

Zornitza Stark Bm Bch, Dm

List of Publications by Year in descending order

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Version: 2024-02-01

108
papers

4,753
citations

145106

33
h-index

134545

62
g-index

116
all docs

116
docs citations

116
times ranked

7981
citing authors

#	ARTICLE	IF	CITATIONS
1	Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. <i>Npj Genomic Medicine</i> , 2018, 3, 16.	1.7	420
2	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. <i>Genetics in Medicine</i> , 2016, 18, 1090-1096.	1.1	332
3	Integrating Genomics into Healthcare: A Global Responsibility. <i>American Journal of Human Genetics</i> , 2019, 104, 13-20.	2.6	264
4	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. <i>JAMA Pediatrics</i> , 2017, 171, 855.	3.3	252
5	Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. <i>Genetics in Medicine</i> , 2017, 19, 867-874.	1.1	194
6	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. <i>JAMA - Journal of the American Medical Association</i> , 2020, 323, 2503.	3.8	160
7	The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. <i>Genetics in Medicine</i> , 2016, 18, 483-493.	1.1	127
8	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. <i>Genetics in Medicine</i> , 2018, 20, 1554-1563.	1.1	125
9	Defects in tRNA Anticodon Loop 2- <i>O</i> -Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations in <i>FTSJ1</i> . <i>Human Mutation</i> , 2015, 36, 1176-1187.	1.1	122
10	Does genomic sequencing early in the diagnostic trajectory make a difference? A follow-up study of clinical outcomes and cost-effectiveness. <i>Genetics in Medicine</i> , 2019, 21, 173-180.	1.1	118
11	Mutations in <i>CSPP1</i> Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 62-72.	2.6	104
12	Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. <i>European Journal of Human Genetics</i> , 2018, 26, 644-651.	1.4	102
13	<i>SYT1</i> -associated neurodevelopmental disorder: a case series. <i>Brain</i> , 2018, 141, 2576-2591.	3.7	98
14	De Novo Loss-of-Function Mutations in <i>USP9X</i> Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , 2016, 98, 373-381.	2.6	95
15	GA4GH: International policies and standards for data sharing across genomic research and healthcare. <i>Cell Genomics</i> , 2021, 1, 100029.	3.0	94
16	<i>ACTB</i> Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 1021-1033.	2.6	83
17	Cpipe: a shared variant detection pipeline designed for diagnostic settings. <i>Genome Medicine</i> , 2015, 7, 68.	3.6	78
18	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. <i>American Journal of Human Genetics</i> , 2019, 105, 7-14.	2.6	75

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19	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. <i>Genetics in Medicine</i> , 2021, 23, 183-191.	1.1	70
20	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. <i>Genetics in Medicine</i> , 2019, 21, 850-860.	1.1	68
21	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	1.1	56
22	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 224-228.	1.4	48
23	Rapid genomic testing for critically ill children: time to become standard of care?. <i>European Journal of Human Genetics</i> , 2022, 30, 142-149.	1.4	45
24	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	1.1	45
25	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1508.	0.6	44
26	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffinâ€“Siris-like syndrome. <i>Journal of Human Genetics</i> , 2017, 62, 741-746.	1.1	43
27	Long-term economic impacts of exome sequencing for suspected monogenic disorders: diagnosis, management, and reproductive outcomes. <i>Genetics in Medicine</i> , 2019, 21, 2586-2593.	1.1	43
28	A novel presentation of homozygous loss-of-function STAT-1 mutation in an infant with hyperinflammationâ€“A case report and review of the literature. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2016, 4, 777-779.	2.0	42
29	Genome-wide sequencing in acutely ill infants: genomic medicineâ€™s critical application?. <i>Genetics in Medicine</i> , 2019, 21, 498-504.	1.1	42
30	Extending the scope of diagnostic chromosome analysis: Detection of single gene defects using high-resolution SNP microarrays. <i>Human Mutation</i> , 2011, 32, 1500-1506.	1.1	41
31	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	0.7	40
32	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 763-770.	2.6	37
33	A head-to-head evaluation of the diagnostic efficacy and costs of trio versus singleton exome sequencing analysis. <i>European Journal of Human Genetics</i> , 2019, 27, 1791-1799.	1.4	37
34	Discussing withholding and withdrawing of lifeâ€“sustaining medical treatment in paediatric inpatients: Audit of current practice. <i>Journal of Paediatrics and Child Health</i> , 2008, 44, 399-403.	0.4	36
35	Genotype and phenotype spectrum of NRAS germline variants. <i>European Journal of Human Genetics</i> , 2017, 25, 823-831.	1.4	36
36	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 318-325.	1.7	36

