Zornitza Stark Bm Bch, Dm

List of Publications by Year
in descending order

108 papers	4,753 citations	126907 33 h-index	118850 62 g-index
116 all docs	116 docs citations	116 times ranked	7543 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. Npj Genomic Medicine, 2018, 3, 16.	3.8	420
2	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	2.4	332
3	Integrating Genomics into Healthcare: A Global Responsibility. American Journal of Human Genetics, 2019, 104, 13-20.	6.2	264
4	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	6.2	252
5	Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. Genetics in Medicine, 2017, 19, 867-874.	2.4	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. JAMA - Journal of the American Medical Association, 2020, 323, 2503.	7.4	160
7	The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. Genetics in Medicine, 2016, 18, 483-493.	2.4	127
8	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. Genetics in Medicine, 2018, 20, 1554-1563.	2.4	125
9	Defects in tRNA Anticodon Loop 2â€²-O-Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations inFTSJ1. Human Mutation, 2015, 36, 1176-1187.	2.5	122
10	Does genomic sequencing early in the diagnostic trajectory make a difference? AÂfollow-up study of clinical outcomes and cost-effectiveness. Genetics in Medicine, 2019, 21, 173-180.	2.4	118
11	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	6.2	104
12	Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. European Journal of Human Genetics, 2018, 26, 644-651.	2.8	102
13	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	7.6	98
14	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95
15	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
16	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
17	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
18	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. American Journal of Human Genetics, 2019, 105, 7-14.	6.2	75

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19	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. Genetics in Medicine, 2021, 23, 183-191.	2.4	70
20	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
21	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
22	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	2.8	48
23	Rapid genomic testing for critically ill children: time to become standard of care?. European Journal of Human Genetics, 2022, 30, 142-149.	2.8	45
24	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
25	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1508.	1.2	44
26	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffinâ€“Siris-like syndrome. Journal of Human Genetics, 2017, 62, 741-746.	2.3	43
27	Long-term economic impacts of exome sequencing for suspected monogenic disorders: diagnosis, management, and reproductive outcomes. Genetics in Medicine, 2019, 21, 2586-2593.	2.4	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. Journal of Allergy and Clinical Immunology: in Practice, 2016, 4, 777-779.	3.8	42
29	Genome-wide sequencing in acutely ill infants: genomic medicineâ€™s critical application?. Genetics in Medicine, 2019, 21, 498-504.	2.4	42
30	Extending the scope of diagnostic chromosome analysis: Detection of single gene defects using high-resolution SNP microarrays. Human Mutation, 2011, 32, 1500-1506.	2.5	41
31	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
32	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
33	A head-to-head evaluation of the diagnostic efficacy and costs of trio versus singleton exome sequencing analysis. European Journal of Human Genetics, 2019, 27, 1791-1799.	2.8	37
34	Discussing withholding and withdrawing of lifeâ€sustaining medical treatment in paediatric inpatients: Audit of current practice. Journal of Paediatrics and Child Health, 2008, 44, 399-403.	0.8	36
35	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
36	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36

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37	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
38	Rapid Challenges: Ethics and Genomic Neonatal Intensive Care. Pediatrics, 2019, 143, S14-S21.	2.1	35
39	Genetic counseling in pediatric acute care: Reflections on ultraâ€rapid genomic diagnoses in neonates. Journal of Genetic Counseling, 2019, 28, 273-282.	1.6	34
40	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
41	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 502-508.	1.6	31
42	The personal utility and uptake of genomic sequencing in pediatric and adult conditions: eliciting societal preferences with three discrete choice experiments. Genetics in Medicine, 2020, 22, 1311-1319.	2.4	31
43	Insights into the genotype-phenotype correlation and molecular function of SLC25A46. Human Mutation, 2018, 39, 1995-2007.	2.5	30
44	Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. European Journal of Human Genetics, 2019, 27, 1493-1501.	2.8	29
45	Triad of tracheoesophageal fistula-esophageal atresia, pulmonary hypoplasia, and duodenal atresia. Journal of Pediatric Surgery, 2007, 42, 1146-1148.	1.6	28
46	De novo intrachromosomal gene conversion from OPN1MW to OPN1LW in the male germline results in Blue Cone Monochromacy. Scientific Reports, 2016, 6, 28253.	3.3	28
47	Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. Genetics in Medicine, 2020, 22, 1976-1985.	2.4	28
48	Parental health spillover effects of paediatric rare genetic conditions. Quality of Life Research, 2020, 29, 2445-2454.	3.1	28
49	Attitudes and Practices of Australian Nephrologists Toward Implementation of Clinical Genomics. Kidney International Reports, 2021, 6, 272-283.	0.8	28
50	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. Nature Communications, 2021, 12, 2678.	12.8	26
51	KBG syndrome: An Australian experience. American Journal of Medical Genetics, Part A, 2017, 173, 1866-1877.	1.2	25
52	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. Genetics in Medicine, 2020, 22, 1986-1993.	2.4	25
53	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	2.8	24
54	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	2.0	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.	1.2	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.	1.2	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.	2.8	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.	2.8	19
59	Clinical genomic testing: what matters to key stakeholders?. European Journal of Human Genetics, 2020, 28, 866-873.	2.8	19
60	Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. Npj Genomic Medicine, 2021, 6, 5.	3.8	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.	2.4	18
62	Apert syndrome: temporal lobe abnormalities on fetal brain imaging. Prenatal Diagnosis, 2015, 35, 179-182.	2.3	17
63	Metronidazole Toxicity in Cockayne Syndrome: A Case Series. Pediatrics, 2015, 136, e706-e708.	2.1	17
64	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€˜Oâ€˜methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	3.8	17
65	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
66	Multiomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. Human Mutation, 2021, 42, 19-24.	2.5	17
67	A homozygous UBA5 pathogenic variant causes a fatal congenital neuropathy. Journal of Medical Genetics, 2020, 57, 835-842.	3.2	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.	0.6	16
69	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
70	Cost-Effectiveness of Targeted Exome Analysis as a Diagnostic Test in Glomerular Diseases. Kidney International Reports, 2021, 6, 2850-2861.	0.8	15
71	The expandingLARS2phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. Human Mutation, 2020, 41, 1425-1434.	2.5	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.	1.2	14

