# Benjamin Glaser

## List of Publications by Citations



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|---|---|---|---|
| 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 | The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443 | 50.4 | 2278 |
| 211 | Pendred syndrome is caused by mutations in a putative sulphate transporter gene (PDS). *Nature Genetics*, **1997**, 17, 411-22 | 36.3 | 950 |
| 210 | The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47 | 50.4 | 704 |
| 209 | Familial hyperinsulinism caused by an activating glucokinase mutation. *New England Journal of Medicine*, **1998**, 338, 226-30 | 59.2 | 482 |
| 208 | Adenosine diphosphate as an intracellular regulator of insulin secretion. *Science*, **1996**, 272, 1785-7 | 33.3 | 452 |
| 207 | Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. *Nature Genetics*, **2014**, 46, 357-63 | 36.3 | 351 |
| 206 | Identification of tissue-specific cell death using methylation patterns of circulating DNA. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, E1826-34 | 11.5 | 350 |
| 205 | Beta-cell proliferation and apoptosis in the developing normal human pancreas and in hyperinsulinism of infancy. *Diabetes*, **2000**, 49, 1325-33 | 0.9 | 316 |
| 204 | Common variants in WFS1 confer risk of type 2 diabetes. *Nature Genetics*, **2007**, 39, 951-3 | 36.3 | 296 |
| 203 | Pancreatic beta-cell glucokinase: closing the gap between theoretical concepts and experimental realities. *Diabetes*, **1998**, 47, 307-15 | 0.9 | 286 |
| 202 | Comprehensive human cell-type methylation atlas reveals origins of circulating cell-free DNA in health and disease. *Nature Communications*, **2018**, 9, 5068 | 17.4 | 281 |
| 201 | Genome-wide survey reveals predisposing diabetes type 2-related DNA methylation variations in human peripheral blood. *Human Molecular Genetics*, **2012**, 21, 371-83 | 5.6 | 272 |
| 200 | A recessive contiguous gene deletion causing infantile hyperinsulinism, enteropathy and deafness identifies the Usher type 1C gene. *Nature Genetics*, **2000**, 26, 56-60 | 36.3 | 257 |
| 199 | Control of pancreatic □-cell regeneration by glucose metabolism. *Cell Metabolism*, **2011**, 13, 440-449 | 24.6 | 229 |
| 198 | A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451 | 50.4 | 223 |
| 197 | Induction of long-term glycemic control in newly diagnosed type 2 diabetic patients by transient intensive insulin treatment. *Diabetes Care*, **1997**, 20, 1353-6 | 14.6 | 211 |
| 196 | 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. *Diabetes*, **2004**, 53, 1134-40 | 0.9 | 195 |

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| 195 | Mutations in the sulonylurea receptor gene are associated with familial hyperinsulinism in Ashkenazi Jews. *Human Molecular Genetics*, **1996**, 5, 1813-22 | 5.6 | 181 |
| 194 | Type 2 diabetes genetic loci informed by multi-trait associations point to disease mechanisms and subtypes: A soft clustering analysis. *PLoS Medicine*, **2018**, 15, e1002654 | 11.6 | 180 |
| 193 | p16(Ink4a)-induced senescence of pancreatic beta cells enhances insulin secretion. *Nature Medicine*, **2016**, 22, 412-20 | 50.5 | 168 |
| 192 | Glibenclamide treatment in permanent neonatal diabetes mellitus due to an activating mutation in Kir6.2. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 5504-7 | 5.6 | 165 |
| 191 | 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. *Diabetes*, **2000**, 49, 667-73 | 0.9 | 148 |
| 190 | Pendred syndrome maps to chromosome 7q21-34 and is caused by an intrinsic defect in thyroid iodine organification. *Nature Genetics*, **1996**, 12, 424-6 | 36.3 | 142 |
| 189 | Genetics of neonatal hyperinsulinism. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2000**, 82, F79-86 | 4.7 | 139 |
| 188 | Functional analyses of novel mutations in the sulfonylurea receptor 1 associated with persistent hyperinsulinemic hypoglycemia of infancy. *Diabetes*, **1998**, 47, 1145-51 | 0.9 | 138 |
| 187 | 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. *Neuroendocrinology*, **2004**, 80, 244-51 | 5.6 | 130 |
| 186 | Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. *Nature*, **2019**, 570, 71-76 | 50.4 | 129 |
| 185 | Aging-Dependent Demethylation of Regulatory Elements Correlates with Chromatin State and Improved  Cell Function. *Cell Metabolism*, **2015**, 22, 619-32 | 24.6 | 129 |
