Catherine S Choong

List of Publications by Year
in descending order

72	2,682	257101 24	189595 50
papers	citations	h-index	g-index

73	73	73	3511
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gigantism and Acromegaly Due to Xq26 Microduplications and GPR101 Mutation. New England Journal of Medicine, 2014, 371, 2363-2374.	13.9	292
2	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	3.8	241
3	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	1.1	173
4	Clinical and genetic characterization of pituitary gigantism: an international collaborative study in 208 patients. Endocrine-Related Cancer, 2015, 22, 745-757.	1.6	155
5	X-linked acrogigantism syndrome: clinical profile and therapeutic responses. Endocrine-Related Cancer, 2015, 22, 353-367.	1.6	151
6	Germ-line and somatic DICER1 mutations in pineoblastoma. Acta Neuropathologica, 2014, 128, 583-595.	3.9	137
7	Trinucleotide repeats in the human androgen receptor: a molecular basis for disease. Journal of Molecular Endocrinology, 1998, 21, 235-257.	1.1	123
8	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56.	2.4	110
9	Androgen receptor levels in prostate cancer epithelial and peritumoral stromal cells identify non-organ confined disease. Prostate, 2005, 63, 19-28.	1.2	103
10	Growth Hormone Research Society perspective on the development of long-acting growth hormone preparations. European Journal of Endocrinology, 2016, 174, C1-C8.	1.9	99
11	Evolution of the Primate Androgen Receptor: A Structural Basis for Disease. Journal of Molecular Evolution, 1998, 47, 334-342.	0.8	72
12	Congenital thyrotoxicosis in premature infants. Clinical Endocrinology, 2001, 54, 371-376.	1.2	56
13	Combined pituitary hormone deficiency in Australian children: clinical and genetic correlates. Clinical Endocrinology, 2003, 58, 785-794.	1.2	50
14	Elevated levels of HERâ€2/neu and androgen receptor in clinically localized prostate cancer identifies metastatic potential. Prostate, 2008, 68, 830-838.	1.2	43
15	Ovarian Sex Cord-Stromal Tumors in Patients With Probable or Confirmed Germline DICER1 Mutations. International Journal of Gynecological Pathology, 2015, 34, 266-274.	0.9	39
16	Growth Hormone Research Society perspective on biomarkers of GH action in children and adults. Endocrine Connections, 2018, 7, R126-R134.	0.8	39
17	A novel missense mutation in the amino-terminal domain of the human androgen receptor gene in a family with partial androgen insensitivity syndrome causes reduced efficiency of protein translation.. Journal of Clinical Investigation, 1996, 98, 1423-1431.	3.9	38
18	The outcome in Australian children with hyperinsulinism of infancy: early extensive surgery in severe cases lowers risk of diabetes. Clinical Endocrinology, 2003, 58, 355-364.	1.2	37

