

# Desheng Liang

## List of Publications by Year in descending order

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129  
papers

2,257  
citations

304743

22  
h-index

289244

40  
g-index

134  
all docs

134  
docs citations

134  
times ranked

3484  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical utility of noninvasive prenatal screening for expanded chromosome disease syndromes. <i>Genetics in Medicine</i> , 2019, 21, 1998-2006.	2.4	158
2	Noninvasive prenatal testing of fetal whole chromosome aneuploidy by massively parallel sequencing. <i>Prenatal Diagnosis</i> , 2013, 33, 409-415.	2.3	134
3	Pregnancy-related complications and adverse pregnancy outcomes in multiple pregnancies resulting from assisted reproductive technology: a meta-analysis of cohort studies. <i>Fertility and Sterility</i> , 2015, 103, 1492-1508.e7.	1.0	109
4	Copy Number Variation Sequencing for Comprehensive Diagnosis of Chromosome Disease Syndromes. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 519-526.	2.8	106
5	Common genetic variants on 1p13.2 associate with risk of autism. <i>Molecular Psychiatry</i> , 2014, 19, 1212-1219.	7.9	85
6	Noninvasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). <i>Clinical Chemistry</i> , 2015, 61, 172-181.	3.2	85
7	MicroRNAs expression in ox-LDL treated HUVECs: MiR-365 modulates apoptosis and Bcl-2 expression. <i>Biochemical and Biophysical Research Communications</i> , 2011, 410, 127-133.	2.1	77
8	Assisted reproductive technology and risk of congenital malformations: a meta-analysis based on cohort studies. <i>Archives of Gynecology and Obstetrics</i> , 2015, 292, 777-798.	1.7	75
9	XRCC2 mutation causes meiotic arrest, azoospermia and infertility. <i>Journal of Medical Genetics</i> , 2018, 55, 628-636.	3.2	54
10	Seamless Genetic Conversion of <i>SMN2</i> to <i>SMN1</i> via CRISPR/Cpf1 and Single-Stranded Oligodeoxynucleotides in Spinal Muscular Atrophy Patient-Specific Induced Pluripotent Stem Cells. <i>Human Gene Therapy</i> , 2018, 29, 1252-1263.	2.7	50
11	MicroRNA let-7c inhibits Bcl-xl expression and regulates ox-LDL-induced endothelial apoptosis. <i>BMB Reports</i> , 2012, 45, 464-469.	2.4	48
12	DMD mutation spectrum analysis in 613 Chinese patients with dystrophinopathy. <i>Journal of Human Genetics</i> , 2015, 60, 435-442.	2.3	47
13	TALE nickase mediates high efficient targeted transgene integration at the human multi-copy ribosomal DNA locus. <i>Biochemical and Biophysical Research Communications</i> , 2014, 446, 261-266.	2.1	45
14	In situ genetic correction of F8 intron 22 inversion in hemophilia A patient-specific iPSCs. <i>Scientific Reports</i> , 2016, 6, 18865.	3.3	43
15	Mutation in PITX2 is associated with ring dermoid of the cornea. <i>Journal of Medical Genetics</i> , 2004, 41, e129-e129.	3.2	39
16	Molecular analysis of hearing loss associated with enlarged vestibular aqueduct in the mainland Chinese: a unique SLC26A4 mutation spectrum. <i>Journal of Human Genetics</i> , 2007, 52, 492-497.	2.3	39
17	A syndactyly type IV locus maps to 7q36. <i>Journal of Human Genetics</i> , 2007, 52, 561-564.	2.3	35
18	A study on a cohort of 301 Chinese patients with isolated methylmalonic acidemia. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 409-423.	3.6	32

