Sarah E Flanagan

List of Publications by Citations

Source: https://exaly.com/author-pdf/6544667/sarah-e-flanagan-publications-by-citations.pdf
Version: 2024-04-28

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.
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	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
129	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. *Nature Genetics*, **2014**, 46, 812-814	36.3	328
128	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
127	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
126	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
125	GATA6 haploinsufficiency causes pancreatic agenesis in humans. *Nature Genetics*, **2011**, 44, 20-22	36.3	195
124	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. *Nature Genetics*, **2014**, 46, 61-64	36.3	187
123	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
122	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
121	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. *Human Molecular Genetics*, **2006**, 15, 1793-800	5.6	175
120	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
119	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
118	Clinical and molecular characterisation of 300 patients with congenital hyperinsulinism. *European Journal of Endocrinology*, **2013**, 168, 557-64	6.5	147
117	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. *Diabetes*, **2016**, 65, 2094-2099	0.9	105
116	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
115	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
114	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

113	Sirolimus therapy in infants with severe hyperinsulinemic hypoglycemia. *New England Journal of Medicine*, **2014**, 370, 1131-7	59.2	97
112	GATA4 mutations are a cause of neonatal and childhood-onset diabetes. *Diabetes*, **2014**, 63, 2888-94	0.9	80
111	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
110	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
109	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
108	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
107	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
106	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. *Nature Communications*, **2017**, 8, 888	17.4	57
105	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
104	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
103	Pancreatic endocrine and exocrine function in children following near-total pancreatectomy for diffuse congenital hyperinsulinism. *PLoS ONE*, **2014**, 9, e98054	3.7	50
102	KATP channel mutations in infants with permanent diabetes diagnosed after 6 months of life. *Pediatric Diabetes*, **2012**, 13, 322-5	3.6	49
101	An ABCC8 gene mutation and mosaic uniparental isodisomy resulting in atypical diffuse congenital hyperinsulinism. *Diabetes*, **2008**, 57, 259-63	0.9	49
100	Genetics of congenital hyperinsulinemic hypoglycemia. *Seminars in Pediatric Surgery*, **2011**, 20, 13-7	2.1	48
99	A CACNA1D mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. *Pediatric Diabetes*, **2017**, 18, 320-323	3.6	46
98	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
97	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
96	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

95	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
94	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. *Pediatric Diabetes*, **2010**, 11, 18-23	3.6	42
93	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
92	Recessively Inherited Mutations Cause Autoimmunity Presenting as Neonatal Diabetes. *Diabetes*, **2017**, 66, 2316-2322	0.9	39
91	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
90	Dominantly acting ABCC8 mutations in patients with medically unresponsive hyperinsulinaemic hypoglycaemia. *Clinical Genetics*, **2011**, 79, 582-7	4	37
89	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
88	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
87	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
86	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
85	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
84	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1279-86	1.6	25
83	The heterogeneity of focal forms of congenital hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E94-9	5.6	25
82	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
81	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
80	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
79	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. *Journal of Clinical Investigation*, **2020**, 130, 6338-6353	15.9	21
78	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

77	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
76	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
75	Prematurity and Genetic Testing for Neonatal Diabetes. *Pediatrics*, **2016**, 138,	7.4	19
74	Monogenic autoimmune diseases of the endocrine system. *Lancet Diabetes and Endocrinology,the*, **2016**, 4, 862-72	18.1	19
73	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
72	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. *Diabetologia*, **2018**, 61, 1027-1036	10.3	17
71	De Novo Mutations in Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. *Diabetes*, **2020**, 69, 477-483	0.9	17
70	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. *Diabetes*, **2016**, 65, 2810-5	0.9	16
69	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
68	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. *Diabetes*, **2019**, 68, 1528-1535	0.9	15
67	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
66	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
65	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
64	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
63	Unravelling the genetic causes of mosaic islet morphology in congenital hyperinsulinism. *Journal of Pathology: Clinical Research*, **2020**, 6, 12-16	5.3	12
62	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
61	Cognitive, Neurological, and Behavioral Features in Adults With Neonatal Diabetes. *Diabetes Care*, **2019**, 42, 215-224	14.6	11
60	Dysmyelination of the cerebral white matter with microdeletion at 6p25. *Indian Pediatrics*, **2011**, 48, 727-32	1.2	10

