> , 2007 , 50, 555-62	10.3	26
87	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. <i>European Journal of Endocrinology</i> , 2017 , 177, 175-186	6.5	24
86	Severe hypo-alpha-lipoproteinemia during treatment with rosiglitazone. <i>Diabetes Care</i> , 2004 , 27, 2577-80	4.6	24
85	Successful treatment of type B insulin resistance with rituximab. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, 1719-22	5.6	22
84	Severe insulin resistance and hypertriglyceridemia after childhood total body irradiation. <i>Endocrine Practice</i> , 2013 , 19, 51-8	3.2	22
83	A clinical approach to severe insulin resistance. <i>Endocrine Development</i> , 2007 , 11, 122-132		22
82	A Pharmacogenetic Approach to the Treatment of Patients With Mutations. <i>Diabetes</i> , 2018 , 67, 1086-1092	2.9	21
81	Mechanistic insights revealed by lipid profiling in monogenic insulin resistance syndromes. <i>Genome Medicine</i> , 2015 , 7, 63	14.4	21
80	Knockdown of the Alström syndrome-associated gene <i>Alms1</i> in 3T3-L1 preadipocytes impairs adipogenesis but has no effect on cell-autonomous insulin action. <i>International Journal of Obesity</i> , 2010 , 34, 1554-8	5.5	21
79	Growth and hormone profiling in children with congenital melanocytic naevi. <i>British Journal of Dermatology</i> , 2015 , 173, 1471-8	4	20
78	Metabolic insights from extreme human insulin resistance phenotypes. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2012 , 26, 145-57	6.5	20
77	Advanced non-alcoholic fatty liver disease and adipose tissue fibrosis in patients with Alström syndrome. <i>Liver International</i> , 2016 , 36, 1704-1712	7.9	20
76	Bi-allelic Loss-of-Function <i>CACNA1B</i> Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019 , 104, 948-956	11	17

75	Truncation of POC1A associated with short stature and extreme insulin resistance. <i>Journal of Molecular Endocrinology</i> , 2015 , 55, 147-58	4.5	16
74	Diagnosis of insulin autoimmune syndrome using polyethylene glycol precipitation and gel filtration chromatography with ex vivo insulin exchange. <i>Clinical Endocrinology</i> , 2017 , 86, 347-353	3.4	16
73	Adiponectin and leptin in human severe insulin resistance - diagnostic utility and biological insights. <i>Biochimie</i> , 2012 , 94, 2172-9	4.6	16
72	Phosphoinositide 3-kinase-related overgrowth: cellular phenotype and future therapeutic options. <i>Lancet, The</i> , 2015 , 385 Suppl 1, S77	4.0	15
71	Familial adult onset hyperinsulinism due to an activating glucokinase mutation: implications for pharmacological glucokinase activation. <i>Clinical Endocrinology</i> , 2014 , 81, 855-61	3.4	15
70	A novel mutation of the insulin receptor gene in a preterm infant with Donohue syndrome and heart failure. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2012 , 25, 363-6	1.6	15
69	The metabolic syndrome- associated small G protein ARL15 plays a role in adipocyte differentiation and adiponectin secretion. <i>Scientific Reports</i> , 2017 , 7, 17593	4.9	14
68	Continuous subcutaneous IGF-1 therapy via insulin pump in a patient with Donohue syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014 , 27, 1237-41	1.6	14
67	Insulin Receptor and the Kidney: Nephrocalcinosis in Patients with Recessive INSR Mutations. <i>Nephron Physiology</i> , 2014 , 128, 55-61		13
66	Type B Insulin Resistance Masquerading as Ovarian Hyperthecosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 1789-1791	5.6	13
65	Assessment and Management of Anti-Insulin Autoantibodies in Varying Presentations of Insulin Autoimmune Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018 , 103, 3845-3855	5.6	13
64	Combined Immunosuppressive Therapy Induces Remission in Patients With Severe Type B Insulin Resistance: A Prospective Cohort Study. <i>Diabetes Care</i> , 2018 , 41, 2353-2360	14.6	13
63	Refining genotype-phenotype correlation in Alström syndrome through study of primary human fibroblasts. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 390-404	2.3	12
62	"Donohue syndrome". <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012 , 97, 1416-7	5.6	12
61	Clinical experience with the AKT1 inhibitor miransertib in two children with PIK3CA-related overgrowth syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 109	4.2	12
60	Somatic mosaicism of the PIK3CA gene identified in a Hungarian girl with macrodactyly and syndactyly. <i>European Journal of Medical Genetics</i> , 2016 , 59, 223-6	2.6	11
59	Evaluation of human dermal fibroblasts directly reprogrammed to adipocyte-like cells as a metabolic disease model. <i>DMM Disease Models and Mechanisms</i> , 2017 , 10, 1411-1420	4.1	11
58	How Useful Are Monogenic Rodent Models for the Study of Human Non-Alcoholic Fatty Liver Disease?. <i>Frontiers in Endocrinology</i> , 2016 , 7, 145	5.7	11

