Charles A Stanley

List of Publications by Citations

207 papers	12,637 citations	69 h-index	104 g-index
225 ext. papers	14,053 ext. citations	6.8 avg, IF	6.21 L-index

#	Paper	IF	Citations
207	Hyperinsulinism and hyperammonemia in infants with regulatory mutations of the glutamate dehydrogenase gene. *New England Journal of Medicine*, **1998**, 338, 1352-7	59.2	555
206	Familial hyperinsulinism caused by an activating glucokinase mutation. *New England Journal of Medicine*, **1998**, 338, 226-30	59.2	482
205	A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. *Nature*, **2007**, 448, 591-4	50.4	424
204	Recommendations from the Pediatric Endocrine Society for Evaluation and Management of Persistent Hypoglycemia in Neonates, Infants, and Children. *Journal of Pediatrics*, **2015**, 167, 238-45	3.6	285
203	Primary carnitine deficiency due to a failure of carnitine transport in kidney, muscle, and fibroblasts. *New England Journal of Medicine*, **1988**, 319, 1331-6	59.2	257
202	Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42	59.2	197
201	Long-chain acyl coenzyme A dehydrogenase deficiency: an inherited cause of nonketotic hypoglycemia. *Pediatric Research*, **1985**, 19, 666-71	3.2	192
200	Chronic cardiomyopathy and weakness or acute coma in children with a defect in carnitine uptake. *Annals of Neurology*, **1991**, 30, 709-16	9.4	188
199	Medium-chain acyl-CoA dehydrogenase deficiency in children with non-ketotic hypoglycemia and low carnitine levels. *Pediatric Research*, **1983**, 17, 877-84	3.2	187
198	Diagnosis and localization of focal congenital hyperinsulinism by 18F-fluorodopa PET scan. *Journal of Pediatrics*, **2007**, 150, 140-5	3.6	179
197	Hyperinsulinism/hyperammonemia syndrome: insights into the regulatory role of glutamate dehydrogenase in ammonia metabolism. *Molecular Genetics and Metabolism*, **2004**, 81 Suppl 1, S45-51	3.7	170
196	Brief report: a deficiency of carnitine-acylcarnitine translocase in the inner mitochondrial membrane. *New England Journal of Medicine*, **1992**, 327, 19-23	59.2	159
195	Carnitine deficiency disorders in children. *Annals of the New York Academy of Sciences*, **2004**, 1033, 42-51	6.5	157
194	Perspective on the Genetics and Diagnosis of Congenital Hyperinsulinism Disorders. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 815-26	5.6	155
193	From disease association to risk assessment: an optimistic view from genome-wide association studies on type 1 diabetes. *PLoS Genetics*, **2009**, 5, e1000678	6	150
192	Hyperinsulinism in infants and children. *Pediatric Clinics of North America*, **1997**, 44, 363-74	3.6	149
191	Medium-chain acyl-CoA dehydrogenase deficiency. Diagnosis by stable-isotope dilution measurement of urinary n-hexanoylglycine and 3-phenylpropionylglycine. *New England Journal of Medicine*, **1988**, 319, 1308-13	59.2	148

190	Mechanisms of Disease: advances in diagnosis and treatment of hyperinsulinism in neonates. *Nature Clinical Practice Endocrinology and Metabolism*, **2007**, 3, 57-68		144
189	Comparative genetic analysis of inflammatory bowel disease and type 1 diabetes implicates multiple loci with opposite effects. *Human Molecular Genetics*, **2010**, 19, 2059-67	5.6	136
188	A multidisciplinary approach to the focal form of congenital hyperinsulinism leads to successful treatment by partial pancreatectomy. *Journal of Pediatric Surgery*, **2004**, 39, 270-5	2.6	136
187	Clinical characteristics and biochemical mechanisms of congenital hyperinsulinism associated with dominant KATP channel mutations. *Journal of Clinical Investigation*, **2008**, 118, 2877-86	15.9	132
