Sarah E Flanagan

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.

130 papers	5,686 citations	39 h-index	74 g-index
138 ext. papers	6,669 ext. citations	7.2 avg, IF	5.2 L-index

#	Paper	IF	Citations
130	Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. *Diabetologia*, **2022**, 65, 246-249	10.3	
129	Molecular Characterization and Management of Congenital Hyperinsulinism: A Tertiary Centre Experience. *Indian Pediatrics*, **2022**, 59, 105-109	1.2	1
128	Variation in Glycemic Outcomes in Focal Forms of Congenital Hyperinsulinism-The UK Perspective.. *Journal of the Endocrine Society*, **2022**, 6, bvac033	0.4	1
127	SavvyCNV: Genome-wide CNV calling from off-target reads.. *PLoS Computational Biology*, **2022**, 18, e1009940	3	1
126	Genetic Etiology of Neonatal Diabetes Mellitus in Vietnamese Infants and Characteristics of Those With Gene Mutations.. *Frontiers in Endocrinology*, **2022**, 13, 866573	5.7	
125	Congenital beta cell defects are not associated with markers of islet autoimmunity, even in the context of high genetic risk for type 1 diabetes.. *Diabetologia*, **2022**, 1	10.3	0
124	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. *Diabetologia*, **2021**, 1	10.3	1
123	Octreotide-LAR is a Useful Alternative for the Management of Diazoxide-Responsive Congenital Hyperinsulinism. *Hormone and Metabolic Research*, **2021**, 53, 723-729	3.1	1
122	Sex-biased islet cell dysfunction is caused by the MODY MAFA S64F variant by inducing premature aging and senescence in males. *Cell Reports*, **2021**, 37, 109813	10.6	3
121	Birth weight and diazoxide unresponsiveness strongly predict the likelihood of congenital hyperinsulinism due to a mutation in ABCC8 or KCNJ11. *European Journal of Endocrinology*, **2021**, 185, 813-818	6.5	1
120	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). *Molecular Genetics & Genomic Medicine*, **2021**, e1674	2.3	0
119	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. *Cell Reports*, **2021**, 35, 108981	10.6	4
118	Identification of GCK-maturity-onset diabetes of the young in cases of neonatal hyperglycemia: A case series and review of clinical features. *Pediatric Diabetes*, **2021**, 22, 876-881	3.6	2
117	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated Permanent Neonatal Diabetes. *Diabetes Care*, **2021**, 44, 35-42	14.6	7
116	Two decades since the fetal insulin hypothesis: what have we learned from genetics?. *Diabetologia*, **2021**, 64, 717-726	10.3	3
115	Genetic and clinical heterogeneity of permanent neonatal diabetes mellitus: a single tertiary centre experience. *Acta Diabetologica*, **2021**, 58, 1689-1700	3.9	0
114	Molecular Genetics, Clinical Characteristics, and Treatment Outcomes of K-Channel Neonatal Diabetes Mellitus in Vietnam National Children Hospital. *Frontiers in Endocrinology*, **2021**, 12, 727083	5.7	3

