

# Catherine S Choong

## List of Publications by Year in descending order

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Version: 2024-02-01

72  
papers

2,682  
citations

257101

24  
h-index

189595

50  
g-index

73  
all docs

73  
docs citations

73  
times ranked

3511  
citing authors

#	ARTICLE	IF	CITATIONS
1	Sleep-disordered breathing in Australian children with Prader-Willi syndrome following initiation of growth hormone therapy. <i>Journal of Paediatrics and Child Health</i> , 2022, 58, 248-255.	0.4	10
2	Monogenic diabetes due to an INSR mutation in a child with severe insulin resistance. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2022, 2022, .	0.2	1
3	Strengths and challenging behaviors in children and adolescents with Prader-Willi syndrome: Two sides to the coin. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1488-1496.	0.7	0
4	Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome. <i>European Journal of Pediatrics</i> , 2022, , 1.	1.3	0
5	Using a trauma informed practice framework to enhance understanding of and identify support strategies for behavioural difficulties in young people with Prader-Willi syndrome. <i>Research in Developmental Disabilities</i> , 2021, 110, 103839.	1.2	3
6	Lessons learnt during the COVID-19 pandemic: Why Australian schools should be prioritised to stay open. <i>Journal of Paediatrics and Child Health</i> , 2021, 57, 1362-1369.	0.4	12
7	Further heterogeneity in Silver-Russell syndrome: PLAG1 deletion in association with a complex chromosomal rearrangement. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3136-3145.	0.7	4
8	The utility of continuous glucose monitoring systems in the management of children with persistent hypoglycaemia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 1567-1572.	0.4	6
9	Cardiovascular Testing Detects Underlying Dysfunction in Childhood Leukemia Survivors. <i>Medicine and Science in Sports and Exercise</i> , 2020, 52, 525-534.	0.2	2
10	Variable persistence of serum testosterone in infants and children exposed to topical testosterone. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1464-1467.	0.4	5
11	Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e2247-e2260.	1.8	37
12	Requirements for improving health and well-being of children with Prader-Willi syndrome and their families. <i>Journal of Paediatrics and Child Health</i> , 2019, 55, 1029-1037.	0.4	11
13	Early markers of cardiovascular injury in childhood leukaemia survivors treated with anthracycline chemotherapy. <i>Cardio-Oncology</i> , 2019, 5, 11.	0.8	2
14	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. <i>Journal of Medical Genetics</i> , 2019, 56, 434-443.	1.5	11
15	Completeness of protocols for clinical trials in children submitted to ethics committees. <i>Journal of Paediatrics and Child Health</i> , 2019, 55, 291-298.	0.4	6
16	Fitness, body composition and vascular health in adolescent and young adult survivors of paediatric brain cancer and cranial radiotherapy. <i>International Journal of Adolescent Medicine and Health</i> , 2019, 31, .	0.6	6
17	Growth Hormone Research Society perspective on biomarkers of GH action in children and adults. <i>Endocrine Connections</i> , 2018, 7, R126-R134.	0.8	39
18	A Case Report of Syndromic Multinodular Goitre in Adolescence: Exploring the Phenotype Overlap between Cowden and DICER1 Syndromes. <i>European Thyroid Journal</i> , 2018, 7, 44-50.	1.2	2

