Khalid Hussain

List of Publications by Year
in descending order

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138	6,262	76196	76769
papers	citations	40 h-index	74 g-index

139	139	139	5594
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	⁶⁸Ga-NODAGA-Exendin-4 PET/CT Improves the Detection of Focal Congenital Hyperinsulinism. Journal of Nuclear Medicine, 2022, 63, 310-315.	2.8	19
2	The epidemiology, clinical, biochemical, immunological and radiological features of youth onset type 2 diabetes mellitus in the state of Qatar. Diabetology International, 2022, 13, 381-386.	0.7	2
3	Homozygous Insulin Promotor Gene Mutation Causing Permanent Neonatal Diabetes Mellitus and Childhood Onset Autoantibody Negative Diabetes in the Same Family. International Medical Case Reports Journal, 2022, Volume 15, 35-41.	0.3	0
4	Infancy onset diabetes mellitus in a patient with a novel homozygous LRBA mutation. Journal of Clinical and Translational Endocrinology: Case Reports, 2022, 23, 100108.	0.4	1
5	Understanding the Mechanism of Diabetes Mellitus in a LRBA-Deficient Patient. Biology, 2022, 11, 612.	1.3	0
6	A Novel Homozygous MC2R Variant Leading to Type-1 Familial Glucocorticoid Deficiency. Journal of the Endocrine Society, 2022, 6, bvac058.	0.1	4
7	Type 2 Diabetes Mellitus in a 7 Year Old Girl. International Medical Case Reports Journal, 2022, Volume 15, 245-250.	0.3	2
8	Understanding the Mechanism of Dysglycemia in a Fanconi-Bickel Syndrome Patient. Frontiers in Endocrinology, 2022, 13, .	1.5	3
9	Sib-pair subgroup familial type 1 diabetes mellitus in children in the state of Qatar. PLoS ONE, 2022, 17, e0271182.	1.1	0
10	Aberrant development of pancreatic beta cells derived from human iPSCs with FOXA2 deficiency. Cell Death and Disease, 2021, 12, 103.	2.7	16
11	Distinctive Microbial Signatures and Gut-Brain Crosstalk in Pediatric Patients with Coeliac Disease and Type 1 Diabetes Mellitus. International Journal of Molecular Sciences, 2021, 22, 1511.	1.8	10
12	Identification of Three Novel and One Known Mutation in the WFS1 Gene in Four Unrelated Turkish Families: The Role of Homozygosity Mapping in the Early Diagnosis. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 34-43.	0.4	1
13	Congenital hyperinsulinism: 2 case reports with different rare variants in ABCC8. Annals of Pediatric Endocrinology and Metabolism, 2021, 26, 60-65.	0.8	2
14	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular Genetics & Genomic Medicine, 2021, , e1674.	0.6	2
15	Epidemiology, genetic landscape and classification of childhood diabetes mellitus in the State of Qatar. Journal of Diabetes Investigation, 2021, 12, 2141-2148.	1.1	8
16	The Epidemiology and Genetic Analysis of Children With Idiopathic Type 1 Diabetes in the State of Qatar. Journal of the Endocrine Society, 2021, 5, bvab131.	0.1	2
17	An induced pluripotent stem cell line derived from a patient with neonatal diabetes and Fanconi-Bickel syndrome caused by a homozygous mutation in the SLC2A2 gene. Stem Cell Research, 2021, 54, 102433.	0.3	0
18	The prevalence, immune profile, and clinical characteristics of children with celiac disease and type 1 diabetes mellitus in the state of Qatar. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1457-1461.	0.4	1

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19	Clinical features, epidemiology, autoantibody status, HLA haplotypes and genetic mechanisms of type 1 diabetes mellitus among children in Qatar. Scientific Reports, 2021, 11, 18887.	1.6	7
20	Maturityâ€onset diabetes of the young (MODY) due to PDX1 mutation in a sibâ€pair diabetes family from Qatar. Clinical Case Reports (discontinued), 2021, 9, e05141.	0.2	6