| 184 | A genome scan for type 2 diabetes susceptibility loci in a genetically isolated population. *Diabetes*, **2001**, 50, 681-5 | 0.9 | 127 |
| 183 | Persistent hyperinsulinemic hypoglycemia of infancy: long-term octreotide treatment without pancreatectomy. *Journal of Pediatrics*, **1993**, 123, 644-50 | 3.6 | 127 |
| 182 | Post genome-wide association studies of novel genes associated with type 2 diabetes show gene-gene interaction and high predictive value. *PLoS ONE*, **2008**, 3, e2031 | 3.7 | 124 |
| 181 | Safety and efficacy of oral octreotide in acromegaly: results of a multicenter phase III trial. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 1699-708 | 5.6 | 114 |
| 180 | 68Ga-DOTA-NOC PET/CT imaging of neuroendocrine tumors: comparison with  In-DTPA-octreotide (OctreoScan ). *Molecular Imaging and Biology*, **2011**, 13, 583-593 | 3.8 | 113 |
| 179 | Reproducibility of glucose measurements using the glucose sensor. *Diabetes Care*, **2002**, 25, 1185-91 | 14.6 | 108 |
| 178 | Genetic heterogeneity in familial hyperinsulinism. *Human Molecular Genetics*, **1998**, 7, 1119-28 | 5.6 | 105 |

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|---|---|---|---|
| 177 | Dysregulation of insulin secretion in children with congenital hyperinsulinism due to sulfonylurea receptor mutations. *Diabetes*, **2001**, 50, 322-8 | 0.9 | 104 |
| 176 | Familial hyperinsulinism maps to chromosome 11p14-15.1, 30 cM centromeric to the insulin gene. *Nature Genetics*, **1994**, 7, 185-8 | 36.3 | 102 |
| 175 | Type 2 diabetes and congenital hyperinsulinism cause DNA double-strand breaks and p53 activity in β cells. *Cell Metabolism*, **2014**, 19, 109-21 | 24.6 | 101 |
| 174 | The H syndrome: a genodermatosis characterized by indurated, hyperpigmented, and hypertrichotic skin with systemic manifestations. *Journal of the American Academy of Dermatology*, **2008**, 59, 79-85 | 4.5 | 97 |
| 173 | A single-nucleotide polymorphism in the RAD51 gene modifies breast cancer risk in BRCA2 carriers, but not in BRCA1 carriers or noncarriers. *British Journal of Cancer*, **2004**, 90, 2002-5 | 8.7 | 90 |
| 172 | Systemic regulation of the age-related decline of pancreatic β cell replication. *Diabetes*, **2013**, 62, 2843-8 | 0.9 | 89 |
| 171 | Weaning triggers a maturation step of pancreatic β cells. *Developmental Cell*, **2015**, 32, 535-45 | 10.2 | 89 |
| 170 | beta-Cell mitochondria exhibit membrane potential heterogeneity that can be altered by stimulatory or toxic fuel levels. *Diabetes*, **2007**, 56, 2569-78 | 0.9 | 89 |
| 169 | Identification of a SIRT1 mutation in a family with type 1 diabetes. *Cell Metabolism*, **2013**, 17, 448-455 | 24.6 | 83 |
| 168 | Hyperinsulinemic hypoglycemia of infancy (nesidioblastosis) in clinical remission: high incidence of diabetes mellitus and persistent beta-cell dysfunction at long-term follow-up. *Journal of Clinical Endocrinology and Metabolism*, **1995**, 80, 386-392 | 5.6 | 83 |
| 167 | Glucagonoma and the glucagonoma syndrome - cumulative experience with an elusive endocrine tumour. *Clinical Endocrinology*, **2011**, 74, 593-8 | 3.4 | 81 |
| 166 | Systemic regulation of the age-related decline of pancreatic β cell replication. Diabetes 2013;62:2843–2848. *Diabetes*, **2013**, 62, 3300-3300 | 0.9 | 78 |
| 165 | Combined treatment with sertraline and liothyronine in major depression: a randomized, double-blind, placebo-controlled trial. *Archives of General Psychiatry*, **2007**, 64, 679-88 | | 78 |
| 164 | Non-invasive detection of human cardiomyocyte death using methylation patterns of circulating DNA. *Nature Communications*, **2018**, 9, 1443 | 17.4 | 77 |
| 163 | p57(KIP2) expression in normal islet cells and in hyperinsulinism of infancy. *Diabetes*, **2001**, 50, 2763-9 | 0.9 | 77 |
| 162 | PAX6 maintains β cell identity by repressing genes of alternative islet cell types. *Journal of Clinical Investigation*, **2017**, 127, 230-243 | 15.9 | 77 |
| 161 | Hyperinsulinism caused by paternal-specific inheritance of a recessive mutation in the sulfonylurea-receptor gene. *Diabetes*, **1999**, 48, 1652-7 | 0.9 | 76 |
| 160 | Large islets, beta-cell proliferation, and a glucokinase mutation. *New England Journal of Medicine*, **2010**, 362, 1348-50 | 59.2 | 71 |