113	Hyperinsulinemic hypoglycemia in children and adolescents: Recent advances in understanding of pathophysiology and management. *Reviews in Endocrine and Metabolic Disorders*, **2020**, 21, 577-597	10.5	19
112	A Novel Mutation Causing Three Phenotypic Forms of Glucose Dysregulation in a Family. *Frontiers in Pediatrics*, **2020**, 8, 320	3.4	2
111	Longitudinal Auxological recovery in a cohort of children with Hyperinsulinaemic Hypoglycaemia. *Orphanet Journal of Rare Diseases*, **2020**, 15, 162	4.2	4
110	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389. *PLoS ONE*, **2020**, 15, e0228417	3.7	11
109	Update of variants identified in the pancreatic β-cell K channel genes KCNJ11 and ABCC8 in individuals with congenital hyperinsulinism and diabetes. *Human Mutation*, **2020**, 41, 884-905	4.7	37
108	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. *Journal of Clinical Investigation*, **2020**, 130, 6338-6353	15.9	21
107	Heterozygous Insulin Receptor Mutation Associated with Neonatal Hyperinsulinemic Hypoglycaemia and Familial Diabetes Mellitus: Case Series. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2020**, 12, 420-426	1.9	2
106	Unravelling the genetic causes of mosaic islet morphology in congenital hyperinsulinism. *Journal of Pathology: Clinical Research*, **2020**, 6, 12-16	5.3	12
105	De Novo Mutations in Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. *Diabetes*, **2020**, 69, 477-483	0.9	17
104	Type 1 diabetes can present before the age of 6 months and is characterised by autoimmunity and rapid loss of beta cells. *Diabetologia*, **2020**, 63, 2605-2615	10.3	9
103	Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	6
102	Clinical Characteristics, Molecular Features, and Long-Term Follow-Up of 15 Patients with Neonatal Diabetes: A Single-Centre Experience. *Hormone Research in Paediatrics*, **2020**, 93, 423-432	3.3	2
101	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389 **2020**, 15, e0228417		
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98	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389 **2020**, 15, e0228417		
97	Congenital Hyperinsulinemic Hypoglycemia and Hyperammonemia due to Pathogenic Variants in GLUD1. *Indian Journal of Pediatrics*, **2019**, 86, 1051-1053	3	1
96	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. *American Journal of Human Genetics*, **2019**, 104, 985-989	11	28

95	Genetic Mechanisms Highlight Shared Pathways for the Pathogenesis of Polygenic Type 1 Diabetes and Monogenic Autoimmune Diabetes. *Current Diabetes Reports*, **2019**, 19, 20	5.6	12
94	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. *Diabetes*, **2019**, 68, 1528-1535	0.9	15
93	A rare case of congenital hyperinsulinism (CHI) due to dual genetic aetiology involving HNF4A and ABCC8. *Journal of Pediatric Endocrinology and Metabolism*, **2019**, 32, 301-304	1.6	2
92	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. *Endocrinology, Diabetes and Metabolism Case Reports*, **2019**, 2019,	1.4	2
91	Congenital Hyperinsulinism and Evolution to Sulfonylurearesponsive Diabetes Later in Life due to a Novel Homozygous p.L171F Mutation. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2019**, 11, 82-87	1.9	10
90	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
89	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
88	Hyperinsulinaemic hypoglycaemia: A new presentation of 16p11.2 deletion syndrome. *Clinical Endocrinology*, **2019**, 90, 766-769	3.4	7
87	Patterns of postmeal insulin secretion in individuals with sulfonylurea-treated neonatal diabetes show predominance of non-K-channel pathways. *BMJ Open Diabetes Research and Care*, **2019**, 7, e000721	4.5	4
86	Identification of novel variants in neonatal diabetes mellitus genes in Egyptian patients with permanent NDM. *International Journal of Diabetes in Developing Countries*, **2019**, 39, 53-59	0.8	
85	Cognitive, Neurological, and Behavioral Features in Adults With Neonatal Diabetes. *Diabetes Care*, **2019**, 42, 215-224	14.6	11
84	Focal Congenital Hyperinsulinism as a Cause for Sudden Infant Death. *Pediatric and Developmental Pathology*, **2019**, 22, 65-69	2.2	4
83	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. *Diabetologia*, **2018**, 61, 1027-1036	10.3	17
82	missense mutation causes familial insulinomatosis and diabetes mellitus. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 1027-1032	11.5	45
81	A type 1 diabetes genetic risk score can discriminate monogenic autoimmunity with diabetes from early-onset clustering of polygenic autoimmunity with diabetes. *Diabetologia*, **2018**, 61, 862-869	10.3	20
80	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 195-204	1.6	8
79	Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation in NEUROD1. *Pediatric Diabetes*, **2018**, 19, 898-904	3.6	14
78	Diazoxide toxicity in a child with persistent hyperinsulinemic hypoglycemia of infancy: mixed hyperglycemic hyperosmolar coma and ketoacidosis. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 943-945	1.6	6

