Vivian Hwa

List of Publications by Year
in descending order

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85	4,915	101384	95083
papers	citations	36 h-index	68 g-index

87	87	87	5092
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Insulin-Like Growth Factor-Binding Protein (IGFBP) Superfamily*. Endocrine Reviews, 1999, 20, 761-787.	8.9	832
2	Growth Hormone Insensitivity Associated with aSTAT5bMutation. New England Journal of Medicine, 2003, 349, 1139-1147.	13.9	492
3	The Neisseria meningitidis haemoglobin receptor: its role in iron utilization and virulence. Molecular Microbiology, 1995, 15, 531-541.	1.2	214
4	Cutting Edge: Decreased Accumulation and Regulatory Function of CD4+CD25high T Cells in Human STAT5b Deficiency. Journal of Immunology, 2006, 177, 2770-2774.	0.4	212
5	Evidence for a Continuum of Genetic, Phenotypic, and Biochemical Abnormalities in Children with Growth Hormone Insensitivity. Endocrine Reviews, 2011, 32, 472-497.	8.9	171
6	Mutations in pregnancyâ€associated plasma protein A2 cause short stature due to low <scp>IGF</scp> â€availability. EMBO Molecular Medicine, 2016, 8, 363-374.	3.3	147
7	Defects in growth hormone receptor signaling. Trends in Endocrinology and Metabolism, 2007, 18, 134-141.	3.1	134
8	Severe Growth Hormone Insensitivity Resulting from Total Absence of Signal Transducer and Activator of Transcription 5b. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 4260-4266.	1.8	132
9	STAT5b Deficiency: An Unsuspected Cause of Growth Failure, Immunodeficiency, and Severe Pulmonary Disease. Journal of Pediatrics, 2011, 158, 701-708.	0.9	110
10	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	9.4	110
11	Human Acid-Labile Subunit Deficiency: Clinical, Endocrine and Metabolic Consequences. Hormone Research, 2009, 72, 129-141.	1.8	109
12	STAT5b deficiency: Lessons from STAT5b gene mutations. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 61-75.	2.2	100
13	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 460-469.	1.8	95
14	Total Absence of Functional Acid Labile Subunit, Resulting in Severe Insulin-Like Growth Factor Deficiency and Moderate Growth Failure. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1826-1831.	1.8	91
15	Dominant-negative STAT5B mutations cause growth hormone insensitivity with short stature and mild immune dysregulation. Nature Communications, 2018, 9, 2105.	5.8	81
16	STAT5B deficiency: Impacts on human growth and immunity. Growth Hormone and IGF Research, 2016, 28, 16-20.	0.5	80
17	The Growth Hormone Cascade and Its Role in Mammalian Growth. Hormone Research in Paediatrics, 2009, 71, 36-40.	0.8	74
18	Binding Properties and Distribution of Insulin-Like Growth Factor Binding Protein-Related Protein 3 (IGFBP-rP3/NovH), an Additional Member of the IGFBP Superfamily1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 1096-1103.	1.8	66