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|---|---|---|---|
| 159 | Plasma human pancreatic polypeptide responses to administered secretin: effects of surgical vagotomy, cholinergic blockade, and chronic pancreatitis. *Journal of Clinical Endocrinology and Metabolism*, **1980**, 50, 1094-9 | 5.6 | 70 |
| 158 | Familial hyperinsulinism with apparent autosomal dominant inheritance: clinical and genetic differences from the autosomal recessive variant. *Journal of Pediatrics*, **1998**, 132, 9-14 | 3.6 | 69 |
| 157 | Improved beta-cell function after intensive insulin treatment in severe non-insulin-dependent diabetes. *European Journal of Endocrinology*, **1988**, 118, 365-73 | 6.5 | 69 |
| 156 | Percutaneous transhepatic venous sampling of gastrin: value in sporadic and familial islet-cell tumors and G-cell hyperfunction. *New England Journal of Medicine*, **1982**, 307, 293-7 | 59.2 | 69 |
| 155 | The plastic pancreas. *Developmental Cell*, **2013**, 26, 3-7 | 10.2 | 68 |
| 154 | Missense polymorphism in the human carboxypeptidase E gene alters enzymatic activity. *Human Mutation*, **2001**, 18, 120-31 | 4.7 | 68 |
| 153 | Hyperglycaemia induces metabolic dysfunction and glycogen accumulation in pancreatic □cells. *Nature Communications*, **2016**, 7, 13496 | 17.4 | 67 |
| 152 | Hyperinsulinism of infancy: novel ABCC8 and KCNJ11 mutations and evidence for additional locus heterogeneity. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 6224-34 | 5.6 | 67 |
| 151 | Metabolic Stress and Compromised Identity of Pancreatic Beta Cells. *Frontiers in Genetics*, **2017**, 8, 21 | 4.5 | 63 |
| 150 | Long-acting somatostatin analogues are an effective treatment for type 1 gastric carcinoid tumours. *European Journal of Endocrinology*, **2008**, 159, 475-82 | 6.5 | 60 |
| 149 | CAG and GGC repeat polymorphisms in the androgen receptor gene and breast cancer susceptibility in BRCA1/2 carriers and non-carriers. *British Journal of Cancer*, **2001**, 85, 36-40 | 8.7 | 59 |
| 148 | □cell dedifferentiation and type 2 diabetes. *New England Journal of Medicine*, **2013**, 368, 572-3 | 59.2 | 57 |
| 147 | Pancreatic polypeptide response to secretin in obesity: effects of glucose intolerance. *Hormone and Metabolic Research*, **1988**, 20, 288-92 | 3.1 | 57 |
| 146 | Disrupting mitochondrial-nuclear coevolution affects OXPHOS complex I integrity and impacts human health. *Genome Biology and Evolution*, **2014**, 6, 2665-80 | 3.9 | 56 |
| 145 | Hyperinsulinemic hypoglycemia in Beckwith-Wiedemann syndrome due to defects in the function of pancreatic beta-cell adenosine triphosphate-sensitive potassium channels. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 4376-82 | 5.6 | 56 |
| 144 | Long-term treatment with the somatostatin analogue SMS 201-995: alternative to pancreatectomy in persistent hyperinsulinaemic hypoglycaemia of infancy. *Digestion*, **1990**, 45 Suppl 1, 27-35 | 3.6 | 56 |
| 143 | Transcript expression-aware annotation improves rare variant interpretation. *Nature*, **2020**, 581, 452-458 | 50.4 | 55 |
| 142 | Single pancreatic beta cells co-express multiple islet hormone genes in mice. *Diabetologia*, **2010**, 53, 128-38 | 10.3 | 55 |

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| 141 | Somatostatin-receptor scintigraphy in the management of gastroenteropancreatic tumors. *American Journal of Gastroenterology*, **1998**, 93, 66-70 | 0.7 | 55 |
| 140 | Thyrotrophin receptor blocking antibodies: incidence, characterization and in-vitro synthesis. *Clinical Endocrinology*, **1987**, 27, 409-21 | 3.4 | 55 |
| 139 | Evaluating drug targets through human loss-of-function genetic variation. *Nature*, **2020**, 581, 459-464 | 50.4 | 53 |
| 138 | Pancreatic beta cells in very old mice retain capacity for compensatory proliferation. *Journal of Biological Chemistry*, **2012**, 287, 27407-14 | 5.4 | 53 |
| 137 | The Genetic Program of Pancreatic β-Cell Replication In Vivo. *Diabetes*, **2016**, 65, 2081-93 | 0.9 | 52 |
| 136 | Hyperinsulinism: molecular aetiology of focal disease. *Archives of Disease in Childhood*, **1998**, 79, 445-7 | 2.2 | 52 |
| 135 | Hyperinsulinism of infancy: the regulated release of insulin by KATP channel-independent pathways. *Diabetes*, **2001**, 50, 329-39 | 0.9 | 51 |