186	Structures of bovine glutamate dehydrogenase complexes elucidate the mechanism of purine regulation. *Journal of Molecular Biology*, **2001**, 307, 707-20	6.5	130
185	Re-evaluating "transitional neonatal hypoglycemia": mechanism and implications for management. *Journal of Pediatrics*, **2015**, 166, 1520-5.e1	3.6	127
184	Mechanism of hyperinsulinism in short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency involves activation of glutamate dehydrogenase. *Journal of Biological Chemistry*, **2010**, 285, 31806-18	5.4	123
183	Green tea polyphenols modulate insulin secretion by inhibiting glutamate dehydrogenase. *Journal of Biological Chemistry*, **2006**, 281, 10214-21	5.4	122
182	Regulation of leucine-stimulated insulin secretion and glutamine metabolism in isolated rat islets. *Journal of Biological Chemistry*, **2003**, 278, 2853-8	5.4	122
181	Glycemic response to glucagon during fasting hypoglycemia: an aid in the diagnosis of hyperinsulinism. *Journal of Pediatrics*, **1980**, 96, 257-9	3.6	121
180	Protein-sensitive and fasting hypoglycemia in children with the hyperinsulinism/hyperammonemia syndrome. *Journal of Pediatrics*, **2001**, 138, 383-9	3.6	120
179	A novel susceptibility locus for type 1 diabetes on Chr12q13 identified by a genome-wide association study. *Diabetes*, **2008**, 57, 1143-6	0.9	118
178	Accuracy of [18F]fluorodopa positron emission tomography for diagnosing and localizing focal congenital hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 4706-11	5.6	115
177	Hyperinsulinism/hyperammonemia syndrome in children with regulatory mutations in the inhibitory guanosine triphosphate-binding domain of glutamate dehydrogenase. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 1782-7	5.6	115
176	A signaling role of glutamine in insulin secretion. *Journal of Biological Chemistry*, **2004**, 279, 13393-401	5.4	114
175	Novel presentations of congenital hyperinsulinism due to mutations in the MODY genes: HNF1A and HNF4A. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E2026-30	5.6	113
174	Follow-up analysis of genome-wide association data identifies novel loci for type 1 diabetes. *Diabetes*, **2009**, 58, 290-5	0.9	112
173	Clinical features and insulin regulation in infants with a syndrome of prolonged neonatal hyperinsulinism. *Journal of Pediatrics*, **2006**, 148, 207-12	3.6	111

172	Late effects after treatment of twenty children with soft tissue sarcomas of the head and neck. Experience at a single institution with a review of the literature. *Cancer*, **1986**, 57, 2070-6	6.4	110
171	Acute fatty liver of pregnancy and long-chain 3-hydroxyacyl□oenzyme A dehydrogenase deficiency. *Hepatology*, **1994**, 19, 339-345	11.2	108
170	The structure of apo human glutamate dehydrogenase details subunit communication and allostery. *Journal of Molecular Biology*, **2002**, 318, 765-77	6.5	106
169	Pancreatic head resection and Roux-en-Y pancreaticojejunostomy for the treatment of the focal form of congenital hyperinsulinism. *Journal of Pediatric Surgery*, **2012**, 47, 130-5	2.6	105
168	Genetic deficiency of medium-chain acyl coenzyme A dehydrogenase: studies in cultured skin fibroblasts and peripheral mononuclear leukocytes. *Pediatric Research*, **1985**, 19, 671-6	3.2	105
167	Type 2 diabetes and congenital hyperinsulinism cause DNA double-strand breaks and p53 activity in □cells. *Cell Metabolism*, **2014**, 19, 109-21	24.6	101
166	Medium-chain and long-chain acyl CoA dehydrogenase deficiency: clinical, pathologic and ultrastructural differentiation from Reye□s syndrome. *Hepatology*, **1986**, 6, 1270-8	11.2	94