21	Corneal confocal microscopy demonstrates minimal evidence of distal neuropathy in children with celiac disease. PLoS ONE, 2020, 15, e0238859.	1.1	4
22	Generation of two human iPSC lines from patients with maturity-onset diabetes of the young type 2 (MODY2) and permanent neonatal diabetes due to mutations in the GCK gene. Stem Cell Research, 2020, 48, 101991.	0.3	5
23	Neonatal diabetes due to homozygous INS gene promoter mutations: Highly variable phenotype, remission and early relapse during the first 3â€‰years of life. Pediatric Diabetes, 2020, 21, 1169-1175.	1.2	6
24	Fanconiâ€“Bickel Syndrome: A Review of the Mechanisms That Lead to Dysglycaemia. International Journal of Molecular Sciences, 2020, 21, 6286.	1.8	26
25	Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4351-e4359.	1.8	10
26	Somatostatin analogues for the treatment of hyperinsulinaemic hypoglycaemia. Therapeutic Advances in Endocrinology and Metabolism, 2020, 11, 204201882096506.	1.4	14
27	Altered Serum Amino Acid and Acylcarnitine Profiles in Hyperinsulinemic Hypoglycemia and Ketotic Hypoglycemia. Frontiers in Endocrinology, 2020, 11, 577373.	1.5	3
28	Corneal nerve loss in children with typeÂ1 diabetes mellitus without retinopathy or microalbuminuria. Journal of Diabetes Investigation, 2020, 11, 1594-1601.	1.1	13
29	Derivation of a human induced pluripotent stem cell line (QBRIi007-A) from a patient carrying a homozygous intronic mutation (c.613-7T>G) in the SLC2A2 gene. Stem Cell Research, 2020, 44, 101736.	0.3	6
30	Haploinsufficiency of the FOXA2 associated with a complex clinical phenotype. Molecular Genetics & Genomic Medicine, 2020, 8, e1086.	0.6	6
31	Functional assessment of variants associated with Wolfram syndrome. Human Molecular Genetics, 2019, 28, 3815-3824.	1.4	10
32	The clinical and genetic characteristics of permanent neonatal diabetes (PNDM) in the state of Qatar. Molecular Genetics & Genomic Medicine, 2019, 7, e00753.	0.6	21
33	Ion Transporters, Channelopathies, and Glucose Disorders. International Journal of Molecular Sciences, 2019, 20, 2590.	1.8	21
34	A novel 3â€² untranslated region mutation in theSLC29A3gene associated with pigmentary hypertrichosis and nonâ€autoimmune insulinâ€dependent diabetes mellitus syndrome. Pediatric Diabetes, 2019, 20, 474-481.	1.2	4
35	The Genetic and Molecular Mechanisms of Congenital Hyperinsulinism. Frontiers in Endocrinology, 2019, 10, 111.	1.5	74
36	A Systematic Review of Childhood Diabetes Research in the Middle East Region. Frontiers in Endocrinology, 2019, 10, 805.	1.5	21

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37	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.	0.3	3
38	Congenital Hyperinsulinism and Evolution to Sulfonylurearesponsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 82-87.	0.4	18
39	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
40	Enteroinsular hormones in two siblings with Donohue syndrome and complete leptin deficiency. Pediatric Diabetes, 2018, 19, 675-679.	1.2	0
41	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.	0.4	7
42	The burden of congenital hyperinsulinism in the United Kingdom: a cost of illness study. Orphanet Journal of Rare Diseases, 2018, 13, 123.	1.2	15
43	Diagnosis and management of hyperinsulinaemic hypoglycaemia. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 551-573.	2.2	34
44	Hyperinsulinaemic hypoglycaemia in children and adults. Lancet Diabetes and Endocrinology,the, 2017, 5, 729-742.	5.5	89
45	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.	0.4	1
46	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.	3.0	99