| 134 | Glucose regulates cyclin D2 expression in quiescent and replicating pancreatic β-cells through glycolysis and calcium channels. *Endocrinology*, **2011**, 152, 2589-98 | 4.8 | 49 |
| 133 | Monitoring liver damage using hepatocyte-specific methylation markers in cell-free circulating DNA. *JCI Insight*, **2018**, 3, | 9.9 | 49 |
| 132 | Recognition and killing of human and murine pancreatic beta cells by the NK receptor NKp46. *Journal of Immunology*, **2011**, 187, 3096-103 | 5.3 | 48 |
| 131 | Germline fumarate hydratase mutations in families with multiple cutaneous and uterine leiomyomata. *Journal of Investigative Dermatology*, **2003**, 121, 741-4 | 4.3 | 48 |
| 130 | Long-term neurodevelopmental outcome in conservatively treated congenital hyperinsulinism. *European Journal of Endocrinology*, **2007**, 157, 491-7 | 6.5 | 47 |
| 129 | Calcium-stimulated insulin secretion in diffuse and focal forms of congenital hyperinsulinism. *Journal of Pediatrics*, **2000**, 137, 239-46 | 3.6 | 46 |
| 128 | Germline fumarate hydratase mutations and evidence for a founder mutation underlying multiple cutaneous and uterine leiomyomata. *Journal of the American Academy of Dermatology*, **2005**, 52, 410-6 | 4.5 | 45 |
| 127 | Toxic multinodular goiter: a variant of autoimmune hyperthyroidism. *Journal of Clinical Endocrinology and Metabolism*, **1987**, 65, 659-64 | 5.6 | 44 |
| 126 | Targeting the cell cycle inhibitor p57Kip2 promotes adult human β cell replication. *Journal of Clinical Investigation*, **2014**, 124, 670-4 | 15.9 | 44 |
| 125 | Persistent hyperinsulinaemic hypoglycaemia of infancy: long-term treatment with the somatostatin analogue Sandostatin. *Clinical Endocrinology*, **1989**, 31, 71-80 | 3.4 | 42 |
| 124 | Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. *PLoS Genetics*, **2018**, 14, e1007329 | 6 | 41 |

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|---|---|---|---|
| 123 | Impact of polymorphisms in WFS1 on prediabetic phenotypes in a population-based sample of middle-aged people with normal and abnormal glucose regulation. *Diabetologia*, **2008**, 51, 1646-52 | 10.3 | 40 |
| 122 | Gene-gene interactions lead to higher risk for development of type 2 diabetes in an Ashkenazi Jewish population. *PLoS ONE*, **2010**, 5, e9903 | 3.7 | 40 |
| 121 | Novel de novo mutation in sulfonylurea receptor 1 presenting as hyperinsulinism in infancy followed by overt diabetes in early adolescence. *Diabetes*, **2008**, 57, 1935-40 | 0.9 | 38 |
| 120 | Hyperinsulinism of the newborn. *Seminars in Perinatology*, **2000**, 24, 150-63 | 3.3 | 37 |
| 119 | Insulin receptor alternative splicing is regulated by insulin signaling and modulates beta cell survival. *Scientific Reports*, **2016**, 6, 31222 | 4.9 | 36 |
| 118 | Pancreatic Cells Express the Fetal Islet Hormone Gastrin in Rodent and Human Diabetes. *Diabetes* , **2017**, 66, 426-436 | 0.9 | 36 |
| 117 | Effects of moderate intensity glycemic control after cardiac surgery. *Annals of Thoracic Surgery*, **2010**, 90, 1825-32 | 2.7 | 36 |
| 116 | Polyglutamine repeat length in the AIB1 gene modifies breast cancer susceptibility in BRCA1 carriers. *International Journal of Cancer*, **2004**, 108, 399-403 | 7.5 | 36 |
| 115 | Cross-sectional and longitudinal study of the pituitary-thyroid axis in patients with thalassaemia major. *Clinical Endocrinology*, **1993**, 38, 55-61 | 3.4 | 36 |
| 114 | Preliminary evidence that a functional polymorphism in type 1 deiodinase is associated with enhanced potentiation of the antidepressant effect of sertraline by triiodothyronine. *Journal of Affective Disorders*, **2009**, 116, 113-6 | 6.6 | 35 |
| 113 | Effects of ageing and senescence on pancreatic cell function. *Diabetes, Obesity and Metabolism*, **2016**, 18 Suppl 1, 58-62 | 6.7 | 33 |
| 112 | Population-specific risk of type 2 diabetes conferred by HNF4A P2 promoter variants: a lesson for replication studies. *Diabetes*, **2008**, 57, 3161-5 | 0.9 | 33 |
| 111 | Gastrin: a distinct fate of neurogenin3 positive progenitor cells in the embryonic pancreas. *PLoS ONE*, **2013**, 8, e70397 | 3.7 | 33 |
| 110 | Identification of a G-Protein Subunit-1 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). *Journal of Bone and Mineral Research*, **2016**, 31, 1207-14 | 6.3 | 33 |
| 109 | CT of the ear in Pendred syndrome. *Radiology*, **2005**, 235, 537-40 | 20.5 | 32 |
