Vivian Hwa

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7428663/publications.pdf

Version: 2024-02-01

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	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
2	Clinical phenotype and musculoskeletal characteristics of patients with aggrecan deficiency. American Journal of Medical Genetics, Part A, 2022, 188, 1193-1203.	0.7	2
3	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
4	STAT5B restrains human B-cell differentiation to maintain humoral immune homeostasis. Journal of Allergy and Clinical Immunology, 2022, 150, 931-946.	1.5	19
5	Genetic causes of growth hormone insensitivity beyond GHR. Reviews in Endocrine and Metabolic Disorders, 2021, 22, 43-58.	2.6	13
6	Developmental Adaptive Immune Defects Associated with STAT5B Deficiency in Three Young Siblings. Journal of Clinical Immunology, 2021, 41, 136-146.	2.0	9
7	Human growth disorders associated with impaired GH action: Defects in STAT5B and JAK2. Molecular and Cellular Endocrinology, 2021, 519, 111063.	1.6	16
8	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
9	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
10	Prenatal diagnosis of Proteus syndrome: Diagnosis of an AKT1 mutation from amniocytes. Birth Defects Research, 2020, 112, 1733-1737.	0.8	4
11	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
12	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
13	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
14	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
15	Nonclassical GH Insensitivity: Characterization of Mild Abnormalities of GH Action. Endocrine Reviews, 2019, 40, 476-505.	8.9	32
16	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
17	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
18	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	9.4	110

#	Article	IF	Citations
19	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
20	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
21	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
22	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
23	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
24	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
25	Dominant-negative STAT5B mutations cause growth hormone insensitivity with short stature and mild immune dysregulation. Nature Communications, 2018, 9, 2105.	5.8	81
26	Growth Hormone Insensitivity. , 2018, , 81-93.		0
27	Biology of the somatotroph axis (after the pituitary). Annales D'Endocrinologie, 2017, 78, 80-82.	0.6	6
28	Expanding Genetic and Functional Diagnoses of <i>IGF1R</i> Haploinsufficiencies. Hormone Research in Paediatrics, 2017, 87, 412-422.	0.8	18
29	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
30	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
31	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
32	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
33	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
34	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
35	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
36	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

#	Article	IF	Citations
37	STAT5B deficiency: Impacts on human growth and immunity. Growth Hormone and IGF Research, 2016, 28, 16-20.	0.5	80
38	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
39	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
40	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
41	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
42	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
43	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
44	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
45	Differentiating the roles of STAT5B and STAT5A in human CD4+ T cells. Clinical Immunology, 2013, 148, 227-236.	1.4	40
46	Severe Growth Deficiency is Associated with STAT5b Mutations that Disrupt Protein Folding and Activity. Molecular Endocrinology, 2013, 27, 150-161.	3.7	15
47	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
48	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
49	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
50	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
51	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
52	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
53	Acid-labile subunit (ALS) deficiency. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 101-113.	2.2	60
54	STAT5b deficiency: Lessons from STAT5b gene mutations. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 61-75.	2.2	100

#	Article	IF	Citations
55	STAT5b Deficiency: An Unsuspected Cause of Growth Failure, Immunodeficiency, and Severe Pulmonary Disease. Journal of Pediatrics, 2011, 158, 701-708.	0.9	110
56	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
57	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
58	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
59	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
60	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
61	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
62	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
63	Human Acid-Labile Subunit Deficiency: Clinical, Endocrine and Metabolic Consequences. Hormone Research, 2009, 72, 129-141.	1.8	109
64	The Growth Hormone Cascade and Its Role in Mammalian Growth. Hormone Research in Paediatrics, 2009, 71, 36-40.	0.8	74
65	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
66	IGFBPs and Cancer. Novartis Foundation Symposium, 2008, , 215-234.	1.2	9
67	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
68	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
69	Defects in growth hormone receptor signaling. Trends in Endocrinology and Metabolism, 2007, 18, 134-141.	3.1	134
70	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
71	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
72	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

#	Article	IF	Citations
73	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
74	Aberrant Folding of a Mutant Stat5b Causes Growth Hormone Insensitivity and Proteasomal Dysfunction. Journal of Biological Chemistry, 2006, 281, 6552-6558.	1.6	28
75	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
76	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
77	Growth Hormone Insensitivity Associated with aSTAT5bMutation. New England Journal of Medicine, 2003, 349, 1139-1147.	13.9	492
78	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
79	Connective Tissue Growth Factor (IGFBP-rP2) Expression and Regulation in Cultured Bovine Endothelial Cells*. Endocrinology, 1999, 140, 1575-1580.	1.4	35
80	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
81	The Insulin-Like Growth Factor-Binding Protein (IGFBP) Superfamily*. Endocrine Reviews, 1999, 20, 761-787.	8.9	832
82	The Insulin-like Growth Factor Binding Protein Superfamily: New Perspectives. Pediatrics, 1999, 104, 1018-1021.	1.0	43
83	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
84	The Neisseria meningitidis haemoglobin receptor: its role in iron utilization and virulence. Molecular Microbiology, 1995, 15, 531-541.	1.2	214
85	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