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19	A novel de novo POGZ mutation in a patient with intellectual disability. <i>Journal of Human Genetics</i> , 2016, 61, 357-359.	2.3	31
20	Targeting of the human F8 at the multicopy rDNA locus in Hemophilia a patient-derived iPSCs using TALEN Nickases. <i>Biochemical and Biophysical Research Communications</i> , 2016, 472, 144-149.	2.1	28
21	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. <i>Human Mutation</i> , 2017, 38, 669-677.	2.5	28
22	Non-viral ex vivo transduction of human hepatocyte cells to express factor VIII using a human ribosomal DNA-targeting vector. <i>Journal of Thrombosis and Haemostasis</i> , 2007, 5, 347-351.	3.8	26
23	Long-read DNA sequencing fully characterized chromothripsis in a patient with Langerâ€Giedion syndrome and Cornelia de Lange syndrome-4. <i>Journal of Human Genetics</i> , 2020, 65, 667-674.	2.3	24
24	Molecular cytogenetic analysis of early spontaneous abortions conceived from varying assisted reproductive technology procedures. <i>Molecular Cytogenetics</i> , 2016, 9, 79.	0.9	23
25	ssODN-Mediated In-Frame Deletion with CRISPR/Cas9 Restores FVIII Function in Hemophilia A-Patient-Derived iPSCs and ECs. <i>Molecular Therapy - Nucleic Acids</i> , 2019, 17, 198-209.	5.1	23
26	Comprehensive Analysis of Congenital Adrenal Hyperplasia Using Long-Read Sequencing. <i>Clinical Chemistry</i> , 2022, 68, 927-939.	3.2	23
27	MiR-223 Regulates Human Embryonic Stem Cell Differentiation by Targeting the IGF-1R/Akt Signaling Pathway. <i>PLoS ONE</i> , 2013, 8, e78769.	2.5	22
28	Targeting of the Human Coagulation Factor IX Gene at rDNA Locus of Human Embryonic Stem Cells. <i>PLoS ONE</i> , 2012, 7, e37071.	2.5	21
29	Targeted next-generation sequencing identifies novel compound heterozygous mutations of DYNC2H1 in a fetus with short rib-polydactyly syndrome, type III. <i>Clinica Chimica Acta</i> , 2015, 447, 47-51.	1.1	20
30	Enhanced tumor growth inhibition by mesenchymal stem cells derived from iPSCs with targeted integration of interleukin24 into rDNA loci. <i>Oncotarget</i> , 2017, 8, 40791-40803.	1.8	20
31	De novo exonic deletion of <i>KDM6A</i> in a Chinese girl with Kabuki syndrome: A case report and brief literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1613-1621.	1.2	19
32	A clear bias in parental origin of de novo pathogenic CNVs related to intellectual disability, developmental delay and multiple congenital anomalies. <i>Scientific Reports</i> , 2017, 7, 44446.	3.3	19
33	Paired CRISPR/Cas9 Nickases Mediate Efficient Site-Specific Integration of F9 into rDNA Locus of Mouse ESCs. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3035.	4.1	19
34	De Novo Mutations of CCNK Cause a Syndromic Neurodevelopmental Disorder with Distinctive Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 448-455.	6.2	17
35	A novel fusion suicide gene yeast CDglyTK plays a role in radio-gene therapy of nasopharyngeal carcinoma. <i>Cancer Gene Therapy</i> , 2004, 11, 790-796.	4.6	16
36	Site-Specific Integration of <i>TRAIL</i> in iPSC-Derived Mesenchymal Stem Cells for Targeted Cancer Therapy. <i>Stem Cells Translational Medicine</i> , 2022, 11, 297-309.	3.3	16