165	Hyperinsulinism in infancy and childhood: when an insulin level is not always enough. *Clinical Chemistry*, **2008**, 54, 256-63	5.5	90
164	The structure and allosteric regulation of mammalian glutamate dehydrogenase. *Archives of Biochemistry and Biophysics*, **2012**, 519, 69-80	4.1	87
163	A syndrome of congenital hyperinsulinism and hyperammonemia. *Journal of Pediatrics*, **1997**, 130, 661-4	3.6	87
162	Sudden neonatal death in carnitine transporter deficiency. *Journal of Pediatrics*, **1997**, 131, 304-5	3.6	86
161	Preoperative evaluation of infants with focal or diffuse congenital hyperinsulinism by intravenous acute insulin response tests and selective pancreatic arterial calcium stimulation. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 288-96	5.6	83
160	Extremes of clinical and enzymatic phenotypes in children with hyperinsulinism caused by glucokinase activating mutations. *Diabetes*, **2009**, 58, 1419-27	0.9	81
159	Untangling the glutamate dehydrogenase allosteric nightmare. *Trends in Biochemical Sciences*, **2008**, 33, 557-64	10.3	81
158	Congenital hyperinsulinism associated ABCC8 mutations that cause defective trafficking of ATP-sensitive K+ channels: identification and rescue. *Diabetes*, **2007**, 56, 2339-48	0.9	80
157	Newborn screening by tandem mass spectrometry for medium-chain Acyl-CoA dehydrogenase deficiency: a cost-effectiveness analysis. *Pediatrics*, **2003**, 112, 1005-15	7.4	80
156	Clinical and molecular characterization of a dominant form of congenital hyperinsulinism caused by a mutation in the high-affinity sulfonylurea receptor. *Diabetes*, **2003**, 52, 2403-10	0.9	78
155	SUN-310 Duodenal Wall Insulinoma with Recurrence as Lymph Node Metastases. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78

154	GLP-1 receptor antagonist exendin-(9-39) elevates fasting blood glucose levels in congenital hyperinsulinism owing to inactivating mutations in the ATP-sensitive K+ channel. *Diabetes*, **2012**, 61, 2585-91	0.9	77
153	Necrotizing enterocolitis in neonates receiving octreotide for the management of congenital hyperinsulinism. *Pediatric Diabetes*, **2010**, 11, 142-7	3.6	77
152	Insights into the structure and regulation of glucokinase from a novel mutation (V62M), which causes maturity-onset diabetes of the young. *Journal of Biological Chemistry*, **2005**, 280, 14105-13	5.4	76
151	Acute insulin responses to leucine in children with the hyperinsulinism/hyperammonemia syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 3724-8	5.6	76
150	Hyperfiltration and renal disease in glycogen storage disease, type I. *Kidney International*, **1989**, 35, 1345-50	5.9	76
149	Glucokinase activation repairs defective bioenergetics of islets of Langerhans isolated from type 2 diabetics. *American Journal of Physiology - Endocrinology and Metabolism*, **2012**, 302, E87-E102	6	71
148	Disorders of glutamate metabolism. *Mental Retardation and Developmental Disabilities Research Reviews*, **2001**, 7, 287-95		71
147	Congenital hyperinsulinism and the surgeon: lessons learned over 35 years. *Journal of Pediatric Surgery*, **1999**, 34, 786-92; discussion 792-3	2.6	71
146	High Risk of Diabetes and Neurobehavioral Deficits in Individuals With Surgically Treated Hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 4133-9	5.6	70
145	Fasting hypoketotic coma in a child with deficiency of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase. *New England Journal of Medicine*, **1997**, 337, 1203-7	59.2	70