| 108 | Glucose metabolism: key endogenous regulator of cell replication and survival. *Diabetes, Obesity and Metabolism*, **2012**, 14 Suppl 3, 101-8 | 6.7 | 31 |
| 107 | Persistent hyperinsulinemic hypoglycemia of infancy ("nesidioblastosis"): autosomal recessive inheritance in 7 pedigrees. *American Journal of Medical Genetics Part A*, **1990**, 37, 511-5 | | 31 |
| 106 | Early intensive insulin treatment for induction of long-term glycaemic control in type 2 diabetes. *Diabetes, Obesity and Metabolism*, **1999**, 1, 67-74 | 6.7 | 30 |

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| 105 | Studies in psychoneuroimmunology: psychological, immunological, and neuroendocrinological parameters in Israeli civilians during and after a period of Scud missile attacks. *Behavioral Medicine*, **1996**, 22, 5-14 | 4.4 | 30 |
| 104 | Truncal vagotomy abolishes the somatostatin response to insulin-induced hypoglycemia in man. *Journal of Clinical Endocrinology and Metabolism*, **1981**, 52, 823-5 | 5.6 | 30 |
| 103 | Dynamical compensation in physiological circuits. *Molecular Systems Biology*, **2016**, 12, 886 | 12.2 | 30 |
| 102 | A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. *Diabetes*, **2017**, 66, 2019-2032 | 0.9 | 29 |
| 101 | Ga-68 DOTA-NOC uptake in the pancreas: pathological and physiological patterns. *Clinical Nuclear Medicine*, **2012**, 37, 57-62 | 1.7 | 29 |
| 100 | Monilethrix: mutational hotspot in the helix termination motif of the human hair basic keratin 6. *Human Heredity*, **2000**, 50, 325-30 | 1.1 | 29 |
| 99 | Gastrointestinal/pancreatic hormone concentrations in the portal venous system of nine patients with organic hyperinsulinism. *Metabolism: Clinical and Experimental*, **1981**, 30, 1001-10 | 12.7 | 29 |
| 98 | Parental diabetes status reveals association of mitochondrial DNA haplogroup J1 with type 2 diabetes. *BMC Medical Genetics*, **2009**, 10, 60 | 2.1 | 28 |
| 97 | Polymorphisms of the HDL receptor gene associated with HDL cholesterol levels in diabetic kindred from three populations. *Human Heredity*, **2003**, 55, 163-70 | 1.1 | 28 |
| 96 | Evidence for extensive locus heterogeneity in Naxos disease. *Journal of Investigative Dermatology*, **2002**, 118, 557-60 | 4.3 | 28 |
| 95 | Neonatal Hyperinsulinism. *Trends in Endocrinology and Metabolism*, **1999**, 10, 55-61 | 8.8 | 28 |
| 94 | Regulation of insulin release in persistent hyperinsulinaemic hypoglycaemia of infancy studied in long-term culture of pancreatic tissue. *Diabetologia*, **1990**, 33, 482-8 | 10.3 | 28 |
| 93 | A New Syndrome of Symptomatic Cutaneous Mastocytoma Producing Vasoactive Intestinal Polypeptide. *Gastroenterology*, **1982**, 82, 963-967 | 13.3 | 28 |
| 92 | Beta cell heterogeneity: an evolving concept. *Diabetologia*, **2017**, 60, 1363-1369 | 10.3 | 27 |
| 91 | Premature aging of leukocyte DNA methylation is associated with type 2 diabetes prevalence. *Clinical Epigenetics*, **2015**, 7, 35 | 7.7 | 27 |
| 90 | Beta Cell Death by Cell-free DNA and Outcome After Clinical Islet Transplantation. *Transplantation*, **2018**, 102, 978-985 | 1.8 | 27 |
| 89 | Detailed investigation of the role of common and low-frequency WFS1 variants in type 2 diabetes risk. *Diabetes*, **2010**, 59, 741-6 | 0.9 | 27 |
| 88 | Differences in mtDNA haplogroup distribution among 3 Jewish populations alter susceptibility to T2DM complications. *BMC Genomics*, **2008**, 9, 198 | 4.5 | 27 |

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|---|---|---|---|
| 87 | Loss of Liver Kinase B1 (LKB1) in Beta Cells Enhances Glucose-stimulated Insulin Secretion Despite Profound Mitochondrial Defects. *Journal of Biological Chemistry*, **2015**, 290, 20934-20946 | 5.4 | 26 |
| 86 | 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. *Diabetologia*, **1998**, 41, 1389-91 | 10.3 | 26 |
| 85 | Predicting diabetic nephropathy using a multifactorial genetic model. *PLoS ONE*, **2011**, 6, e18743 | 3.7 | 25 |
| 84 | Targeted demethylation at the CDKN1C/p57 locus induces human □cell replication. *Journal of Clinical Investigation*, **2019**, 129, 209-214 | 15.9 | 24 |
| 83 | Searching for type 2 diabetes genes on chromosome 20. *Diabetes*, **2002**, 51 Suppl 3, S308-15 | 0.9 | 23 |
