

# Benjamin Glaser

## List of Publications by Year in Descending Order

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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.

212  
papers

16,276  
citations

62  
h-index

123  
g-index

228  
ext. papers

20,913  
ext. citations

10  
avg, IF

5.81  
L-index

#	Paper	IF	Citations
212	Liquid biopsy reveals collateral tissue damage in cancer.. <i>JCI Insight</i> , <b>2022</b> , 7,	9.9	3
211	Remote immune processes revealed by immune-derived circulating cell-free DNA. <i>ELife</i> , <b>2021</b> , 10,	8.9	3
210	NCMP-01. NOVEL BIOMARKERS FOR RADIATION-INDUCED NEUROTOXICITY. <i>Neuro-Oncology</i> , <b>2021</b> , 23, vi147-vi147	1	
209	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , <b>2021</b> , 12, 3505	17.4	5
208	Genetic activation of $\beta$ cell glucokinase in mice causes enhanced glucose-suppression of glucagon secretion during normal and diabetic states. <i>Molecular Metabolism</i> , <b>2021</b> , 49, 101193	8.8	6
207	ChIP-seq of plasma cell-free nucleosomes identifies gene expression programs of the cells of origin. <i>Nature Biotechnology</i> , <b>2021</b> , 39, 586-598	44.5	19
206	Biphasic dynamics of beta cell mass in a mouse model of congenital hyperinsulinism: implications for type 2 diabetes. <i>Diabetologia</i> , <b>2021</b> , 64, 1133-1143	10.3	6
205	What is a $\beta$ cell? - Chapter I in the Human Islet Research Network (HIRN) review series. <i>Molecular Metabolism</i> , <b>2021</b> , 53, 101323	8.8	4
204	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , <b>2020</b> , 581, 459-464	50.4	53
203	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , <b>2020</b> , 581, 434-443	50.4	2278
202	A structural variation reference for medical and population genetics. <i>Nature</i> , <b>2020</b> , 581, 444-451	50.4	223
201	Transcript expression-aware annotation improves rare variant interpretation. <i>Nature</i> , <b>2020</b> , 581, 452-458	50.4	55
200	Long-term outcomes in MEN-1 patients with pancreatic neuroendocrine neoplasms: an Israeli specialist center experience. <i>Endocrine</i> , <b>2020</b> , 68, 222-229	4	4
199	Multiplexing DNA methylation markers to detect circulating cell-free DNA derived from human pancreatic $\beta$ cells. <i>JCI Insight</i> , <b>2020</b> , 5,	9.9	12
198	Circulating Unmethylated Insulin DNA As a Biomarker of Human Beta Cell Death: A Multi-laboratory Assay Comparison. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2020</b> , 105,	5.6	11
197	Single-cell transcriptomics of human islet ontogeny defines the molecular basis of $\beta$ cell dedifferentiation in T2D. <i>Molecular Metabolism</i> , <b>2020</b> , 42, 101057	8.8	21
196	Distinct Imprinting Signatures and Biased Differentiation of Human Androgenetic and Parthenogenetic Embryonic Stem Cells. <i>Cell Stem Cell</i> , <b>2019</b> , 25, 419-432.e9	18	14

195	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. <i>Nature</i> , <b>2019</b> , 570, 71-76	50.4	129
194	Targeted demethylation at the CDKN1C/p57 locus induces human cell replication. <i>Journal of Clinical Investigation</i> , <b>2019</b> , 129, 209-214	15.9	24
193	Non-invasive detection of human cardiomyocyte death using methylation patterns of circulating DNA. <i>Nature Communications</i> , <b>2018</b> , 9, 1443	17.4	77
192	Beta Cell Death by Cell-free DNA and Outcome After Clinical Islet Transplantation. <i>Transplantation</i> , <b>2018</b> , 102, 978-985	1.8	27
191	Thyroidectomy Practice After Implementation of the 2015 American Thyroid Association Guidelines on Surgical Options for Patients With Well-Differentiated Thyroid Carcinoma. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , <b>2018</b> , 144, 427-432	3.9	19
190	IGF-1 levels may increase paradoxically with dopamine agonist treatment for prolactinomas. <i>Pituitary</i> , <b>2018</b> , 21, 406-413	4.3	6
189	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007329	6	41
188	Postnatal Exocrine Pancreas Growth by Cellular Hypertrophy Correlates with a Shorter Lifespan in Mammals. <i>Developmental Cell</i> , <b>2018</b> , 45, 726-737.e3	10.2	18
187	FOXP3 mutations causing early-onset insulin-requiring diabetes but without other features of immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. <i>Pediatric Diabetes</i> , <b>2018</b> , 19, 388-392	3.6	19
186	Monitoring liver damage using hepatocyte-specific methylation markers in cell-free circulating DNA. <i>JCI Insight</i> , <b>2018</b> , 3,	9.9	49
185	Comprehensive human cell-type methylation atlas reveals origins of circulating cell-free DNA in health and disease. <i>Nature Communications</i> , <b>2018</b> , 9, 5068	17.4	281
184	Type 2 diabetes genetic loci informed by multi-trait associations point to disease mechanisms and subtypes: A soft clustering analysis. <i>PLoS Medicine</i> , <b>2018</b> , 15, e1002654	11.6	180
183	Cell DNA Damage Response Promotes Islet Inflammation in Type 1 Diabetes. <i>Diabetes</i> , <b>2018</b> , 67, 2305-2318	18	20
182	Beta cell heterogeneity: an evolving concept. <i>Diabetologia</i> , <b>2017</b> , 60, 1363-1369	10.3	27
181	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. <i>Diabetes</i> , <b>2017</b> , 66, 2019-2032	0.9	29
180	Cells are not uniform after all—Novel insights into molecular heterogeneity of insulin-secreting cells. <i>Diabetes, Obesity and Metabolism</i> , <b>2017</b> , 19 Suppl 1, 147-152	6.7	18
179	Pancreatic Cells Express the Fetal Islet Hormone Gastrin in Rodent and Human Diabetes. <i>Diabetes</i> , <b>2017</b> , 66, 426-436	0.9	36
178	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. <i>Scientific Data</i> , <b>2017</b> , 4, 170179	8.2	22