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19	Familial Short Stature Caused by Haploinsufficiency of the Insulin-Like Growth Factor I Receptor due to Nonsense-Mediated Messenger Ribonucleic Acid Decay. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1740-1747.	1.8	66
20	Acid-labile subunit (ALS) deficiency. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 101-113.	2.2	60
21	Large-Scale Pooled Next-Generation Sequencing of 1077 Genes to Identify Genetic Causes of Short Stature. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1428-E1437.	1.8	60
22	Severe Short Stature Caused by Novel Compound Heterozygous Mutations of the Insulin-Like Growth Factor 1 Receptor (IGF1R). Journal of Clinical Endocrinology and Metabolism, 2012, 97, E243-E247.	1.8	59
23	Impact of Heterozygosity for Acid-Labile Subunit (IGFALS) Gene Mutations on Stature: Results from the International Acid-Labile Subunit Consortium. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 4184-4191.	1.8	52
24	AnXRCC4Splice Mutation Associated With Severe Short Stature, Gonadal Failure, and Early-Onset Metabolic Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E789-E798.	1.8	52
25	Insulin-like growth factor binding protein-3 and-5 are regulated by transforming growth factor-Î² and retinoic acid in the human prostate adenocarcinoma cell line PC-3. Endocrine, 1997, 6, 235-242.	1.1	49
26	Growth Hormone Insensitivity and Severe Short Stature in Siblings: A Novel Mutation at the Exon 13-Intron 13 Junction of the STAT5b Gene. Hormone Research in Paediatrics, 2007, 68, 218-224.	0.8	49
27	Transcriptional Regulation of Insulin-like Growth Factor-I by Interferon-Î³ Requires STAT-5b. Journal of Biological Chemistry, 2004, 279, 2728-2736.	1.6	48
28	A Novel Variant inCDKN1CIs Associated With Intrauterine Growth Restriction, Short Stature, and Early-Adulthood-Onset Diabetes. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2117-E2122.	1.8	45
29	The Insulin-like Growth Factor Binding Protein Superfamily: New Perspectives. Pediatrics, 1999, 104, 1018-1021.	1.0	43
30	Primary Growth Hormone (GH) Insensitivity and Insulin-Like Growth Factor Deficiency Caused by Novel Compound Heterozygous Mutations of the GH Receptor Gene: Genetic and Functional Studies of Simple and Compound Heterozygous States. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2223-2231.	1.8	42
31	Differentiating the roles of STAT5B and STAT5A in human CD4+ T cells. Clinical Immunology, 2013, 148, 227-236.	1.4	40
32	A Novel Missense Mutation in the SH2 Domain of theSTAT5BGene Results in a Transcriptionally Inactive STAT5b Associated with Severe IGF-I Deficiency, Immune Dysfunction, and Lack of Pulmonary Disease. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E830-E839.	1.8	38
33	Identification of a Novel Heterozygous <i>IGF1</i> Splicing Mutation in a Large Kindred with Familial Short Stature. Hormone Research in Paediatrics, 2012, 78, 59-66.	0.8	38
34	A Successful HSCT in a Girl with Novel LRBA Mutation with Refractory Celiac Disease. Journal of Clinical Immunology, 2016, 36, 8-11.	2.0	38
35	Partial growth hormone insensitivity and dysregulatory immune disease associated with de novo germline activating STAT3 mutations. Molecular and Cellular Endocrinology, 2018, 473, 166-177.	1.6	38
36	Interaction of IGF-Binding Protein-Related Protein 1 with a Novel Protein, Neuroendocrine Differentiation Factor, Results in Neuroendocrine Differentiation of Prostate Cancer Cells. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 4504-4511.	1.8	36