| 82 | Growth-hormone-binding protein in patients with acromegaly. *Hormone Research*, **1992**, 37, 205-11 | | 23 |
| 81 | Effects of secretin on the normal and pathological beta-cell. *Journal of Clinical Endocrinology and Metabolism*, **1988**, 66, 1138-43 | 5.6 | 23 |
| 80 | Somatostatin receptor scintigraphy for early detection of regional and distant metastases of medullary carcinoma of the thyroid. *Clinical Nuclear Medicine*, **1999**, 24, 256-60 | 1.7 | 23 |
| 79 | Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179 | 8.2 | 22 |
| 78 | Ashkenazi Jewish mtDNA haplogroup distribution varies among distinct subpopulations: lessons of population substructure in a closed group. *European Journal of Human Genetics*, **2007**, 15, 498-500 | 5.3 | 22 |
| 77 | Compound heterozygosity for the common sulfonylurea receptor mutations can cause mild diazoxide-sensitive hyperinsulinism. *Clinical Pediatrics*, **2002**, 41, 183-6 | 1.2 | 22 |
| 76 | Gastrin-producing ovarian cystadenocarcinoma: sensitivity to secretin and SMS 201-995. *Gastroenterology*, **1989**, 97, 464-7 | 13.3 | 21 |
| 75 | Single-cell transcriptomics of human islet ontogeny defines the molecular basis of □cell dedifferentiation in T2D. *Molecular Metabolism*, **2020**, 42, 101057 | 8.8 | 21 |
| 74 | G0-G1 transition and the restriction point in pancreatic □cells in vivo. *Diabetes*, **2014**, 63, 578-84 | 0.9 | 20 |
| 73 | ABCC8 mutation allele frequency in the Ashkenazi Jewish population and risk of focal hyperinsulinemic hypoglycemia. *Genetics in Medicine*, **2011**, 13, 891-4 | 8.1 | 20 |
| 72 | Unusual causes of I-131 metaiodobenzylguanidine uptake in non-neural crest tissue. *Clinical Nuclear Medicine*, **1991**, 16, 239-42 | 1.7 | 20 |
| 71 | □Cell DNA Damage Response Promotes Islet Inflammation in Type 1 Diabetes. *Diabetes*, **2018**, 67, 2305-2318 | 3.9 | 20 |
| 70 | Thyroidectomy Practice After Implementation of the 2015 American Thyroid Association Guidelines on Surgical Options for Patients With Well-Differentiated Thyroid Carcinoma. *JAMA Otolaryngology - Head and Neck Surgery*, **2018**, 144, 427-432 | 3.9 | 19 |

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| 69 | Malaria and asymptomatic parasitaemia in Gabonese infants under the age of 3 months. *Acta Tropica*, **2005**, 95, 81-5 | 3.2 | 19 |
| 68 | A novel splice-site mutation in ECM-1 gene in a consanguineous family with lipoid proteinosis. *Experimental Dermatology*, **2005**, 14, 891-7 | 4 | 19 |
| 67 | Improvement of sleep apnoea due to acromegaly during short-term treatment with octreotide. *Journal of Internal Medicine*, **1994**, 236, 231-5 | 10.8 | 19 |
| 66 | Somatostatin-receptor imaging of medullary thyroid carcinoma. *Clinical Nuclear Medicine*, **1994**, 19, 416-21 | 2.7 | 19 |
| 65 | ChIP-seq of plasma cell-free nucleosomes identifies gene expression programs of the cells of origin. *Nature Biotechnology*, **2021**, 39, 586-598 | 44.5 | 19 |
| 64 | FOXP3 mutations causing early-onset insulin-requiring diabetes but without other features of immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Pediatric Diabetes*, **2018**, 19, 388-392 | 3.6 | 19 |
| 63 | Postnatal Exocrine Pancreas Growth by Cellular Hypertrophy Correlates with a Shorter Lifespan in Mammals. *Developmental Cell*, **2018**, 45, 726-737.e3 | 10.2 | 18 |
| 62 | Cells are not uniform after all-Novel insights into molecular heterogeneity of insulin-secreting cells. *Diabetes, Obesity and Metabolism*, **2017**, 19 Suppl 1, 147-152 | 6.7 | 18 |
| 61 | Hepatocyte nuclear factor 1alpha coding mutations are an uncommon contributor to early-onset type 2 diabetes in Ashkenazi Jews. *Diabetes*, **1998**, 47, 967-9 | 0.9 | 16 |
| 60 | Abrogation of Autophagy by Chloroquine Alone or in Combination with mTOR Inhibitors Induces Apoptosis in Neuroendocrine Tumor Cells. *Neuroendocrinology*, **2016**, 103, 724-37 | 5.6 | 15 |
| 59 | Secretion of pancreatic polypeptide in man in response to beef ingestion is mediated in part by an extravagal cholinergic mechanism. *Metabolism: Clinical and Experimental*, **1983**, 32, 57-61 | 12.7 | 15 |
| 58 | Type 2 diabetes: hypoinsulinism, hyperinsulinism, or both?. *PLoS Medicine*, **2007**, 4, e148 | 11.6 | 15 |
| 57 | Genetics of NIDDM in France: studies with 19 candidate genes in affected sib pairs. *Diabetes*, **1997**, 46, 1062-1068 | 0.9 | 15 |