144	Regulation of glucagon secretion in normal and diabetic human islets by γ-hydroxybutyrate and glycine. *Journal of Biological Chemistry*, **2013**, 288, 3938-51	5.4	69
143	Linkage-disequilibrium mapping without genotyping. *Nature Genetics*, **1998**, 18, 225-30	36.3	69
142	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
141	Central nervous system hyperexcitability associated with glutamate dehydrogenase gain of function mutations. *Journal of Pediatrics*, **2005**, 146, 388-94	3.6	69
140	Structural studies on ADP activation of mammalian glutamate dehydrogenase and the evolution of regulation. *Biochemistry*, **2003**, 42, 3446-56	3.2	69
139	Expression, purification and characterization of human glutamate dehydrogenase (GDH) allosteric regulatory mutations. *Biochemical Journal*, **2002**, 363, 81-87	3.8	69
138	Mitochondrial carnitine-acylcarnitine translocase deficiency presenting as sudden neonatal death. *Journal of Pediatrics*, **1997**, 131, 220-5	3.6	67
137	Regulation of glutamate metabolism and insulin secretion by glutamate dehydrogenase in hypoglycemic children. *American Journal of Clinical Nutrition*, **2009**, 90, 862S-866S	7	65

136	The hyperinsulinism/hyperammonemia syndrome. *Reviews in Endocrine and Metabolic Disorders*, **2010**, 11, 171-8	10.5	61
135	From clinicogenetic studies of maturity-onset diabetes of the young to unraveling complex mechanisms of glucokinase regulation. *Diabetes*, **2006**, 55, 1713-22	0.9	61
134	Diazoxide-unresponsive congenital hyperinsulinism in children with dominant mutations of the β-cell sulfonylurea receptor SUR1. *Diabetes*, **2011**, 60, 1797-804	0.9	60
133	Effects of hypoglycemia on developmental outcome in children with congenital hyperinsulinism. *Journal of Pediatric Nursing*, **2005**, 20, 109-18	2.2	60
132	Molecular and immunohistochemical analyses of the focal form of congenital hyperinsulinism. *Modern Pathology*, **2006**, 19, 122-9	9.8	60
131	Advances in diagnosis and treatment of hyperinsulinism in infants and children. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 4857-9	5.6	59
130	Biomarkers of Insulin for the Diagnosis of Hyperinsulinemic Hypoglycemia in Infants and Children. *Journal of Pediatrics*, **2016**, 168, 212-219	3.6	58
129	Accuracy of PET/CT Scan in the diagnosis of the focal form of congenital hyperinsulinism. *Journal of Pediatric Surgery*, **2013**, 48, 388-93	2.6	58
128	Insulin-like growth factor binding protein-1 levels in the diagnosis of hypoglycemia caused by hyperinsulinism. *Journal of Pediatrics*, **1997**, 131, 193-9	3.6	58
127	Nephrolithiasis, hypocitraturia, and a distal renal tubular acidification defect in type 1 glycogen storage disease. *Journal of Pediatrics*, **1993**, 122, 392-6	3.6	56
126	Familial and sporadic hyperinsulinism: histopathologic findings and segregation analysis support a single autosomal recessive disorder. *Journal of Pediatrics*, **1991**, 119, 721-4	3.6	55
125	Effects of a GTP-insensitive mutation of glutamate dehydrogenase on insulin secretion in transgenic mice. *Journal of Biological Chemistry*, **2006**, 281, 15064-72	5.4	54
124	Green tea polyphenols control dysregulated glutamate dehydrogenase in transgenic mice by hijacking the ADP activation site. *Journal of Biological Chemistry*, **2011**, 286, 34164-74	5.4	53
123	Exendin-(9-39) corrects fasting hypoglycemia in SUR-1-/- mice by lowering cAMP in pancreatic beta-cells and inhibiting insulin secretion. *Journal of Biological Chemistry*, **2008**, 283, 25786-93	5.4	53
122	Familial leucine-sensitive hypoglycemia of infancy due to a dominant mutation of the beta-cell sulfonylurea receptor. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 4450-6	5.6	53