177	Metabolic Stress and Compromised Identity of Pancreatic Beta Cells. <i>Frontiers in Genetics</i> , <b>2017</b> , 8, 21	4.5	63
176	PAX6 maintains $\beta$ cell identity by repressing genes of alternative islet cell types. <i>Journal of Clinical Investigation</i> , <b>2017</b> , 127, 230-243	15.9	77
175	Diagnosis of ABCC8 Congenital Hyperinsulinism of Infancy in a 20-Year-Old Man Evaluated for Factitious Hypoglycemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2017</b> , 102, 345-349	5.6	7
174	Hyperglycaemia induces metabolic dysfunction and glycogen accumulation in pancreatic $\beta$ cells. <i>Nature Communications</i> , <b>2016</b> , 7, 13496	17.4	67
173	Effects of ageing and senescence on pancreatic $\beta$ cell function. <i>Diabetes, Obesity and Metabolism</i> , <b>2016</b> , 18 Suppl 1, 58-62	6.7	33
172	Insulin receptor alternative splicing is regulated by insulin signaling and modulates beta cell survival. <i>Scientific Reports</i> , <b>2016</b> , 6, 31222	4.9	36
171	Abrogation of Autophagy by Chloroquine Alone or in Combination with mTOR Inhibitors Induces Apoptosis in Neuroendocrine Tumor Cells. <i>Neuroendocrinology</i> , <b>2016</b> , 103, 724-37	5.6	15
170	The Genetic Program of Pancreatic $\beta$ Cell Replication In Vivo. <i>Diabetes</i> , <b>2016</b> , 65, 2081-93	0.9	52
169	Identification of tissue-specific cell death using methylation patterns of circulating DNA. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2016</b> , 113, E1826-34	11.5	350
168	p16(Ink4a)-induced senescence of pancreatic beta cells enhances insulin secretion. <i>Nature Medicine</i> , <b>2016</b> , 22, 412-20	50.5	168
167	The genetic architecture of type 2 diabetes. <i>Nature</i> , <b>2016</b> , 536, 41-47	50.4	704
166	Dynamical compensation in physiological circuits. <i>Molecular Systems Biology</i> , <b>2016</b> , 12, 886	12.2	30
165	Identification of a G-Protein Subunit- $\beta$ 1 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). <i>Journal of Bone and Mineral Research</i> , <b>2016</b> , 31, 1207-14	6.3	33
164	Safety and efficacy of oral octreotide in acromegaly: results of a multicenter phase III trial. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2015</b> , 100, 1699-708	5.6	114
163	Premature aging of leukocyte DNA methylation is associated with type 2 diabetes prevalence. <i>Clinical Epigenetics</i> , <b>2015</b> , 7, 35	7.7	27
162	Aging-Dependent Demethylation of Regulatory Elements Correlates with Chromatin State and Improved $\beta$ Cell Function. <i>Cell Metabolism</i> , <b>2015</b> , 22, 619-32	24.6	129
161	Loss of Liver Kinase B1 (LKB1) in Beta Cells Enhances Glucose-stimulated Insulin Secretion Despite Profound Mitochondrial Defects. <i>Journal of Biological Chemistry</i> , <b>2015</b> , 290, 20934-20946	5.4	26
160	Weaning triggers a maturation step of pancreatic $\beta$ cells. <i>Developmental Cell</i> , <b>2015</b> , 32, 535-45	10.2	89