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37	Novel variants in PAX6 gene caused congenital aniridia in two Chinese families. <i>Eye</i> , 2017, 31, 956-961.	2.1	14
38	Mesenchymal stem cells derived from iPSCs expressing interleukin-24 inhibit the growth of melanoma in the tumor-bearing mouse model. <i>Cancer Cell International</i> , 2020, 20, 33.	4.1	14
39	Confirmation and refinement of a genetic locus of congenital motor nystagmus in Xq26.3-q27.1 in a Chinese family. <i>Human Genetics</i> , 2005, 116, 128-131.	3.8	13
40	Novel compound heterozygous mutation of MLYCD in a Chinese patient with malonic aciduria. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 79-83.	1.1	13
41	Variable phenotypes and outcomes associated with the MMACHC c.609G>A homologous mutation: long term follow-up in a large cohort of cases. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 200.	2.7	13
42	Molecular diagnosis for 55 fetuses with skeletal dysplasias by whole-exome sequencing: A retrospective cohort study. <i>Clinical Genetics</i> , 2021, 100, 219-226.	2.0	13
43	Amyotrophic lateral sclerosis (ALS) linked mutation in Ubiquilin 2 affects stress granule assembly via TIA1. <i>CNS Neuroscience and Therapeutics</i> , 2022, 28, 105-115.	3.9	13
44	Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. <i>Journal of Human Genetics</i> , 2017, 62, 513-516.	2.3	12
45	Translocation breakpoint disrupting the host SNHG14 gene but not coding genes or snoRNAs in typical Prader-Willi syndrome. <i>Journal of Human Genetics</i> , 2019, 64, 647-652.	2.3	12
46	Noninvasive prenatal diagnosis for pregnancies at risk for $\beta^0$ -thalassaemia: a retrospective study. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2021, 128, 448-457.	2.3	12
47	A novel GPR143 splicing mutation in a Chinese family with X-linked congenital nystagmus. <i>Molecular Vision</i> , 2011, 17, 715-22.	1.1	12
48	Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. <i>Human Mutation</i> , 2022, 43, 568-581.	2.5	12
49	Parental origin of apparently balanced <i>de novo</i> complex chromosomal rearrangements investigated by microdissection, whole genome amplification, and microsatellite-mediated haplotype analysis. <i>Clinical Genetics</i> , 2010, 78, 548-553.	2.0	11
50	Novel SIL1 nonstop mutation in a Chinese consanguineous family with Marinesco-Sjögren syndrome and Dandy-Walker syndrome. <i>Clinica Chimica Acta</i> , 2016, 458, 1-4.	1.1	11
51	Clinical and molecular investigation in Chinese patients with glutaric aciduria type I. <i>Clinica Chimica Acta</i> , 2016, 453, 75-79.	1.1	11
52	Truncating mutations of HIBCH tend to cause severe phenotypes in cases with HIBCH deficiency: a case report and brief literature review. <i>Journal of Human Genetics</i> , 2018, 63, 851-855.	2.3	11
53	Noninvasive fetal genotyping in pregnancies at risk for PKU using a comprehensive quantitative cSMART assay for PAH gene mutations: a clinical feasibility study. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2019, 126, 1466-1474.	2.3	11
54	Analysis of 70 patients with hydrocephalus due to cobalamin C deficiency. <i>Neurology</i> , 2020, 95, e3129-e3137.	1.1	11

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55	Silica nanoparticle is a possible safe carrier for gene therapy. <i>Science Bulletin</i> , 2005, 50, 2323-2327.	1.7	10
56	A non-viral vector for potential DMD gene therapy study by targeting a minidystrophin-GFP fusion gene into the hrDNA locus. <i>Acta Biochimica Et Biophysica Sinica</i> , 2009, 41, 1053-1060.	2.0	10
57	Phenotypic expansion of the interstitial 16p13.3 duplication: A case report and review of the literature. <i>Gene</i> , 2013, 531, 502-505.	2.2	10
58	Nonviral Gene Targeting at rDNA Locus of Human Mesenchymal Stem Cells. <i>BioMed Research International</i> , 2013, 2013, 1-10.	1.9	10
59	A New Next-Generation Sequencing-Based Assay for Concurrent Preimplantation Genetic Diagnosis of Charcot-Marie-Tooth Disease Type 1A and Aneuploidy Screening. <i>Journal of Genetics and Genomics</i> , 2016, 43, 155-159.	3.9	10
60	WDR73 missense mutation causes infantile onset intellectual disability and cerebellar hypoplasia in a consanguineous family. <i>Clinica Chimica Acta</i> , 2017, 464, 24-29.	1.1	10
61	CRISPR/Cas12a-Based Ultrasensitive and Rapid Detection of JAK2 V617F Somatic Mutation in Myeloproliferative Neoplasms. <i>Biosensors</i> , 2021, 11, 247.	4.7	10
62	CRISPR/Cas9-Mediated in vivo Genetic Correction in a Mouse Model of Hemophilia A. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 672564.	3.7	10
63	A novel single-base deletion mutation of the RUNX2 gene in a Chinese family with cleidocranial dysplasia. <i>Genetics and Molecular Research</i> , 2011, 10, 3539-3544.	0.2	10
64	Pachygyria, seizures, hypotonia, and growth retardation in a patient with an atypical 1.33Mb inherited microduplication at 22q11.23. <i>Gene</i> , 2015, 569, 46-50.	2.2	9
65	Diagnosis of Joubert Syndrome 10 in a Fetus with Suspected Dandy-Walker Variant by WES: A Novel Splicing Mutation in <i>OFD1</i> . <i>BioMed Research International</i> , 2018, 2018, 1-7.	1.9	9
66	Three Novel Mutations in <i>FBN1</i> and <i>TGFBR2</i> in Patients with the Syndromic Form of Thoracic Aortic Aneurysms and Dissections. <i>International Heart Journal</i> , 2018, 59, 1059-1068.	1.0	9
67	Cas12a and Lateral Flow Strip-Based Test for Rapid and Ultrasensitive Detection of Spinal Muscular Atrophy. <i>Biosensors</i> , 2021, 11, 154.	4.7	9
68	Analysis of the relationship between phenotypes and genotypes in 60 Chinese patients with propionic acidemia: a fourteen-year experience at a tertiary hospital. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 135.	2.7	9
69	The Combination of Suicide Gene Therapy and Radiation Enhances the Killing of Nasopharyngeal Carcinoma Xenographs. <i>Journal of Radiation Research</i> , 2004, 45, 281-289.	1.6	8
70	A father and son with mental retardation, a characteristic face, inv(12), and insertion trisomy 12p12.3-p11.2. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 238-244.	1.2	8
71	Three novel mutations of STK11 gene in Chinese patients with Peutz-Jeghers syndrome. <i>BMC Medical Genetics</i> , 2016, 17, 77.	2.1	8
72	Increase in diagnostic yield achieved for 174 whole-exome sequencing cases reanalyzed 1-2 years after initial analysis. <i>Clinica Chimica Acta</i> , 2021, 523, 163-168.	1.1	8