121	Hyperinsulinism and hyperammonemia syndrome: report of twelve unrelated patients. *Pediatric Research*, **2001**, 50, 353-7	3.2	53
120	Congenital hyperinsulinism in children with paternal 11p uniparental isodisomy and Beckwith-Wiedemann syndrome. *Journal of Medical Genetics*, **2016**, 53, 53-61	5.8	53
119	Genotype-phenotype correlations in children with congenital hyperinsulinism due to recessive mutations of the adenosine triphosphate-sensitive potassium channel genes. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 789-94	5.6	52

118	A novel KCNJ11 mutation associated with congenital hyperinsulinism reduces the intrinsic open probability of beta-cell ATP-sensitive potassium channels. *Journal of Biological Chemistry*, **2006**, 281, 3006-12	5.4	51
117	Carnitine membrane transporter deficiency: a long-term follow up and OCTN2 mutation in the first documented case of primary carnitine deficiency. *Molecular Genetics and Metabolism*, **2002**, 77, 195-201	3.7	51
116	The diagnosis of ectopic focal hyperinsulinism of infancy with [18F]-dopa positron emission tomography. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 2839-42	5.6	49
115	Histopathology of congenital hyperinsulinism: retrospective study with genotype correlations. *Pediatric and Developmental Pathology*, **2003**, 6, 322-33	2.2	49
114	The value of radiologic interventions and (18)F-DOPA PET in diagnosing and localizing focal congenital hyperinsulinism: systematic review and meta-analysis. *Molecular Imaging and Biology*, **2013**, 15, 97-105	3.8	48
113	Dominant form of congenital hyperinsulinism maps to HK1 region on 10q. *Hormone Research in Paediatrics*, **2013**, 80, 18-27	3.3	48
112	Congenital hyperinsulinism: intraoperative biopsy interpretation can direct the extent of pancreatectomy. *American Journal of Surgical Pathology*, **2004**, 28, 1326-35	6.7	48
111	Pregnancy in glycogen storage disease type Ib: gestational care and report of first successful deliveries. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S151-7	5.4	46
110	Expression, purification and characterization of human glutamate dehydrogenase (GDH) allosteric regulatory mutations. *Biochemical Journal*, **2002**, 363, 81-7	3.8	46
109	Calcium-stimulated insulin secretion in diffuse and focal forms of congenital hyperinsulinism. *Journal of Pediatrics*, **2000**, 137, 239-46	3.6	46
108	Surgical treatment of congenital hyperinsulinism: Results from 500 pancreatectomies in neonates and children. *Journal of Pediatric Surgery*, **2019**, 54, 27-32	2.6	46
107	The structure and allosteric regulation of glutamate dehydrogenase. *Neurochemistry International*, **2011**, 59, 445-55	4.4	45
106	The structure and organization of the human carnitine/acylcarnitine translocase (CACT1) gene2. *Biochemical and Biophysical Research Communications*, **1998**, 252, 770-4	3.4	45
105	A specialized team approach to diagnosis and medical versus surgical treatment of infants with congenital hyperinsulinism. *Seminars in Pediatric Surgery*, **2011**, 20, 32-7	2.1	44
104	Destabilization of ATP-sensitive potassium channel activity by novel KCNJ11 mutations identified in congenital hyperinsulinism. *Journal of Biological Chemistry*, **2008**, 283, 9146-56	5.4	42
103	Evolution of glutamate dehydrogenase regulation of insulin homeostasis is an example of molecular exaptation. *Biochemistry*, **2004**, 43, 14431-43	3.2	41
102	The surgical management of insulinomas in children. *Journal of Pediatric Surgery*, **2013**, 48, 2517-24	2.6	40