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37	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. <i>American Journal of Human Genetics</i> , 2021, 108, 1551-1557.	2.6	36
38	Rapid Challenges: Ethics and Genomic Neonatal Intensive Care. <i>Pediatrics</i> , 2019, 143, S14-S21.	1.0	35
39	Genetic counseling in pediatric acute care: Reflections on ultra-rapid genomic diagnoses in neonates. <i>Journal of Genetic Counseling</i> , 2019, 28, 273-282.	0.9	34
40	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	0.7	34
41	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 502-508.	0.7	31
42	The personal utility and uptake of genomic sequencing in pediatric and adult conditions: eliciting societal preferences with three discrete choice experiments. <i>Genetics in Medicine</i> , 2020, 22, 1311-1319.	1.1	31
43	Insights into the genotype-phenotype correlation and molecular function of SLC25A46. <i>Human Mutation</i> , 2018, 39, 1995-2007.	1.1	30
44	Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. <i>European Journal of Human Genetics</i> , 2019, 27, 1493-1501.	1.4	29
45	Triad of tracheoesophageal fistula-esophageal atresia, pulmonary hypoplasia, and duodenal atresia. <i>Journal of Pediatric Surgery</i> , 2007, 42, 1146-1148.	0.8	28
46	De novo intrachromosomal gene conversion from OPN1MW to OPN1LW in the male germline results in Blue Cone Monochromacy. <i>Scientific Reports</i> , 2016, 6, 28253.	1.6	28
47	Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. <i>Genetics in Medicine</i> , 2020, 22, 1976-1985.	1.1	28
48	Parental health spillover effects of paediatric rare genetic conditions. <i>Quality of Life Research</i> , 2020, 29, 2445-2454.	1.5	28
49	Attitudes and Practices of Australian Nephrologists Toward Implementation of Clinical Genomics. <i>Kidney International Reports</i> , 2021, 6, 272-283.	0.4	28
50	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. <i>Nature Communications</i> , 2021, 12, 2678.	5.8	26
51	KBC syndrome: An Australian experience. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1866-1877.	0.7	25
52	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. <i>Genetics in Medicine</i> , 2020, 22, 1986-1993.	1.1	25
53	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. <i>European Journal of Human Genetics</i> , 2017, 25, 1268-1272.	1.4	24
54	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 2-9.	1.0	24