57	A genome-wide RNAi screen identifies the SMC5/6 complex as a non-redundant regulator of a Topo2a-dependent G2 arrest. <i>Nucleic Acids Research</i> , 2019 , 47, 2906-2921	20.1	11
56	Constitutive Activation of AKT2 in Humans Leads to Hypoglycemia Without Fatty Liver or Metabolic Dyslipidemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 2914-2921	5.6	10
55	From bending DNA to diabetes: the curious case of HMGA1. <i>Journal of Biology</i> , 2009 , 8, 64		10
54	Analysis of TBC1D4 in patients with severe insulin resistance. <i>Diabetologia</i> , 2010 , 53, 1239-42	10.3	10
53	Resolution of Hypoglycemia and Cardiovascular Dysfunction After Rituximab Treatment of Insulin Autoimmune Syndrome. <i>Diabetes Care</i> , 2017 , 40, e80-e82	14.6	9
52	H syndrome with a novel homozygous R134C mutation in SLC29A3 gene. <i>International Journal of Dermatology</i> , 2013 , 52, 820-3	1.7	9
51	Functional characterization of a novel insulin receptor mutation contributing to Rabson-Mendenhall syndrome. <i>Clinical Endocrinology</i> , 2007 , 66, 21-6	3.4	9
50	Successful rhIGF1 treatment for over 5 years in a patient with severe insulin resistance due to homozygous insulin receptor mutation. <i>Diabetic Medicine</i> , 2016 , 33, e8-e12	3.5	9
49	Acute knockdown of the insulin receptor or its substrates Irs1 and 2 in 3T3-L1 adipocytes suppresses adiponectin production. <i>Scientific Reports</i> , 2016 , 6, 21105	4.9	8
48	Roux-en-Y Gastric Bypass Surgery in the Management of Familial Partial Lipodystrophy Type 1. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 3616-3620	5.6	8
47	Early Diagnosis of Werner Syndrome Using Exome-Wide Sequencing in a Single, Atypical Patient. <i>Frontiers in Endocrinology</i> , 2011 , 2, 8	5.7	8
46	Founder effect in the Horn of Africa for an insulin receptor mutation that may impair receptor recycling. <i>Diabetologia</i> , 2011 , 54, 1057-65	10.3	8
45	A type III complement factor D deficiency: Structural insights for inhibition of the alternative pathway. <i>Journal of Allergy and Clinical Immunology</i> , 2018 , 142, 311-314.e6	11.5	7
44	Secretome Analysis of Hypoxia-Induced 3T3-L1 Adipocytes Uncovers Novel Proteins Potentially Involved in Obesity. <i>Proteomics</i> , 2018 , 18, e1700260	4.8	7
43	Rabson-Mendenhall syndrome with recurrent cerebral infarcts caused by a novel INSR mutation. <i>International Journal of Dermatology</i> , 2013 , 52, 182-5	1.7	7
42	Severe insulin resistance in disguise: A familial case of reactive hypoglycemia associated with a novel heterozygous INSR mutation. <i>Pediatric Diabetes</i> , 2018 , 19, 670-674	3.6	6
41	Syndromes of Severe Insulin Resistance and/or Lipodystrophy 2010 , 39-52		6
40	IRS2 variants and syndromes of severe insulin resistance. <i>Diabetologia</i> , 2009 , 52, 1208-11	10.3	6