159	Disrupting mitochondrial-nuclear coevolution affects OXPHOS complex I integrity and impacts human health. <i>Genome Biology and Evolution</i> , <b>2014</b> , 6, 2665-80	3.9	56
158	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. <i>Nature Genetics</i> , <b>2014</b> , 46, 357-63	6.3	351
157	Type 2 diabetes and congenital hyperinsulinism cause DNA double-strand breaks and p53 activity in $\beta$ cells. <i>Cell Metabolism</i> , <b>2014</b> , 19, 109-21	24.6	101
156	G0-G1 transition and the restriction point in pancreatic $\beta$ cells in vivo. <i>Diabetes</i> , <b>2014</b> , 63, 578-84	0.9	20
155	Targeting the cell cycle inhibitor p57Kip2 promotes adult human $\beta$ cell replication. <i>Journal of Clinical Investigation</i> , <b>2014</b> , 124, 670-4	15.9	44
154	Systemic regulation of the age-related decline of pancreatic $\beta$ cell replication. <i>Diabetes</i> , <b>2013</b> , 62, 2843-8	0.9	89
153	The plastic pancreas. <i>Developmental Cell</i> , <b>2013</b> , 26, 3-7	10.2	68
152	Systemic regulation of the age-related decline of pancreatic $\beta$ cell replication. <i>Diabetes</i> 2013;62:2843-2848. <i>Diabetes</i> , <b>2013</b> , 62, 3300-3300	0.9	78
151	$\beta$ cell dedifferentiation and type 2 diabetes. <i>New England Journal of Medicine</i> , <b>2013</b> , 368, 572-3	59.2	57
150	Identification of a SIRT1 mutation in a family with type 1 diabetes. <i>Cell Metabolism</i> , <b>2013</b> , 17, 448-455	24.6	83
149	Gastrin: a distinct fate of neurogenin3 positive progenitor cells in the embryonic pancreas. <i>PLoS ONE</i> , <b>2013</b> , 8, e70397	3.7	33
148	The expression of the beta cell-derived autoimmune ligand for the killer receptor nkp46 is attenuated in type 2 diabetes. <i>PLoS ONE</i> , <b>2013</b> , 8, e74033	3.7	13
147	Genome-wide survey reveals predisposing diabetes type 2-related DNA methylation variations in human peripheral blood. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 371-83	5.6	272
146	Glucose metabolism: key endogenous regulator of $\beta$ cell replication and survival. <i>Diabetes, Obesity and Metabolism</i> , <b>2012</b> , 14 Suppl 3, 101-8	6.7	31
145	Pancreatic beta cells in very old mice retain capacity for compensatory proliferation. <i>Journal of Biological Chemistry</i> , <b>2012</b> , 287, 27407-14	5.4	53
144	Relative expression of a dominant mutated ABCC8 allele determines the clinical manifestation of congenital hyperinsulinism. <i>Diabetes</i> , <b>2012</b> , 61, 258-63	0.9	13
143	Ga-68 DOTA-NOC uptake in the pancreas: pathological and physiological patterns. <i>Clinical Nuclear Medicine</i> , <b>2012</b> , 37, 57-62	1.7	29
142	Control of pancreatic $\beta$ cell regeneration by glucose metabolism. <i>Cell Metabolism</i> , <b>2011</b> , 13, 440-449	24.6	229

141	ABCC8 mutation allele frequency in the Ashkenazi Jewish population and risk of focal hyperinsulinemic hypoglycemia. <i>Genetics in Medicine</i> , <b>2011</b> , 13, 891-4	8.1	20
140	Glucagonoma and the glucagonoma syndrome - cumulative experience with an elusive endocrine tumour. <i>Clinical Endocrinology</i> , <b>2011</b> , 74, 593-8	3.4	81
139	<sup>68</sup> Ga-DOTA-NOC PET/CT imaging of neuroendocrine tumors: comparison with <sup>111</sup> In-DTPA-octreotide (OctreoScan®). <i>Molecular Imaging and Biology</i> , <b>2011</b> , 13, 583-593	3.8	113
138	Glucose regulates cyclin D2 expression in quiescent and replicating pancreatic β cells through glycolysis and calcium channels. <i>Endocrinology</i> , <b>2011</b> , 152, 2589-98	4.8	49
137	Recognition and killing of human and murine pancreatic beta cells by the NK receptor NKp46. <i>Journal of Immunology</i> , <b>2011</b> , 187, 3096-103	5.3	48
136	Lessons in human biology from a monogenic pancreatic β cell disease. <i>Journal of Clinical Investigation</i> , <b>2011</b> , 121, 3821-5	15.9	7
135	Predicting diabetic nephropathy using a multifactorial genetic model. <i>PLoS ONE</i> , <b>2011</b> , 6, e18743	3.7	25
134	Detailed investigation of the role of common and low-frequency WFS1 variants in type 2 diabetes risk. <i>Diabetes</i> , <b>2010</b> , 59, 741-6	0.9	27
133	Large islets, beta-cell proliferation, and a glucokinase mutation. <i>New England Journal of Medicine</i> , <b>2010</b> , 362, 1348-50	59.2	71
132	Single pancreatic beta cells co-express multiple islet hormone genes in mice. <i>Diabetologia</i> , <b>2010</b> , 53, 128-38	10.3	55
131	Effects of moderate intensity glycemic control after cardiac surgery. <i>Annals of Thoracic Surgery</i> , <b>2010</b> , 90, 1825-32	2.7	36
130	Gene-gene interactions lead to higher risk for development of type 2 diabetes in an Ashkenazi Jewish population. <i>PLoS ONE</i> , <b>2010</b> , 5, e9903	3.7	40
129	Genetic analysis of complex disease--a roadmap to understanding or a colossal waste of money. <i>Pediatric Endocrinology Reviews</i> , <b>2010</b> , 7, 258-65	1.1	7
128	Preliminary evidence that a functional polymorphism in type 1 deiodinase is associated with enhanced potentiation of the antidepressant effect of sertraline by triiodothyronine. <i>Journal of Affective Disorders</i> , <b>2009</b> , 116, 113-6	6.6	35
127	Parental diabetes status reveals association of mitochondrial DNA haplogroup J1 with type 2 diabetes. <i>BMC Medical Genetics</i> , <b>2009</b> , 10, 60	2.1	28
126	Differences in mtDNA haplogroup distribution among 3 Jewish populations alter susceptibility to T2DM complications. <i>BMC Genomics</i> , <b>2008</b> , 9, 198	4.5	27
125	The H syndrome: a genodermatosis characterized by indurated, hyperpigmented, and hypertrichotic skin with systemic manifestations. <i>Journal of the American Academy of Dermatology</i> , <b>2008</b> , 59, 79-85	4.5	97
124	Novel de novo mutation in sulfonylurea receptor 1 presenting as hyperinsulinism in infancy followed by overt diabetes in early adolescence. <i>Diabetes</i> , <b>2008</b> , 57, 1935-40	0.9	38