| 56 | Distinct Imprinting Signatures and Biased Differentiation of Human Androgenetic and Parthenogenetic Embryonic Stem Cells. *Cell Stem Cell*, **2019**, 25, 419-432.e9 | 18 | 14 |
| 55 | Isolation and characterization of the human PAX4 gene. *Diabetes*, **1998**, 47, 1650-3 | 0.9 | 14 |
| 54 | 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. *American Journal of Medicine*, **1991**, 90, 37S-45S | 2.4 | 14 |
| 53 | Magnetic resonance imaging of the pituitary gland. *Clinical Radiology*, **1986**, 37, 9-14 | 2.9 | 14 |
| 52 | Relative expression of a dominant mutated ABCC8 allele determines the clinical manifestation of congenital hyperinsulinism. *Diabetes*, **2012**, 61, 258-63 | 0.9 | 13 |

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|---|---|---|---|
| 51 | Insulin mutations in diabetes: the clinical spectrum. *Diabetes*, **2008**, 57, 799-800 | 0.9 | 13 |
| 50 | Intragenic single nucleotide polymorphism haplotype analysis of SUR1 mutations in familial hyperinsulinism. *Human Mutation*, **1999**, 14, 23-9 | 4.7 | 13 |
| 49 | The expression of the beta cell-derived autoimmune ligand for the killer receptor nkp46 is attenuated in type 2 diabetes. *PLoS ONE*, **2013**, 8, e74033 | 3.7 | 13 |
| 48 | Sulfonylurea-responsive diabetes in childhood. *Journal of Pediatrics*, **2007**, 150, 553-5 | 3.6 | 12 |
| 47 | Growth hormone reserve in adult beta thalassemia patients. *Endocrine*, **2007**, 31, 33-7 | | 12 |
| 46 | p57Kip2 (cdkn1c): sequence, splice variants and unique temporal and spatial expression pattern in the rat pancreas. *Laboratory Investigation*, **2005**, 85, 364-75 | 5.9 | 12 |
| 45 | Multiplexing DNA methylation markers to detect circulating cell-free DNA derived from human pancreatic  cells. *JCI Insight*, **2020**, 5, | 9.9 | 12 |
| 44 | In-hospital treatment of hyperglycemia: effects of intensified subcutaneous insulin treatment. *Current Medical Research and Opinion*, **2007**, 23, 757-65 | 2.5 | 11 |
| 43 | Gallbladder visualization with In-111 labeled octreotide. *Clinical Nuclear Medicine*, **1994**, 19, 133-5 | 1.7 | 11 |
| 42 | Circulating Unmethylated Insulin DNA As a Biomarker of Human Beta Cell Death: A Multi-laboratory Assay Comparison. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105, | 5.6 | 11 |
| 41 | Isolation and characterization of the human AKT1 gene, identification of 13 single nucleotide polymorphisms (SNPs), and their lack of association with Type II diabetes. *Diabetologia*, **2001**, 44, 910-3 | 10.3 | 10 |
| 40 | Normal proinsulin processing despite beta-cell dysfunction in persistent hyperinsulinaemic hypoglycaemia of infancy (nesidioblastosis). *Diabetologia*, **1996**, 39, 1338-44 | 10.3 | 9 |
| 39 | Recurrence-associated mortality in patients with differentiated thyroid carcinoma. *Journal of Surgical Oncology*, **1993**, 52, 164-8 | 2.8 | 9 |
| 38 | Clinical and molecular heterogeneity of familial hyperinsulinism. *Journal of Pediatrics*, **1998**, 133, 801-2 | 3.6 | 8 |
| 37 | Dominant SUR1 mutation causing autosomal dominant type 2 diabetes. *Lancet, The*, **2003**, 361, 272-3 | 40 | 8 |
| 36 | Glycosylated serum protein levels assayed with highly sensitive immunoradiometric assay accurately reflect glycemic control of diabetic patients. *Diabetes Care*, **1992**, 15, 645-50 | 14.6 | 7 |
| 35 | Sodium valproate and metyrapone for pituitary-dependent Cushing's disease. *Lancet, The*, **1984**, 2, 640 | 40 | 7 |
| 34 | Lessons in human biology from a monogenic pancreatic  cell disease. *Journal of Clinical Investigation*, **2011**, 121, 3821-5 | 15.9 | 7 |

| | | | |
|---|---|---|---|
| 33 | Diagnosis of ABCC8 Congenital Hyperinsulinism of Infancy in a 20-Year-Old Man Evaluated for Factitious Hypoglycemia. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 345-349 | 5.6 | 7 |
| 32 | Genetic analysis of complex disease--a roadmap to understanding or a colossal waste of money. *Pediatric Endocrinology Reviews*, **2010**, 7, 258-65 | 1.1 | 7 |
| 31 | IGF-1 levels may increase paradoxically with dopamine agonist treatment for prolactinomas. *Pituitary*, **2018**, 21, 406-413 | 4.3 | 6 |
| 30 | Uncontrolled insulin secretion from a childhood pancreatic beta-cell adenoma is not due to the functional loss of ATP-sensitive potassium channels. *Endocrine-Related Cancer*, **2002**, 9, 221-6 | 5.7 | 6 |