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37	A Mutant Signal Transducer and Activator of Transcription 5b, Associated with Growth Hormone Insensitivity and Insulin-Like Growth Factor-I Deficiency, Cannot Function as a Signal Transducer or Transcription Factor. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1526-1534.	1.8	36
38	Connective Tissue Growth Factor (IGFBP-rP2) Expression and Regulation in Cultured Bovine Endothelial Cells*. Endocrinology, 1999, 140, 1575-1580.	1.4	35
39	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	2.6	34
40	Nonclassical GH Insensitivity: Characterization of Mild Abnormalities of GH Action. Endocrine Reviews, 2019, 40, 476-505.	8.9	32
41	Mutations in TKT Are the Cause of a Syndrome Including Short Stature, Developmental Delay, and Congenital Heart Defects. American Journal of Human Genetics, 2016, 98, 1235-1242.	2.6	31
42	Three Novel <i>IGFALS</i> Gene Mutations Resulting in Total ALS and Severe Circulating IGF-I/IGFBP-3 Deficiency in Children of Different Ethnic Origins. Hormone Research in Paediatrics, 2009, 71, 100-110.	0.8	29
43	STAT5B mutations in heterozygous state have negative impact on height: another clue in human stature heritability. European Journal of Endocrinology, 2015, 173, 291-296.	1.9	29
44	Aberrant Folding of a Mutant Stat5b Causes Growth Hormone Insensitivity and Proteasomal Dysfunction. Journal of Biological Chemistry, 2006, 281, 6552-6558.	1.6	28
45	Pharmacokinetics of IGF-1 in PAPP-A2-Deficient Patients, Growth Response, and Effects on Glucose and Bone Density. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4568-4577.	1.8	27
46	Novel Dominant-Negative GH Receptor Mutations Expands the Spectrum of GHI and IGF-I Deficiency. Journal of the Endocrine Society, 2017, 1, 345-358.	0.1	26
47	Novel Modulators of the Growth Hormone - Insulin-Like Growth Factor Axis: Pregnancy-Associated Plasma Protein-A2 and Stanniocalcin-2. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 1-8.	0.4	26
48	IGFALS Gene Dosage Effects on Serum IGF-I and Glucose Metabolism, Body Composition, Bone Growth in Length and Width, and the Pharmacokinetics of Recombinant Human IGF-I Administration. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E703-E712.	1.8	25
49	In Vitro and in Vivo Characterization of MOD-4023, a Long-Acting Carboxy-Terminal Peptide (CTP)-Modified Human Growth Hormone. Molecular Pharmaceutics, 2016, 13, 631-639.	2.3	25
50	The Growth Hormone Receptor (GHR)c.899dupCMutation Functions as a Dominant Negative: Insights into the Pathophysiology of IntracellularGHRDefects. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1896-E1904.	1.8	24
51	A Novel Y332C Missense Mutation in the Intracellular Domain of The Human Growth Hormone Receptor Does Not Alter STAT5b Signaling: Redundancy of GHR Intracellular Tyrosines Involved in STAT5b Signaling. Hormone Research in Paediatrics, 2011, 75, 187-199.	0.8	23
52	A novel mutation in <scp>IGFALS</scp>, c.380<scp>T</scp>><scp>C</scp> (p.L127P), associated with short stature, delayed puberty, osteopenia and hyperinsulinaemia in two siblings: insights into the roles of insulin growth factorâ€1 (<scp>IGF</scp>1). Clinical Endocrinology, 2013, 79, 838-844.	1.2	22
53	Growth Hormone (GH) Insensitivity and Insulin-Like Growth Factor-I Deficiency in Inuit Subjects and an Ecuadorian Cohort: Functional Studies of Two Codon 180 GH Receptor Gene Mutations. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1030-1037.	1.8	20
54	Atypical GH Insensitivity Syndrome and Severe Insulin-Like Growth Factor-I Deficiency Resulting from Compound Heterozygous Mutations of the GH Receptor, Including a Novel Frameshift Mutation Affecting the Intracellular Domain. Hormone Research in Paediatrics, 2010, 74, 406-411.	0.8	19

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55	The E180splice mutation in the GHR gene causing Laron syndrome: Witness of a Sephardic Jewish exodus from the Iberian Peninsula to the New World?. American Journal of Medical Genetics, Part A, 2014, 164, 1204-1208.	0.7	19
56	PAPPA2 as a Therapeutic Modulator of IGF-I Bioavailability: in Vivo and in Vitro Evidence. Journal of the Endocrine Society, 2018, 2, 646-656.	0.1	19
57	STAT5B restrains human B-cell differentiation to maintain humoral immune homeostasis. Journal of Allergy and Clinical Immunology, 2022, 150, 931-946.	1.5	19
58	IGF-I in Human Growth: Lessons from Defects in the GH-IGF-I Axis. Nestle Nutrition Institute Workshop Series, 2013, 71, 43-55.	1.5	18
59	Expanding Genetic and Functional Diagnoses of <i>IGF1R</i> Haploinsufficiencies. Hormone Research in Paediatrics, 2017, 87, 412-422.	0.8	18
60	Extreme Elevation of Serum Growth Hormone-Binding Protein Concentrations Resulting from a Novel Heterozygous Splice Site Mutation of the Growth Hormone Receptor Gene. Hormone Research in Paediatrics, 2009, 71, 276-284.	0.8	16
61	Human growth disorders associated with impaired GH action: Defects in STAT5B and JAK2. Molecular and Cellular Endocrinology, 2021, 519, 111063.	1.6	16
62	Severe Growth Deficiency is Associated with STAT5b Mutations that Disrupt Protein Folding and Activity. Molecular Endocrinology, 2013, 27, 150-161.	3.7	15
63	Low IGF-I Bioavailability Impairs Growth and Glucose Metabolism in a Mouse Model of Human PAPPA2 p.Ala1033Val Mutation. Endocrinology, 2019, 160, 1363-1376.	1.4	15
64	Genetic causes of growth hormone insensitivity beyond GHR. Reviews in Endocrine and Metabolic Disorders, 2021, 22, 43-58.	2.6	13
65	Interaction of IGF-Binding Protein-Related Protein 1 with a Novel Protein, Neuroendocrine Differentiation Factor, Results in Neuroendocrine Differentiation of Prostate Cancer Cells. , 0, .		13
66	Anthropometric and biochemical correlates of PAPP-A2, free IGF-I, and IGFBP-3 in childhood. European Journal of Endocrinology, 2020, 182, 363-374.	1.9	12
67	A Novel Homozygous Mutation of the Acid-Labile Subunit <i>(IGFALS)</i> Gene in a Male Adolescent. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 432-438.	0.4	10
68	IGFBPs and Cancer. Novartis Foundation Symposium, 2008, , 215-234.	1.2	9
69	Developmental Adaptive Immune Defects Associated with STAT5B Deficiency in Three Young Siblings. Journal of Clinical Immunology, 2021, 41, 136-146.	2.0	9
70	Signal Transducer and Activator of Transcription 5B Deficiencyâ€“associated Lung Disease. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 1245-1250.	2.5	8
71	Treatment of Short Stature in Aggrecan-deficient Patients With Recombinant Human Growth Hormone: 1-Year Response. Journal of Clinical Endocrinology and Metabolism, 2021, , .	1.8	7
72	Biology of the somatotroph axis (after the pituitary). Annales D'Endocrinologie, 2017, 78, 80-82.	0.6	6