101	Poor specificity of low growth hormone and cortisol levels during fasting hypoglycemia for the diagnoses of growth hormone deficiency and adrenal insufficiency. *Pediatrics*, **2008**, 122, e522-8	7.4	39

100	A mutation in the TMD0-L0 region of sulfonylurea receptor-1 (L225P) causes permanent neonatal diabetes mellitus (PNDM). *Diabetes*, **2007**, 56, 1357-62	0.9	39
99	Dual regulation of insulin-like growth factor binding protein-1 levels by insulin and cortisol during fasting. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 4426-30	5.6	38
98	In vitro and in vivo effects of granulocyte colony-stimulating factor on neutrophils in glycogen storage disease type 1B: granulocyte colony-stimulating factor therapy corrects the neutropenia and the defects in respiratory burst activity and Ca2+ mobilization. *Pediatric Research*, **1994**, 35, 84-90	3.2	38
97	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1 Deficiency. *Journal of Pediatrics*, **2016**, 175, 130-136.e8	3.6	37
96	Multiple ectopic lesions of focal islet adenomatosis identified by positron emission tomography scan in an infant with congenital hyperinsulinism. *Journal of Pediatric Surgery*, **2007**, 42, 188-92	2.6	37
95	Renal handling of carnitine in secondary carnitine deficiency disorders. *Pediatric Research*, **1993**, 34, 89-97	3.2	36
94	Acarbose treatment of postprandial hypoglycemia in children after Nissen fundoplication. *Journal of Pediatrics*, **2001**, 139, 877-9	3.6	35
93	Acute insulin responses to calcium and tolbutamide do not differentiate focal from diffuse congenital hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 925-9	5.6	34
92	Novel Hypoglycemia Phenotype in Congenital Hyperinsulinism Due to Dominant Mutations of Uncoupling Protein 2. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 942-949	5.6	33
91	Determination of insulin for the diagnosis of hyperinsulinemic hypoglycemia. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2013**, 27, 763-9	6.5	33
90	Hypertrophic cardiomyopathy in neonates with congenital hyperinsulinism. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2013**, 98, F351-4	4.7	33
89	Mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: clinical course and description of causal mutations in two patients. *Pediatric Research*, **2001**, 49, 326-31	3.2	33
88	Glutaminolysis and insulin secretion: from bedside to bench and back. *Diabetes*, **2002**, 51 Suppl 3, S421-6	0.9	33
87	Dietary-dependent carnitine deficiency as a cause of nonketotic hypoglycemia in an infant. *Journal of Pediatrics*, **1981**, 99, 551-5	3.6	33
86	Elimination of KATP channels in mouse islets results in elevated [U-13C]glucose metabolism, glutaminolysis, and pyruvate cycling but a decreased gamma-aminobutyric acid shunt. *Journal of Biological Chemistry*, **2008**, 283, 17238-49	5.4	32
85	Glutamate dehydrogenase: structure, allosteric regulation, and role in insulin homeostasis. *Neurochemical Research*, **2014**, 39, 433-45	4.6	31
84	Intragastric feeding in type I glycogen storage disease: factors affecting the control of lactic acidemia. *Pediatric Research*, **1981**, 15, 1504-8	3.2	31
83	Glutamate Dehydrogenase, a Complex Enzyme at a Crucial Metabolic Branch Point. *Neurochemical Research*, **2019**, 44, 117-132	4.6	29

82	Functional and Metabolomic Consequences of K Channel Inactivation in Human Islets. *Diabetes*, **2017**, 66, 1901-1913	0.9	28
81	Pharmacological Correction of Trafficking Defects in ATP-sensitive Potassium Channels Caused by Sulfonylurea Receptor 1 Mutations. *Journal of Biological Chemistry*, **2016**, 291, 21971-21983	5.4	28