| 29 | Sebum measurements for rapid identification of hyperandrogenism due to an ovarian Leydig cell tumor. *International Journal of Dermatology*, **1991**, 30, 276-7 | 1.7 | 6 |
| 28 | ChIP-seq of plasma cell-free nucleosomes identifies cell-of-origin gene expression programs | | 6 |
| 27 | Genetic activation of □cell glucokinase in mice causes enhanced glucose-suppression of glucagon secretion during normal and diabetic states. *Molecular Metabolism*, **2021**, 49, 101193 | 8.8 | 6 |
| 26 | Biphasic dynamics of beta cell mass in a mouse model of congenital hyperinsulinism: implications for type 2 diabetes. *Diabetologia*, **2021**, 64, 1133-1143 | 10.3 | 6 |
| 25 | Sex difference in the sensitivity of the human pancreatic polypeptide cell to autonomic nervous stimulation in man. *Journal of Clinical Endocrinology and Metabolism*, **1983**, 56, 21-5 | 5.6 | 5 |
| 24 | Clustering of Type 2 Diabetes Genetic Loci by Multi-Trait Associations Identifies Disease Mechanisms and Subtypes | | 5 |
| 23 | Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications*, **2021**, 12, 3505 | 17.4 | 5 |
| 22 | Long-term outcomes in MEN-1 patients with pancreatic neuroendocrine neoplasms: an Israeli specialist center experience. *Endocrine*, **2020**, 68, 222-229 | 4 | 4 |
| 21 | TSH producing pituitary tumor: biochemical diagnosis and long-term medical management with octreotide. *Hormone and Metabolic Research*, **1992**, 24, 34-8 | 3.1 | 4 |
| 20 | What is a □cell? - Chapter I in the Human Islet Research Network (HIRN) review series. *Molecular Metabolism*, **2021**, 53, 101323 | 8.8 | 4 |
| 19 | Lipid cell tumor of the ovary: steroid hormone secretory pattern and localization using 75Se-selenomethylcholesterol. *Gynecologic and Obstetric Investigation*, **1989**, 27, 110-2 | 2.5 | 3 |
| 18 | Effect of 6 months' gliclazide treatment on insulin release and sensitivity to endogenous insulin in NIDDM: role of initial CSII-induced normoglycemia. *Diabetes Research and Clinical Practice*, **1991**, 14 Suppl 2, S69-78 | 7.4 | 3 |
| 17 | Effect of acute cimetidine administration on indices of parathyroid hormone action in healthy subjects and patients with primary and secondary hyperparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **1984**, 59, 993-7 | 5.6 | 3 |
| 16 | Leydig-cell tumor of the ovary: visualization using 131I-19-iodocholesterol scintigraphy. *European Journal of Nuclear Medicine and Molecular Imaging*, **1985**, 11, 13-6 | | 3 |

| | | | |
|---|---|---|---|
| 15 | Liquid biopsy reveals collateral tissue damage in cancer.. *JCI Insight*, **2022**, 7, | 9.9 | 3 |
| 14 | Remote immune processes revealed by immune-derived circulating cell-free DNA. *ELife*, **2021**, 10, | 8.9 | 3 |
| 13 | The role of ATP-sensitive K+ channels in familial hyperinsulinism **2000**, 299-325 | | 2 |
| 12 | L-Thyroxine-induced leukopenia in a patient with Hashimoto's disease: involvement of suppressor-cytotoxic T cells. *Journal of Clinical Endocrinology and Metabolism*, **1985**, 61, 980-2 | 5.6 | 2 |
| 11 | Do cyclic AMP concentrations in saliva reflect PTH biologic activity?. *Metabolism: Clinical and Experimental*, **1985**, 34, 505-8 | 12.7 | 2 |
| 10 | Insights into the genetic epidemiology of Crohn and rare diseases in the Ashkenazi Jewish population | | 2 |
| 9 | Rare coding variants in 35 genes associate with circulating lipid levels a multi-ancestry analysis of 170,000 exomes | | 2 |
| 8 | Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries | | 2 |
| 7 | Specific detection of cell-free DNA derived from intestinal epithelial cells using methylation patterns | | 2 |
| 6 | Sequencing of over 100,000 individuals identifies multiple genes and rare variants associated with Crohns disease susceptibility | | 2 |
| 5 | A spot sample test for the estimation of urinary or nephrogenous cAMP in the evaluation of parathyroid function. *Nephron*, **1983**, 35, 264-6 | 3.3 | 1 |
| 4 | Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes | | 1 |
| 3 | Pendred Syndrome **2006**, 11, 154-168 | | |
| 2 | Cellular immune functions in patients with primary hyperparathyroidism: effects of histamine and cimetidine. *Immunopharmacology*, **1985**, 9, 81-6 | | |
| 1 | NCMP-01. NOVEL BIOMARKERS FOR RADIATION-INDUCED NEUROTOXICITY. *Neuro-Oncology*, **2021** , 23, vi147-vi147 | 1 | |