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73	Chromosomal analysis of blastocysts from balanced chromosomal rearrangement carriers. <i>Reproduction</i> , 2016, 151, 455-464.	2.6	7
74	Clinical outcomes for couples containing a reciprocal chromosome translocation carrier without preimplantation genetic diagnosis. <i>International Journal of Gynecology and Obstetrics</i> , 2017, 136, 304-308.	2.3	7
75	Deficiency in GnRH receptor trafficking due to a novel homozygous mutation causes idiopathic hypogonadotropic hypogonadism in three prepubertal siblings. <i>Gene</i> , 2018, 669, 42-46.	2.2	7
76	Development and validation of a haplotype-free technique for non-invasive prenatal diagnosis of spinal muscular atrophy. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23046.	2.1	7
77	Gene Therapy for Hemophilia A: Where We Stand. <i>Current Gene Therapy</i> , 2020, 20, 142-151.	2.0	7
78	Functional properties of peripheral CD <sup>8+</sup> T cells in patients with repeated implantation failure. <i>American Journal of Reproductive Immunology</i> , 2017, 78, e12704.	1.2	6
79	Identification of a novel MIP frameshift mutation associated with congenital cataract in a Chinese family by whole-exome sequencing and functional analysis. <i>Eye</i> , 2018, 32, 1359-1364.	2.1	6
80	Restoration of SMN expression in mesenchymal stem cells derived from gene-targeted patient-specific iPSCs. <i>Journal of Molecular Histology</i> , 2018, 49, 27-37.	2.2	6
81	Six novel Mutation analysis of the androgen receptor gene in 17 Chinese patients with androgen insensitivity syndrome. <i>Clinica Chimica Acta</i> , 2020, 506, 180-186.	1.1	6
82	Loss of PIGK function causes severe infantile encephalopathy and extensive neuronal apoptosis. <i>Human Genetics</i> , 2021, 140, 791-803.	3.8	6
83	Noninvasive Prenatal Testing of Methylmalonic Acidemia cblC Type Using the cSMART Assay for MMACHC Gene Mutations. <i>Frontiers in Genetics</i> , 2021, 12, 750719.	2.3	6
84	A novel deletion mutation in RS1 gene caused X-linked juvenile retinoschisis in a Chinese family. <i>Eye</i> , 2014, 28, 1364-1369.	2.1	5
85	Rare intracranial cholesterol deposition and a homozygous mutation of LDLR in a familial hypercholesterolemia patient. <i>Gene</i> , 2015, 569, 313-317.	2.2	5
86	Two novel NIPBL gene mutations in Chinese patients with Cornelia de Lange syndrome. <i>Gene</i> , 2015, 555, 476-480.	2.2	5
87	Notable Carrier Risks for Individuals Having Two Copies of <i>SMN1</i> in Spinal Muscular Atrophy Families with 2-copy Alleles: Estimation Based on Chinese Meta-analysis Data. <i>Journal of Genetic Counseling</i> , 2017, 26, 72-78.	1.6	5
88	Gene Therapy for Hemophilia and Duchenne Muscular Dystrophy in China. <i>Human Gene Therapy</i> , 2018, 29, 146-150.	2.7	5
89	Detection of Spinal Muscular Atrophy Using a Duplexed Real-Time PCR Approach With Locked Nucleic Acid-Modified Primers. <i>Annals of Laboratory Medicine</i> , 2021, 41, 101-107.	2.5	5
90	Targeted addition of mini-dystrophin into rDNA locus of Duchenne muscular dystrophy patient-derived iPSCs. <i>Biochemical and Biophysical Research Communications</i> , 2021, 545, 40-45.	2.1	5