59	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
58	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
57	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
56	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
55	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
54	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 195-204	1.6	8
53	Partial ABCC8 gene deletion mutations causing diazoxide-unresponsive hyperinsulinaemic hypoglycaemia. *Pediatric Diabetes*, **2012**, 13, 285-9	3.6	8
52	Congenital hyperinsulinism: marked clinical heterogeneity in siblings with identical mutations in the ABCC8 gene. *Clinical Endocrinology*, **2012**, 76, 312-3	3.4	8
51	SavvyCNV: genome-wide CNV calling from off-target reads		8
50	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. *Diabetologia*, **2017**, 60, 2168-2173	10.3	7
49	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
48	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
47	Hyperinsulinaemic hypoglycaemia: A new presentation of 16p11.2 deletion syndrome. *Clinical Endocrinology*, **2019**, 90, 766-769	3.4	7
46	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
45	Genotype and phenotype correlations in Iranian patients with hyperinsulinaemic hypoglycaemia. *BMC Research Notes*, **2015**, 8, 350	2.3	6
44	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
43	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
42	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

41	Sirolimus-Induced Hepatitis in Two Patients with Hyperinsulinemic Hypoglycemia. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2018**, 10, 279-283	1.9	5
40	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
39	Longitudinal Auxological recovery in a cohort of children with Hyperinsulinaemic Hypoglycaemia. *Orphanet Journal of Rare Diseases*, **2020**, 15, 162	4.2	4
38	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
37	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. *Cell Reports*, **2021**, 35, 108981	10.6	4
36	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
35	Focal Congenital Hyperinsulinism as a Cause for Sudden Infant Death. *Pediatric and Developmental Pathology*, **2019**, 22, 65-69	2.2	4
34	Sex-biased islet β 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
33	Two decades since the fetal insulin hypothesis: what have we learned from genetics?. *Diabetologia*, **2021**, 64, 717-726	10.3	3
32	Molecular Genetics, Clinical Characteristics, and Treatment Outcomes of K-Channel Neonatal Diabetes Mellitus in Vietnam National Children's Hospital. *Frontiers in Endocrinology*, **2021**, 12, 727083	5.7	3
31	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
30	A Novel Mutation Causing Three Phenotypic Forms of Glucose Dysregulation in a Family. *Frontiers in Pediatrics*, **2020**, 8, 320	3.4	2
29	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
28	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. *Endocrinology, Diabetes and Metabolism Case Reports*, **2019**, 2019,	1.4	2
27	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
26	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
25	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
24	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

23	Congenital Hyperinsulinemic Hypoglycemia and Hyperammonemia due to Pathogenic Variants in GLUD1. *Indian Journal of Pediatrics*, **2019**, 86, 1051-1053	3	1
22	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
21	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. *Diabetologia*, **2021**, 1	10.3	1
20	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
19	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
18	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
17	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
16	Identification of mutations in the Kir6.2 subunit of the K(ATP) channel. *Methods in Molecular Biology*, **2008**, 491, 235-45	1.4	1
15	Molecular Characterization and Management of Congenital Hyperinsulinism: A Tertiary Centre Experience. *Indian Pediatrics*, **2022**, 59, 105-109	1.2	1
14	Variation in Glycemic Outcomes in Focal Forms of Congenital Hyperinsulinism-The UK Perspective.. *Journal of the Endocrine Society*, **2022**, 6, bvac033	0.4	1
13	SavvyCNV: Genome-wide CNV calling from off-target reads.. *PLoS Computational Biology*, **2022**, 18, e1009940	3	1
12	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). *Molecular Genetics & Genomic Medicine*, **2021**, e1674	2.3	0
11	Genetic and clinical heterogeneity of permanent neonatal diabetes mellitus: a single tertiary centre experience. *Acta Diabetologica*, **2021**, 58, 1689-1700	3.9	0
10	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
9	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	
8	649 A Case of Permanent Neonatal Diabetes Mellitus. *Archives of Disease in Childhood*, **2012**, 97, A188-A188	2.8	
7	Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. *Diabetologia*, **2022**, 65, 246-249	10.3	
6	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	

List of Publications

5	Genetic Etiology of Neonatal Diabetes Mellitus in Vietnamese Infants and Characteristics of Those With Gene Mutations.. *Frontiers in Endocrinology*, **2022**, 13, 866573	5.7
4	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	
3	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	
2	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	
1	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	