

Yu Ding

List of Publications by Year in descending order

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Version: 2024-02-01

20
papers

260
citations

1051969

10
h-index

1113639

15
g-index

21
all docs

21
docs citations

21
times ranked

624
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical Profiles and Genetic Spectra of 814 Chinese Children With Short Stature. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 972-985.	1.8	17
2	Molecular and Phenotypic Expansion of Alstr�m Syndrome in Chinese Patients. <i>Frontiers in Genetics</i> , 2022, 13, 808919.	1.1	5
3	Evaluating the variety of GNAS inactivation disorders and their clinical manifestations in 11 Chinese children. <i>BMC Endocrine Disorders</i> , 2022, 22, 70.	0.9	3
4	Clinical and genetic analysis in a Chinese cohort of children and adolescents with diabetes/persistent hyperglycemia. <i>Journal of Diabetes Investigation</i> , 2021, 12, 48-62.	1.1	3
5	TRMA syndrome with a severe phenotype, cerebral infarction, and novel compound heterozygous SLC19A2 mutation: a case report. <i>BMC Pediatrics</i> , 2019, 19, 233.	0.7	6
6	Molecular and phenotypic spectrum of Noonan syndrome in Chinese patients. <i>Clinical Genetics</i> , 2019, 96, 290-299.	1.0	29
7	New insights from unbiased panel and whole-exome sequencing in a large Chinese cohort with disorders of sex development. <i>European Journal of Endocrinology</i> , 2019, 181, 311-323.	1.9	13
8	Evaluation of basal sex hormone levels for activation of the hypothalamicâ€“pituitaryâ€“gonadal axis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 323-329.	0.4	14
9	Novel compound heterozygous variants in the <i>LHCGR</i> gene identified in a subject with Leydig cell hypoplasia type 1. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 239-245.	0.4	9
10	SOPH Syndrome with Growth Hormone Deficiency, Normal Bone Age, and Novel Compound Heterozygous Mutations in <i>NBAS</i> . <i>Fetal and Pediatric Pathology</i> , 2018, 37, 404-410.	0.4	11
11	Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 178.	1.2	30
12	Increased transactivation and impaired repression of β -catenin-mediated transcription associated with a novel SOX3 missense mutation in an X-linked hypopituitarism pedigree with modest growth failure. <i>Molecular and Cellular Endocrinology</i> , 2018, 478, 133-140.	1.6	5
13	Clinical and molecular genetic characterization of two patients with mutations in the phosphoglucomutase 1 (<i>PGM1</i>) gene. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 781-788.	0.4	7
14	<i>De Novo</i> Mutation of <i>KAT6B</i> Gene Causing Atypical Sayâ€“Barberâ€“Bieseckerâ€“Youngâ€“Simpson Syndrome or Genitopatellar Syndrome. <i>Fetal and Pediatric Pathology</i> , 2017, 36, 130-138.	0.4	5
15	Novel pathogenic ACAN variants in non-syndromic short stature patients. <i>Clinica Chimica Acta</i> , 2017, 469, 126-129.	0.5	35
16	Turner syndrome caused by rare complex structural abnormalities involving chromosome X. <i>Experimental and Therapeutic Medicine</i> , 2017, 14, 2265-2270.	0.8	4
17	Biallelic mutations in GPD1 gene in a Chinese boy mainly presented with obesity, insulin resistance, fatty liver, and short stature. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3189-3194.	0.7	19
18	Clinical and Molecular Characterization of Patients with Fructose 1,6-Bisphosphatase Deficiency. <i>International Journal of Molecular Sciences</i> , 2017, 18, 857.	1.8	27

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19	Novel mutations in the CYP11B2 gene causing aldosterone synthase deficiency. <i>Molecular Medicine Reports</i> , 2016, 13, 3127-3132.	1.1	12
20	Clinical characteristics and follow-up of 5 young Chinese males with gonadotropin-releasing hormone deficiency caused by mutations in the KAL1 gene. <i>Meta Gene</i> , 2016, 7, 64-69.	0.3	5