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55	5q31.3 Microdeletion syndrome: Clinical and molecular characterization of two further cases. American Journal of Medical Genetics, Part A, 2013, 161, 2604-2608.	0.7	23
56	SNP microarray abnormalities in a cohort of 28 infants with congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 2319-2326.	0.7	19
57	Offering pregnant women different levels of genetic information from prenatal chromosome microarray: a prospective study. European Journal of Human Genetics, 2018, 26, 485-494.	1.4	19
58	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. European Journal of Human Genetics, 2019, 27, 1821-1826.	1.4	19
59	Clinical genomic testing: what matters to key stakeholders?. European Journal of Human Genetics, 2020, 28, 866-873.	1.4	19
60	Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. Npj Genomic Medicine, 2021, 6, 5.	1.7	19
61	Is faster better? An economic evaluation of rapid and ultra-rapid genomic testing in critically ill infants and children. Genetics in Medicine, 2022, 24, 1037-1044.	1.1	18
62	Apert syndrome: temporal lobe abnormalities on fetal brain imaging. Prenatal Diagnosis, 2015, 35, 179-182.	1.1	17
63	Metronidazole Toxicity in Cockayne Syndrome: A Case Series. Pediatrics, 2015, 136, e706-e708.	1.0	17
64	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€² methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	1.8	17
65	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
66	Multiomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. Human Mutation, 2021, 42, 19-24.	1.1	17
67	A homozygous <i>UBA5</i> pathogenic variant causes a fatal congenital neuropathy. Journal of Medical Genetics, 2020, 57, 835-842.	1.5	16
68	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.3	16
69	Biallelic loss of function variants in <i>PPP1R21</i> cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	1.1	15
70	Cost-Effectiveness of Targeted Exome Analysis as a Diagnostic Test in Glomerular Diseases. Kidney International Reports, 2021, 6, 2850-2861.	0.4	15
71	The expanding <i>LARS2</i> phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. Human Mutation, 2020, 41, 1425-1434.	1.1	15
72	Copy number variants including RAS pathway genesâ€”How much RASopathy is in the phenotype?. American Journal of Medical Genetics, Part A, 2015, 167, 2685-2690.	0.7	14

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73	Parentsâ€™ experiences of decision making for rapid genomic sequencing in intensive care. <i>European Journal of Human Genetics</i> , 2021, 29, 1804-1810.	1.4	14
74	The value of genomic sequencing in complex pediatric neurological disorders: a discrete choice experiment. <i>Genetics in Medicine</i> , 2021, 23, 155-162.	1.1	13
75	Polymicrogyria in association with hypoglycemia points to mutation in the mTOR pathway. <i>European Journal of Medical Genetics</i> , 2018, 61, 738-740.	0.7	12
76	Preferences and values for rapid genomic testing in critically ill infants and children: a discrete choice experiment. <i>European Journal of Human Genetics</i> , 2021, 29, 1645-1653.	1.4	12
77	A Mouse Splice-Site Mutant and Individuals with Atypical Chromosome 22q11.2 Deletions Demonstrate the Crucial Role for Crkl in Craniofacial and Pharyngeal Development. <i>Molecular Syndromology</i> , 2014, 5, 276-286.	0.3	11
78	A novel <i>AMPD2</i> mutation outside the AMP deaminase domain causes pontocerebellar hypoplasia type 9. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 820-823.	0.7	11
79	Diagnostic and service impact of genomic testing technologies in a neonatal intensive care unit. <i>Journal of Paediatrics and Child Health</i> , 2019, 55, 1309-1314.	0.4	11
80	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. <i>Allergy, Asthma and Clinical Immunology</i> , 2018, 14, 65.	0.9	10
81	â€œDiagnostic shockâ€™: the impact of results from ultrarapid genomic sequencing of critically unwell children on aspects of family functioning. <i>European Journal of Human Genetics</i> , 0, , .	1.4	10
82	Fetal phenotype of 17q12 microdeletion syndrome: renal echogenicity and congenital diaphragmatic hernia in 2 cases. <i>Prenatal Diagnosis</i> , 2015, 35, 1265-1267.	1.1	9
83	Prenatal diagnosis of fragile X syndrome complicated by full mutation retraction. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2485-2487.	0.7	9
84	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 531-535.	0.7	9
85	The leadership behaviors needed to implement clinical genomics at scale: a qualitative study. <i>Genetics in Medicine</i> , 2020, 22, 1384-1390.	1.1	9
86	Rapid Identification of Biallelic <i>SPTB</i> Mutation in a Neonate with Severe Congenital Hemolytic Anemia and Liver Failure. <i>Molecular Syndromology</i> , 2020, 11, 50-55.	0.3	9
87	Teamwork in clinical genomics: A dynamic sociotechnical healthcare setting. <i>Journal of Evaluation in Clinical Practice</i> , 2021, 27, 1369-1380.	0.9	9
88	Lessons learnt from multifaceted diagnostic approaches to the first 150 families in Victoriaâ€™s Undiagnosed Diseases Program. <i>Journal of Medical Genetics</i> , 2022, 59, 748-758.	1.5	9
89	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous <i>ASNS</i> splicing variant in a critically ill neonate. <i>Human Mutation</i> , 2020, 41, 1884-1891.	1.1	8
90	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. <i>Obstetrical and Gynecological Survey</i> , 2020, 75, 662-664.	0.2	7