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19	Graves's disease presenting as life-threatening hypokalaemic periodic paralysis. <i>Journal of Paediatrics and Child Health</i> , 2018, 54, 443-445.	0.4	2
20	A novel, homozygous mutation in desert hedgehog (DHH) in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2018, 2018, 2.	1.6	14
21	Metabolic and Psychological Impact of a Pragmatic Exercise Intervention Program in Adolescent and Young Adult Survivors of Pediatric Cancer-Related Cerebral Insult. <i>Journal of Adolescent and Young Adult Oncology</i> , 2018, 7, 349-357.	0.7	5
22	Exercise training improves vascular function and secondary health measures in survivors of pediatric oncology related cerebral insult. <i>PLoS ONE</i> , 2018, 13, e0201449.	1.1	25
23	Transient pseudohypoadosteronism in infancy secondary to urinary tract infection. <i>Journal of Paediatrics and Child Health</i> , 2017, 53, 458-463.	0.4	20
24	Sequencing of DICER1 in sarcomas identifies biallelic somatic DICER1 mutations in an adult-onset embryonal rhabdomyosarcoma. <i>British Journal of Cancer</i> , 2017, 116, 1621-1626.	2.9	30
25	Low dose growth hormone treatment in infants and toddlers with Prader-Willi syndrome is comparable to higher dosage regimens. <i>Growth Hormone and IGF Research</i> , 2017, 34, 1-7.	0.5	14
26	Short stature and hypoparathyroidism in a child with Kenny-Caffey syndrome type 2 due to a novel mutation in FAM111A gene. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2017, 2017, 1.	1.6	27
27	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	3.8	241
28	Growth Hormone Research Society perspective on the development of long-acting growth hormone preparations. <i>European Journal of Endocrinology</i> , 2016, 174, C1-C8.	1.9	99
29	Early cessation and non-response are important and possibly related problems in growth hormone therapy: An OZGROW analysis. <i>Growth Hormone and IGF Research</i> , 2016, 29, 63-70.	0.5	16
30	Parental pre-pregnancy BMI is a dominant early-life risk factor influencing BMI of offspring in adulthood. <i>Obesity Science and Practice</i> , 2016, 2, 48-57.	1.0	33
31	Germline or somatic GPR101 duplication leads to X-linked acroigantism: a clinico-pathological and genetic study. <i>Acta Neuropathologica Communications</i> , 2016, 4, 56.	2.4	110
32	Functional characterization of multiple DICER1 mutations in an adolescent. <i>Endocrine-Related Cancer</i> , 2016, 23, L1-L5.	1.6	22
33	Ovarian Sex Cord-Stromal Tumors in Patients With Probable or Confirmed Germline DICER1 Mutations. <i>International Journal of Gynecological Pathology</i> , 2015, 34, 266-274.	0.9	39
34	Increased Body Mass Index during Therapy for Childhood Acute Lymphoblastic Leukemia: A Significant and Underestimated Complication. <i>International Journal of Pediatrics (United Kingdom)</i> , 2015, 2015, 1-10.	0.2	7
35	Pseudoexon Activation as a Novel Mechanism for Disease in Two Siblings With Familial Glucocorticoid Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E350-E354.	1.8	13
36	Report and review of described associations of McCune-Albright syndrome and Silver-Russell syndrome. <i>Journal of Paediatrics and Child Health</i> , 2015, 51, 555-560.	0.4	10

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37	Efficacy and safety of sirolimus in a neonate with persistent hypoglycaemia following near-total pancreatectomy for hyperinsulinaemic hypoglycaemia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015, 28, 1391-8.	0.4	15
38	Clinical and genetic characterization of pituitary gigantism: an international collaborative study in 208 patients. <i>Endocrine-Related Cancer</i> , 2015, 22, 745-757.	1.6	155
39	X-linked acrogigantism syndrome: clinical profile and therapeutic responses. <i>Endocrine-Related Cancer</i> , 2015, 22, 353-367.	1.6	151
40	Germ-line and somatic DICER1 mutations in pineoblastoma. <i>Acta Neuropathologica</i> , 2014, 128, 583-595.	3.9	137
41	Gigantism and Acromegaly Due to Xq26 Microduplications and <i>GPR101</i> Mutation. <i>New England Journal of Medicine</i> , 2014, 371, 2363-2374.	13.9	292
42	Comparison of Centers for Disease Control and Prevention and World Health Organization references/standards for height in contemporary Australian children: Analyses of the Raine Study and Australian National Children's Nutrition and Physical Activity cohorts. <i>Journal of Paediatrics and Child Health</i> , 2014, 50, 895-901.	0.4	16
43	Comparison of weight vs body surface area based growth hormone dosing for children: implications for response. <i>Clinical Endocrinology</i> , 2014, 80, 384-394.	1.2	10
44	Efficacy of Hydrochlorothiazide and low renal solute feed in Neonatal Central Diabetes Insipidus with transition to Oral Desmopressin in early infancy. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2014, 2014, 11.	1.6	18
45	NKX2-1 mutations in brain-lung-thyroid syndrome: a case series of four patients. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 373-8.	0.4	16
46	Ski-interacting protein (SKIP) interacts with androgen receptor in the nucleus and modulates androgen-dependent transcription. <i>BMC Biochemistry</i> , 2013, 14, 10.	4.4	14
47	Childhood craniopharyngioma: 20-year institutional experience in Western Australia. <i>Journal of Paediatrics and Child Health</i> , 2013, 49, 403-408.	0.4	22
48	A novel NR5A1 variant in an infant with elevated testosterone from an Australasian cohort of 46, XY patients with disorders of sex development. <i>Clinical Endocrinology</i> , 2013, 78, 545-550.	1.2	24
49	Overexpression of Aromatase Associated With Loss of Heterozygosity of the <i>STK11</i> Gene Accounts for Prepubertal Gynecomastia in Boys with Peutz-Jeghers Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E1979-E1987.	1.8	29
50	Response to growth hormone treatment in Prader-Willi syndrome: Auxological criteria versus genetic diagnosis. <i>Journal of Paediatrics and Child Health</i> , 2013, 49, 1045-1051.	0.4	5
51	GH secretagogue receptor gene polymorphisms are associated with stature throughout childhood. <i>European Journal of Endocrinology</i> , 2012, 166, 1079-1085.	1.9	6
52	Exploring the endocrine manifestations of DICER1 mutations. <i>Trends in Molecular Medicine</i> , 2012, 18, 503-505.	3.5	26
53	Growth hormone regimens in Australia: analysis of the first 3 years of treatment for idiopathic growth hormone deficiency and idiopathic short stature. <i>Clinical Endocrinology</i> , 2012, 77, 62-71.	1.2	20
54	Sterile abscess formation associated with depot leuporelin acetate therapy for central precocious puberty. <i>Journal of Paediatrics and Child Health</i> , 2012, 48, E136-9.	0.4	20