123	Population-specific risk of type 2 diabetes conferred by HNF4A P2 promoter variants: a lesson for replication studies. <i>Diabetes</i> , <b>2008</b> , 57, 3161-5	0.9	33
122	Insulin mutations in diabetes: the clinical spectrum. <i>Diabetes</i> , <b>2008</b> , 57, 799-800	0.9	13
121	Long-acting somatostatin analogues are an effective treatment for type 1 gastric carcinoid tumours. <i>European Journal of Endocrinology</i> , <b>2008</b> , 159, 475-82	6.5	60
120	Post genome-wide association studies of novel genes associated with type 2 diabetes show gene-gene interaction and high predictive value. <i>PLoS ONE</i> , <b>2008</b> , 3, e2031	3.7	124
119	Impact of polymorphisms in WFS1 on prediabetic phenotypes in a population-based sample of middle-aged people with normal and abnormal glucose regulation. <i>Diabetologia</i> , <b>2008</b> , 51, 1646-52	10.3	40
118	Common variants in WFS1 confer risk of type 2 diabetes. <i>Nature Genetics</i> , <b>2007</b> , 39, 951-3	36.3	296
117	Ashkenazi Jewish mtDNA haplogroup distribution varies among distinct subpopulations: lessons of population substructure in a closed group. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 498-500	5.3	22
116	Sulfonylurea-responsive diabetes in childhood. <i>Journal of Pediatrics</i> , <b>2007</b> , 150, 553-5	3.6	12
115	Growth hormone reserve in adult beta thalassemia patients. <i>Endocrine</i> , <b>2007</b> , 31, 33-7		12
114	In-hospital treatment of hyperglycemia: effects of intensified subcutaneous insulin treatment. <i>Current Medical Research and Opinion</i> , <b>2007</b> , 23, 757-65	2.5	11
113	beta-Cell mitochondria exhibit membrane potential heterogeneity that can be altered by stimulatory or toxic fuel levels. <i>Diabetes</i> , <b>2007</b> , 56, 2569-78	0.9	89
112	Long-term neurodevelopmental outcome in conservatively treated congenital hyperinsulinism. <i>European Journal of Endocrinology</i> , <b>2007</b> , 157, 491-7	6.5	47
111	Combined treatment with sertraline and liothyronine in major depression: a randomized, double-blind, placebo-controlled trial. <i>Archives of General Psychiatry</i> , <b>2007</b> , 64, 679-88		78
110	Type 2 diabetes: hypoinsulinism, hyperinsulinism, or both?. <i>PLoS Medicine</i> , <b>2007</b> , 4, e148	11.6	15
109	Pendred Syndrome <b>2006</b> , 11, 154-168		
108	Germline fumarate hydratase mutations and evidence for a founder mutation underlying multiple cutaneous and uterine leiomyomata. <i>Journal of the American Academy of Dermatology</i> , <b>2005</b> , 52, 410-6	4.5	45
107	Malaria and asymptomatic parasitaemia in Gabonese infants under the age of 3 months. <i>Acta Tropica</i> , <b>2005</b> , 95, 81-5	3.2	19
106	p57Kip2 (cdkn1c): sequence, splice variants and unique temporal and spatial expression pattern in the rat pancreas. <i>Laboratory Investigation</i> , <b>2005</b> , 85, 364-75	5.9	12