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19	Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2247-e2260.	1.8	37
20	Parental preâ€pregnancy BMI is a dominant earlyâ€life risk factor influencing BMI of offspring in adulthood.. Obesity Science and Practice, 2016, 2, 48-57.	1.0	33
21	Sequencing of DICER1 in sarcomas identifies biallelic somatic DICER1 mutations in an adult-onset embryonal rhabdomyosarcoma. British Journal of Cancer, 2017, 116, 1621-1626.	2.9	30
22	Overexpression of Aromatase Associated With Loss of Heterozygosity of theSTK11Gene Accounts for Prepubertal Gynecomastia in Boys with Peutz-Jeghers Syndrome. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1979-E1987.	1.8	29
23	Growth hormone treatment for Turner syndrome in Australia reveals that younger age and increased dose interact to improve response. Clinical Endocrinology, 2011, 74, 473-480.	1.2	27
24	Short stature and hypoparathyroidism in a child with Kenny-Caffey syndrome type 2 due to a novel mutation in FAM111A gene. International Journal of Pediatric Endocrinology (Springer), 2017, 2017, 1.	1.6	27
25	Exploring the endocrine manifestations of DICER1 mutations. Trends in Molecular Medicine, 2012, 18, 503-505.	3.5	26
26	Exercise training improves vascular function and secondary health measures in survivors of pediatric oncology related cerebral insult. PLoS ONE, 2018, 13, e0201449.	1.1	25
27	Sertoli-Leydig Cell Tumor of the Ovary, a Rare Cause of Precocious Puberty in a 12-Month-Old Infant. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 49-56.	1.8	24
28	A novel NR 5 A 1 variant in an infant with elevated testosterone from an A ustralasian cohort of 46, XY patients with disorders of sex development. Clinical Endocrinology, 2013, 78, 545-550.	1.2	24
29	Gender Bias in Children Receiving Growth Hormone Treatment. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1191-1198.	1.8	22
30	Childhood craniopharyngioma: 20â€year institutional experience in <scp>W</scp>estern <scp>A</scp>ustralia. Journal of Paediatrics and Child Health, 2013, 49, 403-408.	0.4	22
31	Functional characterization of multiple DICER1 mutations in an adolescent. Endocrine-Related Cancer, 2016, 23, L1-L5.	1.6	22
32	Angiotensinogen gene T235 variant: a marker for the development of persistent microalbuminuria in children and adolescents with type 1 diabetes mellitus. Journal of Diabetes and Its Complications, 2008, 22, 191-198.	1.2	20
33	Growth hormone regimens in Australia: analysis of the first 3â€ƒyears of treatment for idiopathic growth hormone deficiency and idiopathic short stature. Clinical Endocrinology, 2012, 77, 62-71.	1.2	20
34	Sterile abscess formation associated with depot leuprorelin acetate therapy for central precocious puberty. Journal of Paediatrics and Child Health, 2012, 48, E136-9.	0.4	20
35	Transient pseudohypoaldosteronism in infancy secondary to urinary tract infection. Journal of Paediatrics and Child Health, 2017, 53, 458-463.	0.4	20
36	Efficacy of Hydrochlorothiazide and low renal solute feed in Neonatal Central Diabetes Insipidus with transition to Oral Desmopressin in early infancy. International Journal of Pediatric Endocrinology (Springer), 2014, 2014, 11.	1.6	18

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37	Comparison of <scp>C</scp>enters for <scp>D</scp>isease <scp>C</scp>ontrol and <scp>P</scp>revention and <scp>W</scp>orld <scp>H</scp>ealth <scp>O</scp>rganization references/standards for height in contemporary <scp>A</scp>ustralian children: Analyses of the <scp>R</scp>aine <scp>S</scp>tudy and <scp>A</scp>ustralian <scp>N</scp>ational <scp>C</scp>hildren's <scp>N</scp>utrition and <scp>P</scp>hysical <scp>A</scp>ctivity cohorts. Journal of Paediatrics and Child Health, 2014, 50, 895-901.	0.4	16
38	NKX2-1 mutations in brain-lung-thyroid syndrome: a case series of four patients. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 373-8.	0.4	16
39	Early cessation and non-response are important and possibly related problems in growth hormone therapy: An OZGROW analysis. Growth Hormone and IGF Research, 2016, 29, 63-70.	0.5	16
40	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
41	Ski-interacting protein (SKIP) interacts with androgen receptor in the nucleus and modulates androgen-dependent transcription. BMC Biochemistry, 2013, 14, 10.	4.4	14
42	Low dose growth hormone treatment in infants and toddlers with Prader-Willi syndrome is comparable to higher dosage regimens. Growth Hormone and IGF Research, 2017, 34, 1-7.	0.5	14
43	A novel, homozygous mutation in desert hedgehog (DHH) in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. International Journal of Pediatric Endocrinology (Springer), 2018, 2018, 2.	1.6	14
44	Genotype-Phenotype Associations in Patients with Severe Hyperinsulinism of Infancy. Pediatric and Developmental Pathology, 2007, 10, 25-34.	0.5	13
45	NNTPseudoexon Activation as a Novel Mechanism for Disease in Two Siblings With Familial Glucocorticoid Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E350-E354.	1.8	13
46	Lessons learnt during the COVID â€19 pandemic: Why Australian schools should be prioritised to stay open. Journal of Paediatrics and Child Health, 2021, 57, 1362-1369.	0.4	12
47	Requirements for improving health and wellâ€being of children with Praderâ€Willi syndrome and their families. Journal of Paediatrics and Child Health, 2019, 55, 1029-1037.	0.4	11
48	Functional analysis of novel desert hedgehog gene variants improves the clinical interpretation of genomic data and provides a more accurate diagnosis for patients with 46,XY differences of sex development. Journal of Medical Genetics, 2019, 56, 434-443.	1.5	11
49	Comparison of weightâ€•vs body surface areaâ€based growth hormone dosing for children: implications for response. Clinical Endocrinology, 2014, 80, 384-394.	1.2	10
50	Report and review of described associations of <scp>M</scp>ayerâ€<scp>R</scp>okitanskyâ€<scp>K</scp>Ã¼sterâ€<scp>H</scp>auser syndrome and <scp>S</scp>ilverâ€“<scp>R</scp>ussell syndrome. Journal of Paediatrics and Child Health, 2015, 51, 555-560.	0.4	10
51	Sleepâ€disordered breathing in Australian children with Praderâ€Willi syndrome following initiation of growth hormone therapy. Journal of Paediatrics and Child Health, 2022, 58, 248-255.	0.4	10
52	Increased Body Mass Index during Therapy for Childhood Acute Lymphoblastic Leukemia: A Significant and Underestimated Complication. International Journal of Pediatrics (United Kingdom), 2015, 2015, 1-10.	0.2	7
53	Congenital varicella syndrome in the absence of cutaneous lesions. Journal of Paediatrics and Child Health, 2000, 36, 184-185.	0.4	6
54	GH secretagogue receptor gene polymorphisms are associated with stature throughout childhood. European Journal of Endocrinology, 2012, 166, 1079-1085.	1.9	6

