Andrew Dauber

List of Publications by Year
in descending order

78 papers	2,971 citations	172386 29 h-index	182361 51 g-index
81 all docs	81 docs citations	81 times ranked	4841 citing authors

#	Article	IF	Citations
1	Clinical Utility of Anti-Mullerian Hormone in Pediatrics. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 309-323.	1.8	13
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	Longitudinal Associations between Low Serum Bicarbonate and Linear Growth in Children with CKD. Kidney360, 2022, 3, 666-676.	0.9	9
4	Response to Letter to the Editor: â€œA Genome-Wide Pharmacogenetic Study of Growth Hormone Responsivenessâ€• Journal of Clinical Endocrinology and Metabolism, 2021, 106, e409-e410.	1.8	0
5	Developmental Adaptive Immune Defects Associated with STAT5B Deficiency in Three Young Siblings. Journal of Clinical Immunology, 2021, 41, 136-146.	2.0	9
6	Growth Hormone and Insulin-Like Growth Factor Dysregulation in Pediatric Chronic Kidney Disease. Hormone Research in Paediatrics, 2021, 94, 105-114.	0.8	6
7	High-throughput splicing assays identify missense and silent splice-disruptive POU1F1 variants underlying pituitary hormone deficiency. American Journal of Human Genetics, 2021, 108, 1526-1539.	2.6	23
8	Racial and Ethnic Disparities in the Investigation and Treatment of Growth Hormone Deficiency. Journal of Pediatrics, 2021, 236, 238-245.	0.9	10
9	Growth Hormone Stimulation Testing Patterns Contribute to Sex Differences in Pediatric Growth Hormone Treatment. Hormone Research in Paediatrics, 2021, 94, 353-363.	0.8	10
10	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
11	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	1.1	30
12	Algorithm-Driven Electronic Health Record Notification Enhances the Detection of Turner Syndrome. Journal of Pediatrics, 2020, 216, 227-231.	0.9	5
13	Response to Letter to the Editor: â€œGenetic Testing for the Child with Short Stature: Has the Time Come to Change Our Diagnostic Paradigm?â€• Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1927-e1927.	1.8	0
14	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
15	A Genome-Wide Pharmacogenetic Study of Growth Hormone Responsiveness. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3203-3214.	1.8	16
16	Arm Span and Its Relation to Height in a 2- to 17-Year-Old Reference Population and Heterozygous Carriers of ACAN Variants. Hormone Research in Paediatrics, 2020, 93, 164-172.	0.8	13
17	Disorders caused by genetic defects associated with GH-dependent genes: PAPPA2 defects. Molecular and Cellular Endocrinology, 2020, 518, 110967.	1.6	12
18	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

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19	Protein QTL analysis of IGF-I and its binding proteins provides insights into growth biology. Human Molecular Genetics, 2020, 29, 2625-2636.	1.4	2
20	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
21	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198.	0.9	36
22	Diagnosis, Genetics, and Therapy of Short Stature in Children: A Growth Hormone Research Society International Perspective. Hormone Research in Paediatrics, 2019, 92, 1-14.	0.8	181
23	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
24	Genetic Testing for the Child With Short Statureâ€"Has the Time Come To Change Our Diagnostic Paradigm?. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2766-2769.	1.8	23
25	Nonclassical GH Insensitivity: Characterization of Mild Abnormalities of GH Action. Endocrine Reviews, 2019, 40, 476-505.	8.9	32
26	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	2.6	88
27	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
28	Growth and Clinical Characteristics of Children with Floating-Harbor Syndrome: Analysis of Current Original Data and a Review of the Literature. Hormone Research in Paediatrics, 2019, 92, 115-123.	0.8	7
29	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
30	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	9.4	110
31	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
32	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
33	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
34	New genetic tools in the diagnosis of growth defects. Growth Hormone and IGF Research, 2018, 38, 24-28.	0.5	3
35	Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. Hormone Research in Paediatrics, 2018, 90, 407-413.	0.8	10
36	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