47	Gastrointestinal dysmotility and pancreatic insufficiency in 2 siblings with Donohue syndrome. Pediatric Diabetes, 2017, 18, 839-843.	1.2	5
48	Tocilizumab for the Treatment of SLC29A3 Mutation Positive PHID Syndrome. Pediatrics, 2017, 140, .	1.0	27
49	Analysis of large-scale sequencing cohorts does not support the role of variants inUCP2as a cause of hyperinsulinaemic hypoglycaemia. Human Mutation, 2017, 38, 1442-1444.	1.1	17
50	Constitutive Activation of AKT2 in Humans Leads to Hypoglycemia Without Fatty Liver or Metabolic Dyslipidemia. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2914-2921.	1.8	15
51	Diagnosis and treatment of hyperinsulinaemic hypoglycaemia and its implications for paediatric endocrinology. International Journal of Pediatric Endocrinology (Springer), 2017, 2017, 9.	1.6	17
52	Congenital Hyperinsulinism: Diagnosis and Treatment Update. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 69-87.	0.4	91
53	Sirolimus precipitating diabetes mellitus in a patient with congenital hyperinsulinaemic hypoglycaemia due to autosomal dominant ABCC8 mutation. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1219-1222.	0.4	8
54	Coexistence of Mosaic Uniparental Isodisomy and a KCNJ11 Mutation Presenting as Diffuse Congenital Hyperinsulinism and Hemihypertrophy. Hormone Research in Paediatrics, 2016, 85, 426-427.	0.8	1

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55	Hyperinsulinemic Hypoglycemia â€“ The Molecular Mechanisms. Frontiers in Endocrinology, 2016, 7, 29.	1.5	53
56	Assessment of Nifedipine therapy in Hyperinsulinemic Hypoglycemia due to mutations in the ABCC8 gene.. Journal of Clinical Endocrinology and Metabolism, 2016, 102, jc.2016-2916.	1.8	20
57	Severe Hyperinsulinaemic Hypoglycaemia in Beckwith-Wiedemann Syndrome due to Paternal Uniparental Disomy of 11p15.5 Managed with Sirolimus Therapy. Hormone Research in Paediatrics, 2016, 85, 353-357.	0.8	10
58	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. Diabetes, 2016, 65, 2810-2815.	0.3	22
59	What is a normal blood glucose?. Archives of Disease in Childhood, 2016, 101, 569-574.	1.0	143
60	Sirolimus therapy in a child with partially diazoxide-responsive hyperinsulinaemic hypoglycaemia. Endocrinology, Diabetes and Metabolism Case Reports, 2016, 2016, .	0.2	3
61	A Novel Homozygous Mutation in the KCNJ11 Gene of a Neonate with Congenital Hyperinsulinism and Successful Management with Sirolimus. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 478-481.	0.4	17
62	High Incidence of HeterozygousABCC8andHNF1AMutations in Czech Patients With Congenital Hyperinsulinism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1540-E1549.	1.8	32
63	Molecular mechanisms of congenital hyperinsulinism and prospective therapeutic targets. Expert Opinion on Orphan Drugs, 2015, 3, 887-898.	0.5	2
64	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.	0.4	10
65	Recommendations from the Pediatric Endocrine Society for Evaluation andÂManagement of Persistent Hypoglycemia in Neonates, Infants, andÂChildren. Journal of Pediatrics, 2015, 167, 238-245.	0.9	431
66	Hyperinsulinemic Hypoglycemia. Pediatric Clinics of North America, 2015, 62, 1017-1036.	0.9	33
67	Molecular mechanisms of congenital hyperinsulinism due to autosomal dominant mutations inABCC8. Human Molecular Genetics, 2015, 24, 5142-5153.	1.4	26
68	Use of Long-Acting Somatostatin Analogue (Lanreotide) in an Adolescent with Diazoxide-Responsive Congenital Hyperinsulinism and Its Psychological Impact. Hormone Research in Paediatrics, 2015, 84, 355-360.	0.8	26
69	Molecular mechanisms of congenital hyperinsulinism. Journal of Molecular Endocrinology, 2015, 54, R119-R129.	1.1	62
