

# Zhan Qi

## List of Publications by Year in descending order

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#	ARTICLE	IF	CITATIONS
1	Newborn screening with targeted sequencing: a multicenter investigation and a pilot clinical study in China. <i>Journal of Genetics and Genomics</i> , 2022, 49, 13-19.	3.9	19
2	Spectrum Analysis of Albinism Genes in a Large Cohort of Chinese Index Patients. <i>Journal of Investigative Dermatology</i> , 2022, 142, 1752-1755.e3.	0.7	6
3	Genetic variants and mutational spectrum of Chinese Hermanskyâ€Pudlak syndrome patients. <i>Pigment Cell and Melanoma Research</i> , 2021, 34, 111-121.	3.3	9
4	The first Hermanskyâ€Pudlak syndrome type 9 patient with two novel variants in Chinese population. <i>Journal of Dermatology</i> , 2021, 48, 676-680.	1.2	7
5	Exome sequencing as the firstâ€tier test for pediatric respiratory diseases: A singleâ€center study. <i>Human Mutation</i> , 2021, 42, 891-900.	2.5	2
6	Clinical Application of Whole Exome Sequencing for Monogenic Disorders in PICU of China. <i>Frontiers in Genetics</i> , 2021, 12, 677699.	2.3	6
7	Parallel Tests of Whole Exome Sequencing and Copy Number Variant Sequencing Increase the Diagnosis Yields of Rare Pediatric Disorders. <i>Frontiers in Genetics</i> , 2020, 11, 473.	2.3	8
8	Simpsonâ€Golabiâ€Behmel syndrome with 46,XY disorders of sex development: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 285-289.	1.2	1
9	Application of multiplex ligation-dependent probe amplification in the genetic testing of oculocutaneous albinism. <i>Chinese Medical Journal</i> , 2019, 132, 2011-2012.	2.3	3
10	Instability of BLOCâ€2 and BLOCâ€3 in Chinese patients with Hermanskyâ€Pudlak syndrome. <i>Pigment Cell and Melanoma Research</i> , 2019, 32, 373-380.	3.3	12
11	Neonates are armed with deviated immune cell proportion and cytokine reduction but higher T cell proliferation potentiality. <i>Acta Biochimica Et Biophysica Sinica</i> , 2018, 50, 934-937.	2.0	5
12	Whole-exome sequencing identified compound heterozygous variants in MMKS in a Chinese pedigree with Bardet-Biedl syndrome. <i>Science China Life Sciences</i> , 2017, 60, 739-745.	4.9	21
13	Clinical feature and waveform in infantile nystagmus syndrome in children with FRMD7 gene mutations. <i>Science China Life Sciences</i> , 2017, 60, 707-713.	4.9	8
14	Genetic Analysis and Follow-Up of 25 Neonatal Diabetes Mellitus Patients in China. <i>Journal of Diabetes Research</i> , 2016, 2016, 1-9.	2.3	28
15	Identification of AAAS gene mutation in Allgrove syndrome: A report of three cases. <i>Experimental and Therapeutic Medicine</i> , 2015, 10, 1277-1282.	1.8	16
16	Toll-like receptor 1 (TLR1) Gene SNP rs5743618 is associated with increased risk for tuberculosis in Han Chinese children. <i>Tuberculosis</i> , 2015, 95, 197-203.	1.9	33
17	rs2243268 and rs2243274 of Interleukin-4 (IL-4) gene are associated with reduced risk for extrapulmonary and severe tuberculosis in Chinese Han children. <i>Infection, Genetics and Evolution</i> , 2014, 23, 121-128.	2.3	14