105	A novel splice-site mutation in ECM-1 gene in a consanguineous family with lipid proteinosis. <i>Experimental Dermatology</i> , <b>2005</b> , 14, 891-7	4	19
104	CT of the ear in Pendred syndrome. <i>Radiology</i> , <b>2005</b> , 235, 537-40	20.5	32
103	Hyperinsulinemic hypoglycemia in Beckwith-Wiedemann syndrome due to defects in the function of pancreatic beta-cell adenosine triphosphate-sensitive potassium channels. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2005</b> , 90, 4376-82	5.6	56
102	Rapid and sustained relief from the symptoms of carcinoid syndrome: results from an open 6-month study of the 28-day prolonged-release formulation of lanreotide. <i>Neuroendocrinology</i> , <b>2004</b> , 80, 244-51	5.6	130
101	Glibenclamide treatment in permanent neonatal diabetes mellitus due to an activating mutation in Kir6.2. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 5504-7	5.6	165
100	Hyperinsulinism of infancy: novel ABCC8 and KCNJ11 mutations and evidence for additional locus heterogeneity. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 6224-34	5.6	67
99	A single-nucleotide polymorphism in the RAD51 gene modifies breast cancer risk in BRCA2 carriers, but not in BRCA1 carriers or noncarriers. <i>British Journal of Cancer</i> , <b>2004</b> , 90, 2002-5	8.7	90
98	Polyglutamine repeat length in the AIB1 gene modifies breast cancer susceptibility in BRCA1 carriers. <i>International Journal of Cancer</i> , <b>2004</b> , 108, 399-403	7.5	36
97	A common polymorphism in the upstream promoter region of the hepatocyte nuclear factor-4 alpha gene on chromosome 20q is associated with type 2 diabetes and appears to contribute to the evidence for linkage in an ashkenazi jewish population. <i>Diabetes</i> , <b>2004</b> , 53, 1134-40	0.9	195
96	Polymorphisms of the HDL receptor gene associated with HDL cholesterol levels in diabetic kindred from three populations. <i>Human Heredity</i> , <b>2003</b> , 55, 163-70	1.1	28
95	Germline fumarate hydratase mutations in families with multiple cutaneous and uterine leiomyomata. <i>Journal of Investigative Dermatology</i> , <b>2003</b> , 121, 741-4	4.3	48
94	Dominant SUR1 mutation causing autosomal dominant type 2 diabetes. <i>Lancet, The</i> , <b>2003</b> , 361, 272-3	40	8
93	Evidence for extensive locus heterogeneity in Naxos disease. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 118, 557-60	4.3	28
92	Compound heterozygosity for the common sulfonylurea receptor mutations can cause mild diazoxide-sensitive hyperinsulinism. <i>Clinical Pediatrics</i> , <b>2002</b> , 41, 183-6	1.2	22
91	Reproducibility of glucose measurements using the glucose sensor. <i>Diabetes Care</i> , <b>2002</b> , 25, 1185-91	14.6	108
90	Searching for type 2 diabetes genes on chromosome 20. <i>Diabetes</i> , <b>2002</b> , 51 Suppl 3, S308-15	0.9	23
89	Uncontrolled insulin secretion from a childhood pancreatic beta-cell adenoma is not due to the functional loss of ATP-sensitive potassium channels. <i>Endocrine-Related Cancer</i> , <b>2002</b> , 9, 221-6	5.7	6
88	Missense polymorphism in the human carboxypeptidase E gene alters enzymatic activity. <i>Human Mutation</i> , <b>2001</b> , 18, 120-31	4.7	68