39	Caenorhabditis elegans DAF-2 as a Model for Human Insulin Receptoropathies. <i>G3: Genes, Genomes, Genetics</i> , 2017 , 7, 257-268	3.2	5
38	Anti-Insulin Receptor Antibodies Improve Hyperglycemia in a Mouse Model of Human Insulin Receptoropathy. <i>Diabetes</i> , 2020 , 69, 2481-2489	0.9	5
37	Somatic PIK3R1 variation as a cause of vascular malformations and overgrowth. <i>Genetics in Medicine</i> , 2021 , 23, 1882-1888	8.1	5
36	Intractable Hypoglycemia in the Setting of Autoimmune Overlap Syndrome. <i>Pediatrics</i> , 2017 , 139,	7.4	4
35	Gastrointestinal dysmotility and pancreatic insufficiency in 2 siblings with Donohue syndrome. <i>Pediatric Diabetes</i> , 2017 , 18, 839-843	3.6	4
34	Evaluation of anti-insulin receptor antibodies as potential novel therapies for human insulin receptoropathy using cell culture models. <i>Diabetologia</i> , 2018 , 61, 1662-1675	10.3	4
33	NODAL/TGFβ signalling mediates the self-sustained stemness induced by homozygosity in pluripotent stem cells. <i>DMM Disease Models and Mechanisms</i> , 2021 ,	4.1	4
32	Ovarian Hyperandrogenism and Response to Gonadotropin-releasing Hormone Analogues in Primary Severe Insulin Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, 2367-2383	5.6	4
31	Syndromes of Severe Insulin Resistance and/or Lipodystrophy 2016 , 307-324		4
30	Neurokinin B and its receptor in hypogonadotropic hypogonadism. <i>Frontiers of Hormone Research</i> , 2010 , 39, 133-141	3.5	3
29	Truncation of Pik3r1 causes severe insulin resistance uncoupled from obesity and dyslipidaemia by increased energy expenditure. <i>Molecular Metabolism</i> , 2020 , 40, 101020	8.8	3
28	A standard of care for individuals with PIK3CA-related disorders: An international expert consensus statement. <i>Clinical Genetics</i> , 2022 , 101, 32-47	4	3
27	Genome-Wide Association Study of NAFLD Using Electronic Health Records. <i>Hepatology Communications</i> , 2021 ,	6	3
26	Sex hormone-binding globulin and risk of type 2 diabetes. <i>New England Journal of Medicine</i> , 2009 , 361, 2677; author reply 2677-8	59.2	3
25	Syndromes Associated with Mutations in the Insulin Signalling Pathway. <i>Frontiers in Diabetes</i> , 2017 , 104-118	1.1	2
24	Novel mutation in insulin receptor gene identified after muscle biopsy in a Niuean woman with severe insulin resistance. <i>Diabetic Medicine</i> , 2015 , 32, e24-8	3.5	2
23	Human pluripotent stem cell-based models suggest preadipocyte senescence as a possible cause of metabolic complications of Werner and Bloom Syndromes. <i>Scientific Reports</i> , 2020 , 10, 7490	4.9	2
22	Luminescent peptide tagging enables efficient screening for CRISPR-mediated knock-in in human induced pluripotent stem cells. <i>Wellcome Open Research</i> , 2019 , 4, 37	4.8	2

21	Type B insulin resistance syndrome associated with connective tissue disease and psoriasis. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2020 , 2020,	1.4	2
20	Luminescent peptide tagging enables efficient screening for CRISPR-mediated knock-in in human induced pluripotent stem cells. <i>Wellcome Open Research</i> , 2019 , 4, 37	4.8	2
19	Transcriptomically-inferred PI3K activity and stemness show a counterintuitive correlation with PIK3CA genotype in breast cancer		2
18	NODAL/TGFβ signalling mediates the self-sustained stemness induced by PIK3CAH1047R homozygosity in pluripotent stem cells		2
17	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. <i>Genetics in Medicine</i> , 2021 , 23, 1484-1491	8.1	2
16	Genetic Disorders of Insulin Action: Far More than Diabetes. <i>Current Obesity Reports</i> , 2013 , 2, 293-300	8.4	1
15	Loss of NPC1 function in a patient with a co-inherited novel insulin receptor mutation does not grossly modify the severity of the associated insulin resistance. <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33 Suppl 3, S227-32	5.4	1
14	Severe hypoinsulinaemic hypoglycaemia in a premature infant associated with poor weight gain and reduced adipose tissue. <i>Hormone Research in Paediatrics</i> , 2007 , 68, 91-8	3.3	1
13	Luminescent peptide tagging enables efficient screening for CRISPR-mediated knock-in in human induced pluripotent stem cells. <i>Wellcome Open Research</i> , 4, 37	4.8	1
12	Oncogenic PIK3CA promotes cellular stemness in an allele dose-dependent manner		1
11	Unusual Glycemic Presentations in a Child with a Novel Heterozygous Intragenic INSR Deletion. <i>Hormone Research in Paediatrics</i> , 2020 , 93, 396-401	3.3	1
10	Identification of rare loss of function genetic variation regulating body fat distribution. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 ,	5.6	1
9	Atypical neurofibromatosis type 1 with unilateral limb hypertrophy mimicking overgrowth syndrome. <i>Clinical and Experimental Dermatology</i> , 2017 , 42, 763-766	1.8	0
8	Positive correlation between transcriptomic stemness and PI3K/AKT/mTOR signaling scores in breast cancer, and a counterintuitive relationship with PIK3CA genotype. <i>PLoS Genetics</i> , 2021 , 17, e1009876	6.7	0
7	Increased C-Peptide Immunoreactivity in Insulin Autoimmune Syndrome (Hirata Disease) Due to High Molecular Weight Proinsulin. <i>Clinical Chemistry</i> , 2021 , 67, 854-862	5.5	0
6	Insulin Resistance and Diabetes Associated with Lipodystrophies. <i>Frontiers in Diabetes</i> , 2017 , 119-133	0.6	
5	Whole-Exome Sequencing of Patients with Severe Disorders of Insulin Action. <i>Frontiers in Diabetes</i> , 2014 , 87-101	0.6	
4	PPARδ and Glucose Homeostasis 2005 , 237-267		

3 An Evidence-Based Approach to Type 2 Diabetes **2006**, 303-321

2 Lipodystrophies **2019**, 1221-1227

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