70	Efficacy and safety of sirolimus in a neonate with persistent hypoglycaemia following near-total pancreatectomy for hyperinsulinaemic hypoglycaemia. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1391-8.	0.4	15
71	The Diagnosis and Management of Hyperinsulinaemic Hypoglycaemia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 86-97.	0.4	58
72	A novel homozygous SLC19A2 mutation in a Portuguese patient with diabetes mellitus and thiamine-responsive megaloblastic anaemia. International Journal of Pediatric Endocrinology (Springer), 2015, 2015, 6.	1.6	15

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73	Re-Evaluating â€œTransitional Neonatal Hypoglycemiaâ€• Mechanism and Implications for Management. Journal of Pediatrics, 2015, 166, 1520-1525.e1.	0.9	179
74	Sirolimus therapy in a patient with severe hyperinsulinaemic hypoglycaemia due to a compound heterozygous ABCC8 gene mutation. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 695-9.	0.4	20
75	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.	1.9	52
76	Genotype and phenotype correlations in Iranian patients with hyperinsulinaemic hypoglycaemia. BMC Research Notes, 2015, 8, 350.	0.6	10
77	Insulin Receptor and the Kidney: Nephrocalcinosis in Patients with Recessive INSR Mutations. Nephron Physiology, 2015, 128, 55-61.	1.5	18
78	Postprandial Hyperinsulinaemic Hypoglycaemia Secondary to a Congenital Portosystemic Shunt. Hormone Research in Paediatrics, 2015, 83, 217-220.	0.8	11
79	Protein-induced hyperinsulinaemic hypoglycaemia due to a homozygous HADH mutation in three siblings of a Saudi family. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1073-7.	0.4	19
80	Pancreatic Endocrine and Exocrine Function in Children following Near-Total Pancreatectomy for Diffuse Congenital Hyperinsulinism. PLoS ONE, 2014, 9, e98054.	1.1	63
81	Neonatal diabetes in an infant of diabetic mother: same novel INS missense mutation in the mother and her offspring. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 745-8.	0.4	4
82	Congenital hyperinsulinism: clinical and molecular characterisation of compound heterozygous ABCC8 mutation responsive to Diazoxide therapy. International Journal of Pediatric Endocrinology (Springer), 2014, 2014, 24.	1.6	15
83	Clinical and histological heterogeneity of congenital hyperinsulinism due to paternally inherited heterozygous ABCC8/KCNJ11 mutations. European Journal of Endocrinology, 2014, 171, 685-695.	1.9	36
84	Congenital hyperinsulinism. Medicina (Lithuania), 2014, 50, 190-195.	0.8	8
85	Neonatal Hypoglycemia. Indian Journal of Pediatrics, 2014, 81, 58-65.	0.3	19
86	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	9.4	255
87	Clinical characteristics and phenotypeâ€“genotype analysis in Turkish patients with congenital hyperinsulinism; predominance of recessive KATP channel mutations. European Journal of Endocrinology, 2014, 170, 885-892.	1.9	17
88	Insulinoma in childhood: clinical, radiological, molecular and histological aspects of nine patients. European Journal of Endocrinology, 2014, 170, 741-747.	1.9	27
89	Sirolimus Therapy in Infants with Severe Hyperinsulinemic Hypoglycemia. New England Journal of Medicine, 2014, 370, 1131-1137.	13.9	116
90	Persistent hyperinsulinaemic hypoglycaemia in infancy. Seminars in Pediatric Surgery, 2014, 23, 76-82.	0.5	32

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91	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, 146-154.	7.2	123
92	Long-Term Follow-Up of Children With Congenital Hyperinsulinism on Octreotide Therapy. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3660-3667.	1.8	53