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55	Completeness of protocols for clinical trials in children submitted to ethics committees. Journal of Paediatrics and Child Health, 2019, 55, 291-298.	0.4	6
56	Fitness, body composition and vascular health in adolescent and young adult survivors of paediatric brain cancer and cranial radiotherapy. International Journal of Adolescent Medicine and Health, 2019, 31, .	0.6	6
57	The utility of continuous glucose monitoring systems in the management of children with persistent hypoglycaemia. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1567-1572.	0.4	6
58	Reduced expression and normal nucleotide sequence of androgen receptor gene coding and promoter regions in a family with partial androgen insensitivity syndrome. Clinical Endocrinology, 1997, 46, 281-288.	1.2	5
59	The influence of secular trend for height on ascertainment and eligibility for growth hormone treatment. Clinical Endocrinology, 2010, 73, 760-768.	1.2	5
60	Structural chromosome disruption of the NR3C2 gene causing pseudohypoaldosteronism type 1 presenting in infancy. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 555-9.	0.4	5
61	Response to growth hormone treatment in <scp>P</scp>raderâ€“<scp>W</scp>illi syndrome: Auxological criteria versus genetic diagnosis. Journal of Paediatrics and Child Health, 2013, 49, 1045-1051.	0.4	5
62	Metabolic and Psychological Impact of a Pragmatic Exercise Intervention Program in Adolescent and Young Adult Survivors of Pediatric Cancer-Related Cerebral Insult. Journal of Adolescent and Young Adult Oncology, 2018, 7, 349-357.	0.7	5
63	Variable persistence of serum testosterone in infants and children exposed to topical testosterone. Journal of Paediatrics and Child Health, 2020, 56, 1464-1467.	0.4	5
64	Further heterogeneity in Silverâ€“Russell syndrome: PLAG1 deletion in association with a complex chromosomal rearrangement. American Journal of Medical Genetics, Part A, 2021, 185, 3136-3145.	0.7	4
65	Using a trauma informed practice framework to enhance understanding of and identify support strategies for behavioural difficulties in young people with Prader-Willi syndrome. Research in Developmental Disabilities, 2021, 110, 103839.	1.2	3
66	A Case Report of Syndromic Multinodular Goitre in Adolescence: Exploring the Phenotype Overlap between Cowden and DICER1 Syndromes. European Thyroid Journal, 2018, 7, 44-50.	1.2	2
67	Gravesâ€™ disease presenting as lifeâ€threatening hypokalaemic periodic paralysis. Journal of Paediatrics and Child Health, 2018, 54, 443-445.	0.4	2
68	Early markers of cardiovascular injury in childhood leukaemia survivors treated with anthracycline chemotherapy. Cardio-Oncology, 2019, 5, 11.	0.8	2
69	Cardiovascular Testing Detects Underlying Dysfunction in Childhood Leukemia Survivors. Medicine and Science in Sports and Exercise, 2020, 52, 525-534.	0.2	2
70	Monogenic diabetes due to an INSR mutation in a child with severe insulin resistance. Endocrinology, Diabetes and Metabolism Case Reports, 2022, 2022, .	0.2	1
71	Strengths and challenging behaviors in children and adolescents with<scp>Praderâ€“Willi</scp>syndrome: Two sides to the coin. American Journal of Medical Genetics, Part A, 2022, 188, 1488-1496.	0.7	0
72	Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome. European Journal of Pediatrics, 2022, , 1.	1.3	0