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73	Severe growth failure associated with a novel heterozygous nonsense mutation in the GHR transmembrane domain leading to elevated growth hormone binding protein. Clinical Endocrinology, 2020, 92, 331-337.	1.2	6
74	Targeted Searches of the Electronic Health Record and Genomics Identify an Etiology in Three Patients with Short Stature and High IGF-I Levels. Hormone Research in Paediatrics, 2019, 92, 186-195.	0.8	5
75	Rare CNVs provide novel insights into the molecular basis of GH and IGF-1 insensitivity. European Journal of Endocrinology, 2020, 183, 581-595.	1.9	5
76	Isolated growth hormone deficiency due to the R183H mutation in <scp>GH</scp>1: Clinical analysis of a fourâ€generation family. Clinical Endocrinology, 2017, 87, 874-876.	1.2	4
77	Two Siblings with a Mutation in <i>CCDC8</i> Presenting with Mild Short Stature: A Case of 3-M Syndrome. Hormone Research in Paediatrics, 2017, 88, 364-370.	0.8	4
78	Prenatal diagnosis of Proteus syndrome: Diagnosis of an AKT1 mutation from amniocytes. Birth Defects Research, 2020, 112, 1733-1737.	0.8	4
79	Growth Hormone Receptor (GHR) 6Î© Pseudoexon Activation: A Novel Cause of Severe Growth Hormone Insensitivity. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e401-e416.	1.8	4
80	A Novel Mutation in Insulin-Like Growth Factor 1 Receptor (c.641-2A>G) Is Associated with Impaired Growth, Hypoglycemia, and Modified Immune Phenotypes. Hormone Research in Paediatrics, 2020, 93, 322-334.	0.8	3
81	Clinical phenotype and musculoskeletal characteristics of patients with aggrecan deficiency. American Journal of Medical Genetics, Part A, 2022, 188, 1193-1203.	0.7	2
82	OR07-6 Integrating Targeted Bioinformatic Searches of the Electronic Health Records and Genomic Testing Identifies a Molecular Diagnosis in Three Patients with Undiagnosed Short Stature. Journal of the Endocrine Society, 2019, 3, .	0.1	1
83	Laron syndrome related to homozygous growth hormone receptor c.784>C mutation in a patient with hypoplastic pulmonary arteries. Cardiovascular Journal of Africa, 2019, 30, e7-e8.	0.2	1
84	Growth Hormone Insensitivity. , 2018, , 81-93.		0
85	OR07-5 A Cross-Sectional Study of IGF-I Bioavailability through Childhood: Associations with PAPP-A2 and Anthropometric Data. Journal of the Endocrine Society, 2019, 3, .	0.1	0