77	Comprehensive screening shows that mutations in the known syndromic genes are rare in infants presenting with hyperinsulinaemic hypoglycaemia. *Clinical Endocrinology*, **2018**, 89, 621-627	3.4	4
76	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. *Lancet Diabetes and Endocrinology,the*, **2018**, 6, 637-646	18.1	77
75	Sirolimus-Induced Hepatitis in Two Patients with Hyperinsulinemic Hypoglycemia. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2018**, 10, 279-283	1.9	5
74	Monogenic Diabetes Not Caused By Mutations in Mody Genes: A Very Heterogenous Group of Diabetes. *Experimental and Clinical Endocrinology and Diabetes*, **2018**, 126, 612-618	2.3	5
73	Clinical Diversity in Focal Congenital Hyperinsulinism in Infancy Correlates With Histological Heterogeneity of Islet Cell Lesions. *Frontiers in Endocrinology*, **2018**, 9, 619	5.7	9
72	First case of neonatal diabetes with KCNJ11 Q52R mutation successfully switched from insulin to sulphonylurea treatment. *Journal of Diabetes Investigation*, **2017**, 8, 716-719	3.9	8
71	Dominant ER Stress-Inducing Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. *Diabetes*, **2017**, 66, 2044-2053	0.9	56
70	Recessively Inherited Mutations Cause Autoimmunity Presenting as Neonatal Diabetes. *Diabetes*, **2017**, 66, 2316-2322	0.9	39
69	Atypical Forms of Congenital Hyperinsulinism in Infancy Are Associated With Mosaic Patterns of Immature Islet Cells. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 3261-3267	5.6	21
68	Clinical presentation and treatment response to diazoxide in two siblings with congenital hyperinsulinism as a result of a novel compound heterozygous ABCC8 missense mutation. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 471-474	1.6	1
67	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 2529-2539	12.7	73
66	A CACNA1D mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. *Pediatric Diabetes*, **2017**, 18, 320-323	3.6	46
65	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. *Nature Communications*, **2017**, 8, 888	17.4	57
64	Analysis of large-scale sequencing cohorts does not support the role of variants in UCP2 as a cause of hyperinsulinaemic hypoglycaemia. *Human Mutation*, **2017**, 38, 1442-1444	4.7	15
63	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. *Diabetologia*, **2017**, 60, 2168-2173	10.3	7
62	Case report: maternal mosaicism resulting in inheritance of a novel GATA6 mutation causing pancreatic agenesis and neonatal diabetes mellitus. *Diagnostic Pathology*, **2017**, 12, 1	3	20
61	An Nonsense Mutation Causing Neonatal Diabetes Through Altered Transcript Expression. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2017**, 9, 260-264	1.9	10
60	Prematurity and Genetic Testing for Neonatal Diabetes. *Pediatrics*, **2016**, 138,	7.4	19