80	Protein-sensitive hypoglycemia without leucine sensitivity in hyperinsulinism caused by K(ATP) channel mutations. *Journal of Pediatrics*, **2006**, 149, 47-52	3.6	27
79	Detection of inborn errors of fatty acid oxidation from acylcarnitine analysis of plasma and blood spots with the radioisotopic exchange-high-performance liquid chromatographic method. *Journal of Pediatrics*, **1993**, 122, 708-14	3.6	27
78	Pancreatic surgery in infants with Beckwith-Wiedemann syndrome and hyperinsulinism. *Journal of Pediatric Surgery*, **2013**, 48, 2511-6	2.6	26
77	Acute Insulin Responses to Leucine in Children with the Hyperinsulinism/Hyperammonemia Syndrome		26
76	Aberrant mRNA splicing associated with coding region mutations in children with carnitine-acylcarnitine translocase deficiency. *Molecular Genetics and Metabolism*, **2001**, 74, 248-55	3.7	25
75	Endocardial fibroelastosis and primary carnitine deficiency due to a defect in the plasma membrane carnitine transporter. *Clinical Cardiology*, **1996**, 19, 243-6	3.3	25
74	Genetic disorders of mitochondrial fatty acid oxidation. *Current Opinion in Pediatrics*, **1994**, 6, 476-81	3.2	25
73	Mutational analysis of allosteric activation and inhibition of glucokinase. *Biochemical Journal*, **2011**, 440, 203-15	3.8	24
72	Sulfonylurea receptor 1 mutations that cause opposite insulin secretion defects with chemical chaperone exposure. *Journal of Biological Chemistry*, **2009**, 284, 7951-9	5.4	24
71	Familial carnitine transporter defect: A treatable cause of cardiomyopathy in children. *American Heart Journal*, **2000**, 139, S96-S106	4.9	24
70	Two genetic forms of hyperinsulinemic hypoglycemia caused by dysregulation of glutamate dehydrogenase. *Neurochemistry International*, **2011**, 59, 465-72	4.4	23
69	Measurement of tissue acyl-CoAs using flow-injection tandem mass spectrometry: acyl-CoA profiles in short-chain fatty acid oxidation defects. *Molecular Genetics and Metabolism*, **2012**, 107, 679-83	3.7	22
68	Management of hyperinsulinism in infants. *Journal of Pediatrics*, **1991**, 119, 755-7	3.6	22
67	Development of hepatic fatty acid oxidation and ketogenesis in the newborn guinea pig. *Pediatric Research*, **1983**, 17, 224-9	3.2	22
66	Systemic carnitine deficiency simulating Reye syndrome. *Journal of Pediatrics*, **1984**, 105, 679	3.6	22
65	Congenital Hyperinsulinism in Infants with Turner Syndrome: Possible Association with Monosomy X and KDM6A Haploinsufficiency. *Hormone Research in Paediatrics*, **2018**, 89, 413-422	3.3	21

64	First prenatal diagnosis of the carnitine transporter defect. *American Journal of Medical Genetics Part A*, **1996**, 66, 21-4		21
63	Nesidioblastosis no longer! It's all about genetics. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, 617-9	5.6	20
62	Rare forms of congenital hyperinsulinism. *Seminars in Pediatric Surgery*, **2011**, 20, 38-44	2.1	18
61	Hepatocyte nuclear factor 4 gene mutation associated with familial neonatal hyperinsulinism and maturity-onset diabetes of the young. *Journal of Pediatrics*, **2011**, 158, 852-4	3.6	18
60	Plasma ketones in newborn infants: absence of suckling ketosis. *Journal of Pediatrics*, **1981**, 98, 628-30	3.6	18
59	Pancreatic adenomas in infants and children: current surgical management. *Journal of Pediatric Surgery*, **1978**, 13, 591-6	2.6	18
58	Histologic and Molecular Profile of Pediatric Insulinomas: Evidence of a Paternal Parent-of-Origin Effect. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 914-22	5.6	17