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37	The Clinical Cases of Geleophysic Dysplasia: One Gene, Different Phenotypes. Case Reports in Endocrinology, 2018, 2018, 1-7.	0.2	9
38	Insights and Implications of Genome-Wide Association Studies of Height. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3155-3168.	1.8	31
39	Dominant-negative STAT5B mutations cause growth hormone insensitivity with short stature and mild immune dysregulation. Nature Communications, 2018, 9, 2105.	5.8	81
40	Ezh2 Mutations Found in the Weaver Overgrowth Syndrome Cause a Partial Loss of H3K27 Histone Methyltransferase Activity. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 1470-1478.	1.8	33
41	Genetic Evaluation of 114 Chinese Short Stature Children in the Next Generation Era: a Single Center Study. Cellular Physiology and Biochemistry, 2018, 49, 295-305.	1.1	28
42	Paternally Inherited DLK1 Deletion Associated With Familial Central Precocious Puberty. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1557-1567.	1.8	145
43	Expanding Genetic and Functional Diagnoses of <i>IGF1R</i> Haploinsufficiencies. Hormone Research in Paediatrics, 2017, 87, 412-422.	0.8	18
44	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
45	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
46	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
47	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
48	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
49	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
50	Genomic insights into growth and its disorders. Current Opinion in Endocrinology, Diabetes and Obesity, 2016, 23, 51-56.	1.2	10
51	Determinants of Power in Gene-Based Burden Testing for Monogenic Disorders. American Journal of Human Genetics, 2016, 99, 527-539.	2.6	39
52	Treatment With Recombinant Human Insulin-Like Growth Factor-1 Improves Growth in Patients With PAPP-A2 Deficiency. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3879-3883.	1.8	40
53	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
54	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

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55	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
56	De novo mutations in ARID1B associated with both syndromic and non-syndromic short stature. BMC Genomics, 2015, 16, 701.	1.2	27
57	Insights from exome sequencing for endocrine disorders. Nature Reviews Endocrinology, 2015, 11, 455-464.	4.3	32
58	Short and tall stature: a new paradigm emerges. Nature Reviews Endocrinology, 2015, 11, 735-746.	4.3	212
59	Idiopathic short stature due to novel heterozygous mutation of the aggrecan gene. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 927-32.	0.4	44
60	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
61	Whole Exome Sequencing to Identify Genetic Causes of Short Stature. Hormone Research in Paediatrics, 2014, 82, 44-52.	0.8	76
62	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
63	A Novel Deletion of IGF1 in a Patient With Idiopathic Short Stature Provides Insight Into IGF1 Haploinsufficiency. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E153-E159.	1.8	38
64	Short Stature, Accelerated Bone Maturation, and Early Growth Cessation Due to Heterozygous Aggrecan Mutations. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1510-E1518.	1.8	109
65	Genetic Evaluation of Short Stature. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3080-3092.	1.8	128
66	Whole Exome Sequencing Reveals a Novel Mutation in CUL7 in a Patient with an Undiagnosed Growth Disorder. Journal of Pediatrics, 2013, 162, 202-204.e1.	0.9	32
67	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. American Journal of Human Genetics, 2013, 93, 798-811.	2.6	82
68	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
69	Redefining the progeroid form of ehlersâ€“danlos syndrome: Report of the fourth patient with B4GALT7 deficiency and review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 2519-2527.	0.7	31
70	Closed-Loop Insulin Therapy Improves Glycemic Control in Children Aged <7 Years. Diabetes Care, 2013, 36, 222-227.	4.3	70
71	Genetic Defect inCYP24A1, the Vitamin D 24-Hydroxylase Gene, in a Patient with Severe Infantile Hypercalcemia. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E268-E274.	1.8	113
72	Novel Microcephalic Primordial Dwarfism Disorder Associated with Variants in the Centrosomal Protein Ninein. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2140-E2151.	1.8	64

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73	Genome-wide Association of Copy-Number Variation Reveals an Association between Short Stature and the Presence of Low-Frequency Genomic Deletions. American Journal of Human Genetics, 2011, 89, 751-759.	2.6	63
74	Genome-Wide Association Studies in Pediatric Endocrinology. Hormone Research in Paediatrics, 2011, 75, 322-328.	0.8	5
75	Delayed Puberty Due to a Novel Mutation in CHD7 Causing CHARGE Syndrome. Pediatrics, 2010, 126, e1594-e1598.	1.0	16
76	Nocturnal Dexamethasone versus Hydrocortisone for the Treatment of Children with Congenital Adrenal Hyperplasia. International Journal of Pediatric Endocrinology (Springer), 2010, 2010, 1-8.	1.6	8
77	Monitoring of Therapy in Congenital Adrenal Hyperplasia. Clinical Chemistry, 2010, 56, 1245-1251.	1.5	58
78	Procalcitonin Levels in Febrile Infants After Recent Immunization. Pediatrics, 2008, 122, e1119-e1122.	1.0	11