93	Pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes mellitus (PHID) syndrome is associated with severe chronic inflammation and cardiomyopathy, and represents a new monogenic autoinflammatory syndrome. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 877-82.	0.4	31
94	Paradoxical Hypoglycaemia Associated with Diazoxide Therapy for Hyperinsulinaemic Hypoglycaemia. Hormone Research in Paediatrics, 2013, 80, 129-133.	0.8	9
95	Clinical and molecular characterisation of hyperinsulinaemic hypoglycaemia in infants born small-for-gestational age. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2013, 98, F356-F358.	1.4	44
96	Clinical and molecular characterisation of 300 patients with congenital hyperinsulinism. European Journal of Endocrinology, 2013, 168, 557-564.	1.9	190
97	An Evaluation of Growth Hormone and IGF-1 Responses in Neonates with Hyperinsulinaemic Hypoglycaemia. International Journal of Endocrinology, 2013, 2013, 1-5.	0.6	8
98	The molecular mechanisms, diagnosis and management of congenital hyperinsulinism. Indian Journal of Endocrinology and Metabolism, 2013, 17, 19.	0.2	48
99	Octreotide-Induced Long QT Syndrome in a Child with Congenital Hyperinsulinemia and a Novel Missense Mutation (p.Met115Val) in theABCC8Gene. Hormone Research in Paediatrics, 2013, 80, 299-303.	0.8	13
100	Heterogeneity in Phenotype of Usher-Congenital Hyperinsulinism Syndrome. Diabetes Care, 2013, 36, 557-561.	4.3	14
101	18F-DOPA PET and enhanced CT imaging for congenital hyperinsulinism. Nuclear Medicine Communications, 2013, 34, 601-608.	0.5	34
102	Paternally inherited ABCC8 mutation causing diffuse congenital hyperinsulinism. Endocrinology, Diabetes and Metabolism Case Reports, 2013, 2013, 130041.	0.2	7
103	Hyperinsulinaemic Hypoglycaemia: Genetic Mechanisms, Diagnosis and Management. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 169-181.	0.4	54
104	Hyperinsulinaemic hypoglycaemia: genetic mechanisms, diagnosis and management. Journal of Inherited Metabolic Disease, 2012, 35, 589-601.	1.7	116
105	Congenital hyperinsulinism: marked clinical heterogeneity in siblings with identical mutations in the ABCC8 gene. Clinical Endocrinology, 2012, 76, 312-313.	1.2	8
106	Recurrent spontaneous hypoglycaemia causes loss of neurogenic and neuroglycopaenic signs in infants with congenital hyperinsulinism. Clinical Endocrinology, 2012, 76, 548-554.	1.2	18
107	Incidence, genetics, and clinical phenotype of permanent neonatal diabetes mellitus in northwest Saudi Arabia. Pediatric Diabetes, 2012, 13, 499-505.	1.2	84
108	The predictive value of preoperative fluorine-18-l-3,4-dihydroxyphenylalanine positron emission tomographyâ€“computed tomography scans in children with congenital hyperinsulinism of infancy. Journal of Pediatric Surgery, 2011, 46, 204-208.	0.8	47

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109	Genetics of congenital hyperinsulinemic hypoglycemia. Seminars in Pediatric Surgery, 2011, 20, 13-17.	0.5	55
110	Investigations for neonatal hypoglycaemia. Clinical Biochemistry, 2011, 44, 465-466.	0.8	13
111	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-E502.	1.8	51
112	Characterization of ABCC8 and KCNJ11 gene mutations and phenotypes in Korean patients with congenital hyperinsulinism. European Journal of Endocrinology, 2011, 164, 919-926.	1.9	28
113	Pancreatic Î²-cell KATP channels: Hypoglycaemia and hyperglycaemia. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 157-163.	2.6	54
114	Mutations in pancreatic ÃŸ-cell Glucokinase as a cause of hyperinsulinaemic hypoglycaemia and neonatal diabetes mellitus. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 179-183.	2.6	26