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91	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. <i>JAMA Neurology</i> , 2022, 79, 405.	4.5	7
92	Rare cause of maternal and neonatal hypercalcaemia. <i>Journal of Paediatrics and Child Health</i> , 2019, 55, 232-235.	0.4	6
93	Treatment limitation and advance planning: Hospital-wide audit of paediatric death. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 893-899.	0.4	6
94	Goldberg's Shprintzen syndrome is determined by the absence, or reduced expression levels, of KIFBP. <i>Human Mutation</i> , 2020, 41, 1906-1917.	1.1	6
95	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. <i>International Journal of Molecular Sciences</i> , 2022, 23, 986.	1.8	5
96	Consent for rapid genomic sequencing for critically ill children: legal and ethical issues. <i>Monash Bioethics Review</i> , 2021, 39, 117-129.	0.4	5
97	Predictive genetic testing for neurodegenerative conditions: how should conflicting interests within families be managed?. <i>Journal of Medical Ethics</i> , 2016, 42, 640-642.	1.0	4
98	Novel missense mutations in a conserved loop between ERCC6 (CSB) helicase motifs V and VI: Insights into Cockayne syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 773-776.	0.7	4
99	Clinical and Molecular Differences between 4-Year-Old Monozygous Male Twins Mosaic for Normal, Premutation and Fragile X Full Mutation Alleles. <i>Genes</i> , 2019, 10, 279.	1.0	4
100	Clinical versus research genomics in kidney disease. <i>Nature Reviews Nephrology</i> , 2021, 17, 570-571.	4.1	4
101	Neonatal Bartter syndrome diagnosed by rapid genomics following low risk pre-conception carrier screening. <i>Journal of Paediatrics and Child Health</i> , 2022, 58, 758-761.	0.4	4
102	Ethylmalonic encephalopathy masquerading as meningococccemia. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006193.	0.5	3
103	Can Rapid Nanopore Sequencing Bring Genomic Testing to the Bedside?. <i>Clinical Chemistry</i> , 2022, 68, 1484-1485.	1.5	3
104	Use of ultra-rapid whole-exome sequencing to diagnose congenital central hypoventilation syndrome. <i>Pediatric Pulmonology</i> , 2020, 55, 855-857.	1.0	2
105	Distinct diagnostic trajectories in <i>NBAS</i> -associated acute liver failure highlights the need for timely functional studies. <i>JIMD Reports</i> , 2022, 63, 240-249.	0.7	2
106	Methyl-CpG binding domain 4, DNA glycosylase (<i>MBD4</i>)-associated neoplasia syndrome associated with a homozygous missense variant in <i>MBD4</i> : Expansion of an emerging phenotype. <i>British Journal of Haematology</i> , 2022, , .	1.2	2
107	The HIDDEN Protocol: An Australian Prospective Cohort Study to Determine the Utility of Whole Genome Sequencing in Kidney Failure of Unknown Aetiology. <i>Frontiers in Medicine</i> , 0, 9, .	1.2	2
108	Response to Ferket et al.. <i>Genetics in Medicine</i> , 2020, 22, 1910.	1.1	0