

# Zhan Qi

## List of Publications by Year in descending order

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356  
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#	ARTICLE	IF	CITATIONS
1	Toll-like receptor 1(TLR1) Gene SNP rs5743618 is associated with increased risk for tuberculosis in Han Chinese children. Tuberculosis, 2015, 95, 197-203.	1.9	33
2	Genetic Analysis and Follow-Up of 25 Neonatal Diabetes Mellitus Patients in China. Journal of Diabetes Research, 2016, 2016, 1-9.	2.3	28
3	Whole-exome sequencing identified compound heterozygous variants in MMKS in a Chinese pedigree with Bardet-Biedl syndrome. Science China Life Sciences, 2017, 60, 739-745.	4.9	21
4	Newborn screening with targeted sequencing: a multicenter investigation and a pilot clinical study in China. Journal of Genetics and Genomics, 2022, 49, 13-19.	3.9	19
5	Identification of AAAS gene mutation in Allgrove syndrome: A report of three cases. Experimental and Therapeutic Medicine, 2015, 10, 1277-1282.	1.8	16
6	rs2243268 and rs2243274 of Interleukin-4 (IL-4) gene are associated with reduced risk for extrapulmonary and severe tuberculosis in Chinese Han children. Infection, Genetics and Evolution, 2014, 23, 121-128.	2.3	14
7	Instability of BLOC2 and BLOC3 in Chinese patients with Hermansky-Pudlak syndrome. Pigment Cell and Melanoma Research, 2019, 32, 373-380.	3.3	12
8	Genetic variants and mutational spectrum of Chinese Hermansky-Pudlak syndrome patients. Pigment Cell and Melanoma Research, 2021, 34, 111-121.	3.3	9
9	Clinical feature and waveform in infantile nystagmus syndrome in children with FRMD7 gene mutations. Science China Life Sciences, 2017, 60, 707-713.	4.9	8
10	Parallel Tests of Whole Exome Sequencing and Copy Number Variant Sequencing Increase the Diagnosis Yields of Rare Pediatric Disorders. Frontiers in Genetics, 2020, 11, 473.	2.3	8
11	The first Hermansky-Pudlak syndrome type 9 patient with two novel variants in Chinese population. Journal of Dermatology, 2021, 48, 676-680.	1.2	7
12	Clinical Application of Whole Exome Sequencing for Monogenic Disorders in PICU of China. Frontiers in Genetics, 2021, 12, 677699.	2.3	6
13	Spectrum Analysis of Albinism Genes in a Large Cohort of Chinese Index Patients. Journal of Investigative Dermatology, 2022, 142, 1752-1755.e3.	0.7	6
14	Neonates are armed with deviated immune cell proportion and cytokine reduction but higher T cell proliferation potentiality. Acta Biochimica Et Biophysica Sinica, 2018, 50, 934-937.	2.0	5
15	Application of multiplex ligation-dependent probe amplification in the genetic testing of oculocutaneous albinism. Chinese Medical Journal, 2019, 132, 2011-2012.	2.3	3
16	Exome sequencing as the first-tier test for pediatric respiratory diseases: A single-center study. Human Mutation, 2021, 42, 891-900.	2.5	2
17	Simpson-Golabi-Behmel syndrome with 46,XY disorders of sex development: A case report. American Journal of Medical Genetics, Part A, 2019, 179, 285-289.	1.2	1