115	Congenital hyperinsulinism and neonatal diabetes mellitus. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 155-156.	2.6	4
116	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, 3105-3110.	3.3	185
117	SLC29A3 gene is mutated in pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome and interacts with the insulin signaling pathway. Human Molecular Genetics, 2009, 18, 2257-2265.	1.4	100
118	Hyperinsulinismâ€“hyperammonaemia syndrome: novel mutations in the GLUD1 gene and genotypeâ€“phenotype correlations. European Journal of Endocrinology, 2009, 161, 731-735.	1.9	81
119	Wolcott-Rallison Syndrome Is the Most Common Genetic Cause of Permanent Neonatal Diabetes in Consanguineous Families. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4162-4170.	1.8	127
120	Diabetes mellitus, exocrine pancreatic deficiency, hypertrichosis, hyperpigmentation, and chronic inflammation: confirmation of a syndrome. Pediatric Diabetes, 2009, 10, 193-197.	1.2	25
121	Hyperinsulinism in Developmental Syndromes. Endocrine Development, 2009, 14, 95-113.	1.3	56
122	Diagnosis and Management of Hyperinsulinaemic Hypoglycaemia of Infancy. Hormone Research in Paediatrics, 2008, 69, 2-13.	0.8	76
123	Persistent Hyperinsulinemic Hypoglycemia and Maturity-Onset Diabetes of the Young Due to Heterozygous HNF4A Mutations. Diabetes, 2008, 57, 1659-1663.	0.3	133
124	An ABCC8 Gene Mutation and Mosaic Uniparental Isodisomy Resulting in Atypical Diffuse Congenital Hyperinsulinism. Diabetes, 2008, 57, 259-263.	0.3	58
125	Insulin Mutation Screening in 1,044 Patients With Diabetes. Diabetes, 2008, 57, 1034-1042.	0.3	347
126	Hyperinsulinaemic hypoglycaemia: biochemical basis and the importance of maintaining normoglycaemia during management. Archives of Disease in Childhood, 2007, 92, 568-570.	1.0	75

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127	Insights in Congenital Hyperinsulinism. , 2007, 11, 106-121.		18
128	Mosaic Turner Syndrome and Hyperinsulinaemic Hypoglycaemia. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 1451-7.	0.4	24
129	From congenital hyperinsulinism to diabetes mellitus: the role of pancreatic beta-cell KATP channels. Pediatric Diabetes, 2005, 6, 103-113.	1.2	21
130	Ketotic hypoglycaemia in children with diazoxide responsive hyperinsulinism of infancy. European Journal of Pediatrics, 2005, 164, 387-390.	1.3	6
131	Serum Glucagon Counterregulatory Hormonal Response to Hypoglycemia Is Blunted in Congenital Hyperinsulinism. Diabetes, 2005, 54, 2946-2951.	0.3	53
132	Hyperinsulinism of infancy associated with a novel splice site mutation in the SCHAD gene. Journal of Pediatrics, 2005, 146, 706-708.	0.9	68
133	Medications used in the treatment of hypoglycemia due to congenital hyperinsulinism of infancy (HI). Pediatric Endocrinology Reviews, 2004, 2 Suppl 1, 163-7.	1.2	12
134	A child presenting with disordered consciousness, hallucinations, screaming episodes and abdominal pain. European Journal of Pediatrics, 2002, 161, 127-129.	1.3	13
135	A child presenting with disordered consciousness, hallucinations, screaming episodes and abdominal pain. European Journal of Pediatrics, 2002, 161, 127-129.	1.3	14
136	Hyperinsulinism in short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency reveals the importance of l²-oxidation in insulin secretion. Journal of Clinical Investigation, 2001, 108, 457-465.	3.9	246
137	A recessive contiguous gene deletion causing infantile hyperinsulinism, enteropathy and deafness identifies the Usher type 1C gene. Nature Genetics, 2000, 26, 56-60.	9.4	307
138	Management of hyperinsulinism in infancy and childhood. Annals of Medicine, 2000, 32, 544-551.	1.5	41