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91	Expression of reconstructive hFVIII in the hrDNA by using hrDNA targeting vector. Science Bulletin, 2005, 50, 2187-2192.	1.7	4
92	A family with partial duplication/deletion 4p due to a balanced t (4; 15) (p16.2; p11.2) translocation. American Journal of Medical Genetics, Part A, 2011, 155, 656-659.	1.2	4
93	Mutation Analysis in Chinese Patients with Cornelia de Lange Syndrome. Genetic Testing and Molecular Biomarkers, 2012, 16, 1130-1134.	0.7	4
94	An Xp21.3p11.4 duplication observed in a boy with intellectual deficiency and speech delay and his asymptomatic mother. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 467-470.	1.6	4
95	Brief report adult patient presenting an interstitial (9) (q21.32q31.1) direct duplication resulting from the malsegregation of a paternal balanced insertional translocation. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 294-299.	1.6	4
96	De Novo ring chromosome 11 and non-reciprocal translocation of 11p15.3-pter to 21qter in a patient with congenital heart disease. Molecular Cytogenetics, 2015, 8, 88.	0.9	4
97	Novel de novo nonsense mutation of the PHEX gene (p.Lys50Ter) in a Chinese patient with hypophosphatemic rickets. Gene, 2015, 565, 150-154.	2.2	4
98	Partial trisomy 2q33.3-q37.3 in a patient with an inverted duplicated neocentric marker chromosome. Molecular Cytogenetics, 2015, 8, 10.	0.9	4
99	A novel MSX1 intronic mutation associated with autosomal dominant non-syndromic oligodontia in a large Chinese family pedigree. Clinica Chimica Acta, 2016, 461, 135-140.	1.1	4
100	Targeted exome sequencing identifies novel compound heterozygous mutations in P3H1 in a fetus with osteogenesis imperfecta type VIII. Clinica Chimica Acta, 2017, 464, 170-175.	1.1	4
101	28 novel mutations identified from 33 Chinese patients with cilia-related kidney disorders. Clinica Chimica Acta, 2020, 501, 207-215.	1.1	4
102	The rare Alu element-mediated chimerism of multiple de novo complex rearrangement sequences in GAN result in giant axonal neuropathy. Clinica Chimica Acta, 2020, 502, 91-98.	1.1	4
103	A novel <i>GJB1</i> mutation associated with X-linked Charcot-Marie-Tooth disease in a large Chinese family pedigree. Molecular Genetics & Genomic Medicine, 2020, 8, e1127.	1.2	4
104	IL-24 armored CAR19-T cells show enhanced antitumor activity and persistence. Signal Transduction and Targeted Therapy, 2021, 6, 14.	17.1	4
105	An Episomal CRISPR/Cas12a System for Mediating Efficient Gene Editing. Life, 2021, 11, 1262.	2.4	4
106	Ectopic Expression of FVIII in HPCs and MSCs Derived from hiPSCs with Site-Specific Integration of ITGA2B Promoter-Driven BDDF8 Gene in Hemophilia A. International Journal of Molecular Sciences, 2022, 23, 623.	4.1	4
107	Targeted B-domain deletion restores F8 function in human endothelial cells and mice. Signal Transduction and Targeted Therapy, 2022, 7, .	17.1	4
108	Mutation analysis of the SRY, NR5A1, and DHH genes in six Chinese 46,XY women. Journal of Maternal-Fetal and Neonatal Medicine, 2011, 24, 863-866.	1.5	3