87	Isolation and characterization of the human AKT1 gene, identification of 13 single nucleotide polymorphisms (SNPs), and their lack of association with Type II diabetes. <i>Diabetologia</i> , <b>2001</b> , 44, 910-3	10.3	10
86	p57(KIP2) expression in normal islet cells and in hyperinsulinism of infancy. <i>Diabetes</i> , <b>2001</b> , 50, 2763-9	0.9	77
85	Hyperinsulinism of infancy: the regulated release of insulin by KATP channel-independent pathways. <i>Diabetes</i> , <b>2001</b> , 50, 329-39	0.9	51
84	Dysregulation of insulin secretion in children with congenital hyperinsulinism due to sulfonylurea receptor mutations. <i>Diabetes</i> , <b>2001</b> , 50, 322-8	0.9	104
83	A genome scan for type 2 diabetes susceptibility loci in a genetically isolated population. <i>Diabetes</i> , <b>2001</b> , 50, 681-5	0.9	127
82	CAG and GGC repeat polymorphisms in the androgen receptor gene and breast cancer susceptibility in BRCA1/2 carriers and non-carriers. <i>British Journal of Cancer</i> , <b>2001</b> , 85, 36-40	8.7	59
81	Monilethrix: mutational hotspot in the helix termination motif of the human hair basic keratin 6. <i>Human Heredity</i> , <b>2000</b> , 50, 325-30	1.1	29
80	A recessive contiguous gene deletion causing infantile hyperinsulinism, enteropathy and deafness identifies the Usher type 1C gene. <i>Nature Genetics</i> , <b>2000</b> , 26, 56-60	36.3	257
79	Hyperinsulinism of the newborn. <i>Seminars in Perinatology</i> , <b>2000</b> , 24, 150-63	3.3	37
78	Genetics of neonatal hyperinsulinism. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , <b>2000</b> , 82, F79-86	4.7	139
77	The role of ATP-sensitive K <sup>+</sup> channels in familial hyperinsulinism <b>2000</b> , 299-325		2
76	Beta-cell proliferation and apoptosis in the developing normal human pancreas and in hyperinsulinism of infancy. <i>Diabetes</i> , <b>2000</b> , 49, 1325-33	0.9	316
75	Molecular basis and characterization of the hyperinsulinism/hyperammonemia syndrome: predominance of mutations in exons 11 and 12 of the glutamate dehydrogenase gene. HI/HA Contributing Investigators. <i>Diabetes</i> , <b>2000</b> , 49, 667-73	0.9	148
74	Calcium-stimulated insulin secretion in diffuse and focal forms of congenital hyperinsulinism. <i>Journal of Pediatrics</i> , <b>2000</b> , 137, 239-46	3.6	46
73	Hyperinsulinism caused by paternal-specific inheritance of a recessive mutation in the sulfonylurea-receptor gene. <i>Diabetes</i> , <b>1999</b> , 48, 1652-7	0.9	76
72	Early intensive insulin treatment for induction of long-term glycaemic control in type 2 diabetes. <i>Diabetes, Obesity and Metabolism</i> , <b>1999</b> , 1, 67-74	6.7	30
71	Intragenic single nucleotide polymorphism haplotype analysis of SUR1 mutations in familial hyperinsulinism. <i>Human Mutation</i> , <b>1999</b> , 14, 23-9	4.7	13
70	Neonatal Hyperinsulinism. <i>Trends in Endocrinology and Metabolism</i> , <b>1999</b> , 10, 55-61	8.8	28

69	Somatostatin receptor scintigraphy for early detection of regional and distant metastases of medullary carcinoma of the thyroid. <i>Clinical Nuclear Medicine</i> , <b>1999</b> , 24, 256-60	1.7	23
68	Mapping of the human insulin receptor substrate-2 gene, identification of a linked polymorphic marker and linkage analysis in families with Type II diabetes: no evidence for a major susceptibility role. <i>Diabetologia</i> , <b>1998</b> , 41, 1389-91	10.3	26
67	Clinical and molecular heterogeneity of familial hyperinsulinism. <i>Journal of Pediatrics</i> , <b>1998</b> , 133, 801-2	3.6	8
66	Familial hyperinsulinism with apparent autosomal dominant inheritance: clinical and genetic differences from the autosomal recessive variant. <i>Journal of Pediatrics</i> , <b>1998</b> , 132, 9-14	3.6	69
65	Pancreatic beta-cell glucokinase: closing the gap between theoretical concepts and experimental realities. <i>Diabetes</i> , <b>1998</b> , 47, 307-15	0.9	286
64	Familial hyperinsulinism caused by an activating glucokinase mutation. <i>New England Journal of Medicine</i> , <b>1998</b> , 338, 226-30	59.2	482
63	Genetic heterogeneity in familial hyperinsulinism. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 1119-28	5.6	105
62	Hepatocyte nuclear factor 1alpha coding mutations are an uncommon contributor to early-onset type 2 diabetes in Ashkenazi Jews. <i>Diabetes</i> , <b>1998</b> , 47, 967-9	0.9	16
61	Somatostatin-receptor scintigraphy in the management of gastroenteropancreatic tumors. <i>American Journal of Gastroenterology</i> , <b>1998</b> , 93, 66-70	0.7	55
60	Hyperinsulinism: molecular aetiology of focal disease. <i>Archives of Disease in Childhood</i> , <b>1998</b> , 79, 445-7	2.2	52
59	Functional analyses of novel mutations in the sulfonylurea receptor 1 associated with persistent hyperinsulinemic hypoglycemia of infancy. <i>Diabetes</i> , <b>1998</b> , 47, 1145-51	0.9	138
58	Isolation and characterization of the human PAX4 gene. <i>Diabetes</i> , <b>1998</b> , 47, 1650-3	0.9	14
57	Induction of long-term glycemic control in newly diagnosed type 2 diabetic patients by transient intensive insulin treatment. <i>Diabetes Care</i> , <b>1997</b> , 20, 1353-6	14.6	211
56	Pendred syndrome is caused by mutations in a putative sulphate transporter gene (PDS). <i>Nature Genetics</i> , <b>1997</b> , 17, 411-22	36.3	950
55	Genetics of NIDDM in France: studies with 19 candidate genes in affected sib pairs. <i>Diabetes</i> , <b>1997</b> , 46, 1062-1068	0.9	15
54	Adenosine diphosphate as an intracellular regulator of insulin secretion. <i>Science</i> , <b>1996</b> , 272, 1785-7	33.3	452
53	Normal proinsulin processing despite beta-cell dysfunction in persistent hyperinsulinaemic hypoglycaemia of infancy (nesidioblastosis). <i>Diabetologia</i> , <b>1996</b> , 39, 1338-44	10.3	9
52	Pendred syndrome maps to chromosome 7q21-34 and is caused by an intrinsic defect in thyroid iodine organification. <i>Nature Genetics</i> , <b>1996</b> , 12, 424-6	36.3	142