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55	Growth hormone treatment for Turner syndrome in Australia reveals that younger age and increased dose interact to improve response. <i>Clinical Endocrinology</i> , 2011, 74, 473-480.	1.2	27
56	Extending the phenotypes associated with <i>DICER1</i> mutations. <i>Human Mutation</i> , 2011, 32, 1381-1384.	1.1	173
57	Structural chromosome disruption of the <i>NR3C2</i> gene causing pseudohypoaldosteronism type 1 presenting in infancy. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2011, 24, 555-9.	0.4	5
58	The influence of secular trend for height on ascertainment and eligibility for growth hormone treatment. <i>Clinical Endocrinology</i> , 2010, 73, 760-768.	1.2	5
59	Gender Bias in Children Receiving Growth Hormone Treatment. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 1191-1198.	1.8	22
60	Elevated levels of HER2/neu and androgen receptor in clinically localized prostate cancer identifies metastatic potential. <i>Prostate</i> , 2008, 68, 830-838.	1.2	43
61	Angiotensinogen gene T235 variant: a marker for the development of persistent microalbuminuria in children and adolescents with type 1 diabetes mellitus. <i>Journal of Diabetes and Its Complications</i> , 2008, 22, 191-198.	1.2	20
62	Genotype-Phenotype Associations in Patients with Severe Hyperinsulinism of Infancy. <i>Pediatric and Developmental Pathology</i> , 2007, 10, 25-34.	0.5	13
63	Androgen receptor levels in prostate cancer epithelial and peritumoral stromal cells identify non-organ confined disease. <i>Prostate</i> , 2005, 63, 19-28.	1.2	103
64	The outcome in Australian children with hyperinsulinism of infancy: early extensive surgery in severe cases lowers risk of diabetes. <i>Clinical Endocrinology</i> , 2003, 58, 355-364.	1.2	37
65	Combined pituitary hormone deficiency in Australian children: clinical and genetic correlates. <i>Clinical Endocrinology</i> , 2003, 58, 785-794.	1.2	50
66	Sertoli-Leydig Cell Tumor of the Ovary, a Rare Cause of Precocious Puberty in a 12-Month-Old Infant. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 49-56.	1.8	24
67	Congenital thyrotoxicosis in premature infants. <i>Clinical Endocrinology</i> , 2001, 54, 371-376.	1.2	56
68	Congenital varicella syndrome in the absence of cutaneous lesions. <i>Journal of Paediatrics and Child Health</i> , 2000, 36, 184-185.	0.4	6
69	Evolution of the Primate Androgen Receptor: A Structural Basis for Disease. <i>Journal of Molecular Evolution</i> , 1998, 47, 334-342.	0.8	72
70	Trinucleotide repeats in the human androgen receptor: a molecular basis for disease. <i>Journal of Molecular Endocrinology</i> , 1998, 21, 235-257.	1.1	123
71	Reduced expression and normal nucleotide sequence of androgen receptor gene coding and promoter regions in a family with partial androgen insensitivity syndrome. <i>Clinical Endocrinology</i> , 1997, 46, 281-288.	1.2	5
72	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. <i>Journal of Clinical Investigation</i> , 1996, 98, 1423-1431.	3.9	38