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109	A lesson from a reported pathogenic variant in Peutz-Jeghers syndrome: a case report. <i>Familial Cancer</i> , 2017, 16, 417-422.	1.9	3
110	First report on an X-linked hypohidrotic ectodermal dysplasia family with X chromosome inversion: Breakpoint mapping reveals the pathogenic mechanism and preimplantation genetics diagnosis achieves an unaffected birth. <i>Clinica Chimica Acta</i> , 2017, 475, 78-84.	1.1	3
111	REDBot: Natural language process methods for clinical copy number variation reporting in prenatal and products of conception diagnosis. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1488.	1.2	3
112	Behavioral and Gene Expression Analysis of Stxbp6-Knockout Mice. <i>Brain Sciences</i> , 2021, 11, 436.	2.3	3
113	Novel variants in OSGEP leading to Galloway-Mowat syndrome by altering its subcellular localization. <i>Clinica Chimica Acta</i> , 2021, 523, 297-303.	1.1	3
114	Identification of four novel mutations in BTK from six Chinese families with X-linked agammaglobulinemia. <i>Clinica Chimica Acta</i> , 2022, 531, 48-55.	1.1	3
115	Cas14a1-Mediated Nucleic Acid Diagnostics for Spinal Muscular Atrophy. <i>Biosensors</i> , 2022, 12, 268.	4.7	3
116	Generation of reporter hESCs by targeting <i>EGFP</i> at the CD144 locus to facilitate the endothelial differentiation. <i>Development Growth and Differentiation</i> , 2018, 60, 205-215.	1.5	2
117	Ectopic expression of factor VIII in MSCs and hepatocytes derived from rDNA targeted hESCs. <i>Clinica Chimica Acta</i> , 2019, 495, 656-663.	1.1	2
118	Identification of pathogenic mutations in 6 Chinese families with multiple exostoses by whole-exome sequencing and multiplex ligation-dependent probe amplification. <i>Medicine (United States)</i> , 2019, 98, e15692.	1.0	2
119	Prenatal Diagnosis in a Fetus With X-Linked Recessive Chondrodysplasia Punctata: Identification and Functional Study of a Novel Missense Mutation in ARSE. <i>Frontiers in Genetics</i> , 2021, 12, 722694.	2.3	2
120	Functional analysis of three genetic disorder related PITX2 mutants. <i>Science Bulletin</i> , 2006, 51, 164-169.	1.7	1
121	Investigation of hrDNA targeting vector-mediated tumor-specific suicide gene therapy for hepatocellular carcinoma. <i>Science Bulletin</i> , 2006, 51, 2342-2350.	1.7	1
122	Three patients with Wolf-Hirschhorn syndrome carrying a satellited chromosome 4p. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 549-552.	1.6	1
123	A novel FOXL2 mutation in a Chinese family with blepharophimosis, ptosis, epicanthus inversus syndrome. <i>Human Genome Variation</i> , 2015, 2, 15008.	0.7	1
124	Simultaneous Identification of Both MFSD8 and RDH12 Pathogenic Variants in a Chinese Family Affected With Retinitis Pigmentosa. <i>Frontiers in Genetics</i> , 2021, 12, 715100.	2.3	1
125	A patient with apparently reciprocal translocation and cryptic 10p deletion. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1753-1755.	1.2	0
126	Identification of a novel SHOX mutation in a Chinese family with isolated Madelung deformity. <i>Journal of Genetics</i> , 2014, 93, 809-812.	0.7	0



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127	Morphometric analysis and developmental comparison of embryos from carriers with balanced chromosomal rearrangements in preimplantation genetic diagnosis cycles. <i>Reproduction, Fertility and Development</i> , 2016, 28, 1953.	0.4	0
128	Novel GZF1 pathogenic variants identified in two Chinese patients with Larsen syndrome. <i>Clinical Genetics</i> , 2021, 99, 281-285.	2.0	0
129	Biochemical and genetic characteristics of 40 neonates with carnitine deficiency. <i>Journal of Central South University (Medical Sciences)</i> , 2020, 45, 1164-1171.	0.1	0