51	Mutations in the sulonylurea receptor gene are associated with familial hyperinsulinism in Ashkenazi Jews. <i>Human Molecular Genetics</i> , <b>1996</b> , 5, 1813-22	5.6	181
50	Studies in psychoneuroimmunology: psychological, immunological, and neuroendocrinological parameters in Israeli civilians during and after a period of Scud missile attacks. <i>Behavioral Medicine</i> , <b>1996</b> , 22, 5-14	4.4	30
49	Hyperinsulinemic hypoglycemia of infancy (nesidioblastosis) in clinical remission: high incidence of diabetes mellitus and persistent beta-cell dysfunction at long-term follow-up. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1995</b> , 80, 386-392	5.6	83
48	Familial hyperinsulinism maps to chromosome 11p14-15.1, 30 cM centromeric to the insulin gene. <i>Nature Genetics</i> , <b>1994</b> , 7, 185-8	36.3	102
47	Improvement of sleep apnoea due to acromegaly during short-term treatment with octreotide. <i>Journal of Internal Medicine</i> , <b>1994</b> , 236, 231-5	10.8	19
46	Gallbladder visualization with In-111 labeled octreotide. <i>Clinical Nuclear Medicine</i> , <b>1994</b> , 19, 133-5	1.7	11
45	Somatostatin-receptor imaging of medullary thyroid carcinoma. <i>Clinical Nuclear Medicine</i> , <b>1994</b> , 19, 416-21	2.7	19
44	Persistent hyperinsulinemic hypoglycemia of infancy: long-term octreotide treatment without pancreatectomy. <i>Journal of Pediatrics</i> , <b>1993</b> , 123, 644-50	3.6	127
43	Recurrence-associated mortality in patients with differentiated thyroid carcinoma. <i>Journal of Surgical Oncology</i> , <b>1993</b> , 52, 164-8	2.8	9
42	Cross-sectional and longitudinal study of the pituitary-thyroid axis in patients with thalassaemia major. <i>Clinical Endocrinology</i> , <b>1993</b> , 38, 55-61	3.4	36
41	TSH producing pituitary tumor: biochemical diagnosis and long-term medical management with octreotide. <i>Hormone and Metabolic Research</i> , <b>1992</b> , 24, 34-8	3.1	4
40	Glycosylated serum protein levels assayed with highly sensitive immunoradiometric assay accurately reflect glycemic control of diabetic patients. <i>Diabetes Care</i> , <b>1992</b> , 15, 645-50	14.6	7
39	Growth-hormone-binding protein in patients with acromegaly. <i>Hormone Research</i> , <b>1992</b> , 37, 205-11		23
38	Unusual causes of I-131 metaiodobenzylguanidine uptake in non-neural crest tissue. <i>Clinical Nuclear Medicine</i> , <b>1991</b> , 16, 239-42	1.7	20
37	Sebum measurements for rapid identification of hyperandrogenism due to an ovarian Leydig cell tumor. <i>International Journal of Dermatology</i> , <b>1991</b> , 30, 276-7	1.7	6
36	Effect of 6-month gliclazide treatment on insulin release and sensitivity to endogenous insulin in NIDDM: role of initial continuous subcutaneous insulin infusion-induced normoglycemia. <i>American Journal of Medicine</i> , <b>1991</b> , 90, 37S-45S	2.4	14
35	Effect of 6 months' gliclazide treatment on insulin release and sensitivity to endogenous insulin in NIDDM: role of initial CSII-induced normoglycemia. <i>Diabetes Research and Clinical Practice</i> , <b>1991</b> , 14 Suppl 2, S69-78	7.4	3
34	Long-term treatment with the somatostatin analogue SMS 201-995: alternative to pancreatectomy in persistent hyperinsulinaemic hypoglycaemia of infancy. <i>Digestion</i> , <b>1990</b> , 45 Suppl 1, 27-35	3.6	56