57	Short-chain 3-hydroxyacyl-coenzyme A dehydrogenase associates with a protein super-complex integrating multiple metabolic pathways. *PLoS ONE*, **2012**, 7, e35048	3.7	16
56	Dissecting the spectrum of fatty acid oxidation disorders. *Journal of Pediatrics*, **1998**, 132, 384-6	3.6	16
55	Regulation of K Channel Trafficking in Pancreatic Cells by Protein Histidine Phosphorylation. *Diabetes*, **2018**, 67, 849-860	0.9	14
54	Mitochondrial GTP insensitivity contributes to hypoglycemia in hyperinsulinemia hyperammonemia by inhibiting glucagon release. *Diabetes*, **2014**, 63, 4218-29	0.9	14
53	Intussusception after pancreatic surgery in children: a case series. *Journal of Pediatric Surgery*, **2010**, 45, 1496-9	2.6	14
52	Intragenic single nucleotide polymorphism haplotype analysis of SUR1 mutations in familial hyperinsulinism. *Human Mutation*, **1999**, 14, 23-9	4.7	13
51	A novel atypical presentation of insulin autoimmune syndrome (Hirata's disease) in a child. *Journal of Pediatric Endocrinology and Metabolism*, **2013**, 26, 1163-6	1.6	12
50	Postmortem recognition of fatty acid oxidation disorders. *Pediatric Pathology*, **1991**, 11, 365-70		12
49	Medications used in the treatment of hypoglycemia due to congenital hyperinsulinism of infancy (HI). *Pediatric Endocrinology Reviews*, **2004**, 2 Suppl 1, 163-7	1.1	12
48	A severe case of hyperinsulinism due to hemizygous activating mutation of glutamate dehydrogenase. *Pediatric Diabetes*, **2017**, 18, 911-916	3.6	11
47	Clinical heterogeneity of hyperinsulinism due to HNF1A and HNF4A mutations. *Pediatric Diabetes*, **2018**, 19, 910-916	3.6	11

46	Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: clinical and functional characterization of two novel ABCC8 mutations. *Gene*, **2013**, 516, 122-5	3.8	10
45	Hypoglycemia in the neonate. *Pediatric Endocrinology Reviews*, **2006**, 4 Suppl 1, 76-81	1.1	10
44	Prenatal diagnosis and postnatal management of diffuse congenital hyperinsulinism: a case report. *Fetal Diagnosis and Therapy*, **2006**, 21, 515-8	2.4	9
43	Disorders of Mitochondrial Fatty Acid Oxidation and Related Metabolic Pathways **2006**, 175-190		9
42	Clinical and molecular heterogeneity of familial hyperinsulinism. *Journal of Pediatrics*, **1998**, 133, 801-2	3.6	8
41	Suppression of insulin oversecretion by subcutaneous recombinant human insulin-like growth factor I in children with congenital hyperinsulinism due to defective beta-cell sulfonylurea receptor. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 3117-24	5.6	8
40	Interferon-gamma corrects the respiratory burst defect in vitro in monocyte-derived macrophages from glycogen storage disease type 1b patients. *Pediatric Research*, **1993**, 34, 265-9	3.2	8
39	Novel dominant K channel mutations in infants with congenital hyperinsulinism: Validation by in vitro expression studies and in vivo carrier phenotyping. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2214-2227	2.5	7
38	Blood glucose control during selective arterial stimulation and venous sampling for localization of focal hyperinsulinism lesions in anesthetized children. *Anesthesia and Analgesia*, **2004**, 99, 1044-1048	3.9	7
37	Pathophysiology of Diffuse ATP-Sensitive Potassium Channel Hyperinsulinism. *Frontiers in Diabetes*, **2012**, 18-29	0.6	6
36	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 542-551	2.5	5
35	Hypoglycemia in the toddler and child **2014**, 920-955.e1		5
34	Hypoglycemia in the newborn and infant **2014**, 157-185.e2		5
33	Disorders of Carbohydrate Metabolism **2005**, 1410-1422		5
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