59	Conservatively treated Congenital Hyperinsulinism (CHI) due to K-ATP channel gene mutations: reducing severity over time. *Orphanet Journal of Rare Diseases*, **2016**, 11, 163	4.2	32
58	Genetic characteristics, clinical spectrum, and incidence of neonatal diabetes in the Emirate of AbuDhabi, United Arab Emirates. *American Journal of Medical Genetics, Part A*, **2016**, 170, 602-9	2.5	26
57	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. *Diabetes*, **2016**, 65, 2810-5	0.9	16
56	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. *Diabetologia*, **2016**, 59, 1162-6	10.3	54
55	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. *Diabetes*, **2016**, 65, 2094-2099	0.9	105
54	Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in ATP sensitivity markedly affect diabetes risk. *Diabetologia*, **2016**, 59, 1430-1436	10.3	18
53	Clinical and genetic features of Argentinian children with diabetes-onset before 12months of age: Successful transfer from insulin to oral sulfonylurea. *Diabetes Research and Clinical Practice*, **2016**, 117, 104-10	7.4	7
52	Monogenic autoimmune diseases of the endocrine system. *Lancet Diabetes and Endocrinology,the*, **2016**, 4, 862-72	18.1	19
51	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. *Lancet, The*, **2015**, 386, 957-63	40	186
50	Clinical characteristics and molecular genetic analysis of 22 patients with neonatal diabetes from the South-Eastern region of Turkey: predominance of non-KATP channel mutations. *European Journal of Endocrinology*, **2015**, 172, 697-705	6.5	40
49	Genotype and phenotype correlations in Iranian patients with hyperinsulinaemic hypoglycaemia. *BMC Research Notes*, **2015**, 8, 350	2.3	6
48	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. *European Journal of Human Genetics*, **2015**, 23, 1744-8	5.3	24
47	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1279-86	1.6	25
46	Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 345-51	1.6	6
45	Anemia in a Child with Deafness: Be Vigilant for a Rare Cause!. *Indian Journal of Hematology and Blood Transfusion*, **2015**, 31, 394-5	0.7	
44	Increased plasma incretin concentrations identifies a subset of patients with persistent congenital hyperinsulinism without KATP channel gene defects. *Journal of Pediatrics*, **2015**, 166, 191-4	3.6	7
43	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. *Nature Genetics*, **2014**, 46, 61-64	36.3	187
42	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. *Nature Genetics*, **2014**, 46, 812-814	36.3	328

41	Permanent neonatal diabetes misdiagnosed as type 1 diabetes in a 28-year-old female: a life-changing diagnosis. *Diabetes Research and Clinical Practice*, **2014**, 106, e22-4	7.4	1
40	Sirolimus therapy in infants with severe hyperinsulinemic hypoglycemia. *New England Journal of Medicine*, **2014**, 370, 1131-7	59.2	97
39	Analysis of transcription factors key for mouse pancreatic development establishes NKX2-2 and MNX1 mutations as causes of neonatal diabetes in man. *Cell Metabolism*, **2014**, 19, 146-54	24.6	102
38	Long-term follow-up of children with congenital hyperinsulinism on octreotide therapy. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3660-7	5.6	42
37	Pancreatic endocrine and exocrine function in children following near-total pancreatectomy for diffuse congenital hyperinsulinism. *PLoS ONE*, **2014**, 9, e98054	3.7	50
36	Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability. *Human Molecular Genetics*, **2014**, 23, 6432-40	5.6	30
35	GATA4 mutations are a cause of neonatal and childhood-onset diabetes. *Diabetes*, **2014**, 63, 2888-94	0.9	80
34	Next-generation sequencing reveals deep intronic cryptic ABCC8 and HADH splicing founder mutations causing hyperinsulinism by pseudoexon activation. *American Journal of Human Genetics*, **2013**, 92, 131-6	11	66
33	Clinical and molecular characterisation of 300 patients with congenital hyperinsulinism. *European Journal of Endocrinology*, **2013**, 168, 557-64	6.5	147
32	GATA6 mutations cause a broad phenotypic spectrum of diabetes from pancreatic agenesis to adult-onset diabetes without exocrine insufficiency. *Diabetes*, **2013**, 62, 993-7	0.9	104
31	Partial ABCC8 gene deletion mutations causing diazoxide-unresponsive hyperinsulinaemic hypoglycaemia. *Pediatric Diabetes*, **2012**, 13, 285-9	3.6	8
30	KATP channel mutations in infants with permanent diabetes diagnosed after 6 months of life. *Pediatric Diabetes*, **2012**, 13, 322-5	3.6	49
29	Congenital hyperinsulinism: marked clinical heterogeneity in siblings with identical mutations in the ABCC8 gene. *Clinical Endocrinology*, **2012**, 76, 312-3	3.4	8
28	The heterogeneity of focal forms of congenital hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E94-9	5.6	25
27	649 A Case of Permanent Neonatal Diabetes Mellitus. *Archives of Disease in Childhood*, **2012**, 97, A188-A188		
26	GATA6 haploinsufficiency causes pancreatic agenesis in humans. *Nature Genetics*, **2011**, 44, 20-22	36.3	195
25	Clinical characteristics of recessive and dominant congenital hyperinsulinism due to mutation(s) in the ABCC8/KCNJ11 genes encoding the ATP-sensitive potasium channel in the pancreatic beta cell. *Journal of Pediatric Endocrinology and Metabolism*, **2011**, 24, 1019-23	1.6	13
24	Genetics of congenital hyperinsulinemic hypoglycemia. *Seminars in Pediatric Surgery*, **2011**, 20, 13-7	2.1	48