33	Regulation of insulin release in persistent hyperinsulinaemic hypoglycaemia of infancy studied in long-term culture of pancreatic tissue. <i>Diabetologia</i> , <b>1990</b> , 33, 482-8	10.3	28
32	Persistent hyperinsulinemic hypoglycemia of infancy ("nesidioblastosis"): autosomal recessive inheritance in 7 pedigrees. <i>American Journal of Medical Genetics Part A</i> , <b>1990</b> , 37, 511-5		31
31	Persistent hyperinsulinaemic hypoglycaemia of infancy: long-term treatment with the somatostatin analogue Sandostatin. <i>Clinical Endocrinology</i> , <b>1989</b> , 31, 71-80	3.4	42
30	Gastrin-producing ovarian cystadenocarcinoma: sensitivity to secretin and SMS 201-995. <i>Gastroenterology</i> , <b>1989</b> , 97, 464-7	13.3	21
29	Lipid cell tumor of the ovary: steroid hormone secretory pattern and localization using <sup>75</sup> Se-selenomethylcholesterol. <i>Gynecologic and Obstetric Investigation</i> , <b>1989</b> , 27, 110-2	2.5	3
28	Effects of secretin on the normal and pathological beta-cell. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1988</b> , 66, 1138-43	5.6	23
27	Pancreatic polypeptide response to secretin in obesity: effects of glucose intolerance. <i>Hormone and Metabolic Research</i> , <b>1988</b> , 20, 288-92	3.1	57
26	Improved beta-cell function after intensive insulin treatment in severe non-insulin-dependent diabetes. <i>European Journal of Endocrinology</i> , <b>1988</b> , 118, 365-73	6.5	69
25	Toxic multinodular goiter: a variant of autoimmune hyperthyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1987</b> , 65, 659-64	5.6	44
24	Thyrotrophin receptor blocking antibodies: incidence, characterization and in-vitro synthesis. <i>Clinical Endocrinology</i> , <b>1987</b> , 27, 409-21	3.4	55
23	Magnetic resonance imaging of the pituitary gland. <i>Clinical Radiology</i> , <b>1986</b> , 37, 9-14	2.9	14
22	L-Thyroxine-induced leukopenia in a patient with Hashimoto's disease: involvement of suppressor-cytotoxic T cells. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1985</b> , 61, 980-2	5.6	2
21	Leydig-cell tumor of the ovary: visualization using <sup>131</sup> I-19-iodocholesterol scintigraphy. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , <b>1985</b> , 11, 13-6		3
20	Cellular immune functions in patients with primary hyperparathyroidism: effects of histamine and cimetidine. <i>Immunopharmacology</i> , <b>1985</b> , 9, 81-6		
19	Do cyclic AMP concentrations in saliva reflect PTH biologic activity?. <i>Metabolism: Clinical and Experimental</i> , <b>1985</b> , 34, 505-8	12.7	2
18	Effect of acute cimetidine administration on indices of parathyroid hormone action in healthy subjects and patients with primary and secondary hyperparathyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1984</b> , 59, 993-7	5.6	3
17	Sodium valproate and metyrapone for pituitary-dependent Cushing's disease. <i>Lancet, The</i> , <b>1984</b> , 2, 640	40	7
16	Secretion of pancreatic polypeptide in man in response to beef ingestion is mediated in part by an extravagal cholinergic mechanism. <i>Metabolism: Clinical and Experimental</i> , <b>1983</b> , 32, 57-61	12.7	15

15	Sex difference in the sensitivity of the human pancreatic polypeptide cell to autonomic nervous stimulation in man. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1983</b> , 56, 21-5	5.6	5
14	A spot sample test for the estimation of urinary or nephrogenous cAMP in the evaluation of parathyroid function. <i>Nephron</i> , <b>1983</b> , 35, 264-6	3.3	1
13	Percutaneous transhepatic venous sampling of gastrin: value in sporadic and familial islet-cell tumors and G-cell hyperfunction. <i>New England Journal of Medicine</i> , <b>1982</b> , 307, 293-7	59.2	69
12	A New Syndrome of Symptomatic Cutaneous Mastocytoma Producing Vasoactive Intestinal Polypeptide. <i>Gastroenterology</i> , <b>1982</b> , 82, 963-967	13.3	28
11	Gastrointestinal/pancreatic hormone concentrations in the portal venous system of nine patients with organic hyperinsulinism. <i>Metabolism: Clinical and Experimental</i> , <b>1981</b> , 30, 1001-10	12.7	29
10	Truncal vagotomy abolishes the somatostatin response to insulin-induced hypoglycemia in man. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1981</b> , 52, 823-5	5.6	30
9	Plasma human pancreatic polypeptide responses to administered secretin: effects of surgical vagotomy, cholinergic blockade, and chronic pancreatitis. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1980</b> , 50, 1094-9	5.6	70
8	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population		2
7	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes		1
6	Rare coding variants in 35 genes associate with circulating lipid levels in a multi-ancestry analysis of 170,000 exomes		2
5	Clustering of Type 2 Diabetes Genetic Loci by Multi-Trait Associations Identifies Disease Mechanisms and Subtypes		5
4	Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries		2
3	Specific detection of cell-free DNA derived from intestinal epithelial cells using methylation patterns		2
2	ChIP-seq of plasma cell-free nucleosomes identifies cell-of-origin gene expression programs		6
1	Sequencing of over 100,000 individuals identifies multiple genes and rare variants associated with Crohn's disease susceptibility		2