23	Dominantly acting ABCC8 mutations in patients with medically unresponsive hyperinsulinaemic hypoglycaemia. *Clinical Genetics*, **2011**, 79, 582-7	4	37
22	Dysmyelination of the cerebral white matter with microdeletion at 6p25. *Indian Pediatrics*, **2011**, 48, 727-9	1.2	10
21	Genome-wide homozygosity analysis reveals HADH mutations as a common cause of diazoxide-responsive hyperinsulinemic-hypoglycemia in consanguineous pedigrees. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E498-502	5.6	44
20	Characterization of ABCC8 and KCNJ11 gene mutations and phenotypes in Korean patients with congenital hyperinsulinism. *European Journal of Endocrinology*, **2011**, 164, 919-26	6.5	24
19	In vitro recovery of ATP-sensitive potassium channels in β-cells from patients with congenital hyperinsulinism of infancy. *Diabetes*, **2011**, 60, 1223-8	0.9	16
18	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. *Pediatric Diabetes*, **2010**, 11, 18-23	3.6	42
17	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 3105-10	11.5	149
16	Using SIFT and PolyPhen to predict loss-of-function and gain-of-function mutations. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 533-7	1.6	247
15	3-Hydroxyacyl-coenzyme A dehydrogenase deficiency and hyperinsulinemic hypoglycemia: characterization of a novel mutation and severe dietary protein sensitivity. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 2221-5	5.6	64
14	Tooth discoloration in patients with neonatal diabetes after transfer onto glibenclamide: a previously unreported side effect. *Diabetes Care*, **2009**, 32, 1428-30	14.6	35
13	Wolcott-Rallison syndrome is the most common genetic cause of permanent neonatal diabetes in consanguineous families. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4162-70	5.6	103
12	Update of mutations in the genes encoding the pancreatic beta-cell K(ATP) channel subunits Kir6.2 (KCNJ11) and sulfonylurea receptor 1 (ABCC8) in diabetes mellitus and hyperinsulinism. *Human Mutation*, **2009**, 30, 170-80	4.7	181
11	Effective treatment with oral sulfonylureas in patients with diabetes due to sulfonylurea receptor 1 (SUR1) mutations. *Diabetes Care*, **2008**, 31, 204-9	14.6	203
10	An ABCC8 gene mutation and mosaic uniparental isodisomy resulting in atypical diffuse congenital hyperinsulinism. *Diabetes*, **2008**, 57, 259-63	0.9	49
9	Identification of mutations in the Kir6.2 subunit of the K(ATP) channel. *Methods in Molecular Biology*, **2008**, 491, 235-45	1.4	1
8	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. *Diabetes*, **2007**, 56, 1930-7	0.9	273
7	Origin of de novo KCNJ11 mutations and risk of neonatal diabetes for subsequent siblings. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 1773-7	5.6	46
6	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. *American Journal of Human Genetics*, **2007**, 81, 375-82	11	161

5	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. *Human Molecular Genetics*, **2006**, 15, 1793-800	5.6	175
4	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. *New England Journal of Medicine*, **2006**, 355, 467-77	59.2	740
3	Neonatal diabetes is more than just a paediatric problem: 57 years of diabetes from a Kir6.2 mutation. *Practical Diabetes International: the International Journal for Diabetes Care Teams Worldwide*, **2005**, 22, 342-344		2
2	High-dose glibenclamide can replace insulin therapy despite transitory diarrhea in early-onset diabetes caused by a novel R201L Kir6.2 mutation. *Diabetes Care*, **2005**, 28, 758-9	14.6	75
1	SavvyCNV: genome-wide CNV calling from off-target reads		8