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73	Parentsâ€™ experiences of decision making for rapid genomic sequencing in intensive care. European Journal of Human Genetics, 2021, 29, 1804-1810.	2.8	14
74	The value of genomic sequencing in complex pediatric neurological disorders: a discrete choice experiment. Genetics in Medicine, 2021, 23, 155-162.	2.4	13
75	Polymicrogyria in association with hypoglycemia points to mutation in the mTOR pathway. European Journal of Medical Genetics, 2018, 61, 738-740.	1.3	12
76	Preferences and values for rapid genomic testing in critically ill infants and children: a discrete choice experiment. European Journal of Human Genetics, 2021, 29, 1645-1653.	2.8	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. Molecular Syndromology, 2014, 5, 276-286.	0.8	11
78	A novel AMPD2 mutation outside the AMP deaminase domain causes pontocerebellar hypoplasia type 9. American Journal of Medical Genetics, Part A, 2017, 173, 820-823.	1.2	11
79	Diagnostic and service impact of genomic testing technologies in a neonatal intensive care unit. Journal of Paediatrics and Child Health, 2019, 55, 1309-1314.	0.8	11
80	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. Allergy, Asthma and Clinical Immunology, 2018, 14, 65.	2.0	10
81	â€˜Diagnostic shockâ€™: the impact of results from ultrarapid genomic sequencing of critically unwell children on aspects ofÂfamily functioning. European Journal of Human Genetics, 0, , .	2.8	10
82	Fetal phenotype of 17q12 microdeletion syndrome: renal echogenicity and congenital diaphragmatic hernia in 2 cases. Prenatal Diagnosis, 2015, 35, 1265-1267.	2.3	9
83	Prenatal diagnosis of fragile X syndrome complicated by full mutation retraction. American Journal of Medical Genetics, Part A, 2015, 167, 2485-2487.	1.2	9
84	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535.	1.2	9
85	The leadership behaviors needed to implement clinical genomics at scale: a qualitative study. Genetics in Medicine, 2020, 22, 1384-1390.	2.4	9
86	Rapid Identification of Biallelic <i>SPTB</i> Mutation in a Neonate with Severe Congenital Hemolytic Anemia and Liver Failure. Molecular Syndromology, 2020, 11, 50-55.	0.8	9
87	Teamwork in clinical genomics: A dynamic sociotechnical healthcare setting. Journal of Evaluation in Clinical Practice, 2021, 27, 1369-1380.	1.8	9
88	Lessons learnt from multifaceted diagnostic approaches to the first 150 families in Victoriaâ€™s Undiagnosed Diseases Program. Journal of Medical Genetics, 2022, 59, 748-758.	3.2	9
89	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. Human Mutation, 2020, 41, 1884-1891.	2.5	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. Obstetrical and Gynecological Survey, 2020, 75, 662-664.	0.4	7

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91	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	9.0	7
92	Rare cause of maternal and neonatal hypercalcaemia. Journal of Paediatrics and Child Health, 2019, 55, 232-235.	0.8	6
93	Treatment limitation and advance planning: Hospitalâ€wide audit of paediatric death. Journal of Paediatrics and Child Health, 2020, 56, 893-899.	0.8	6
94	Goldbergâ€“Shprintzen syndrome is determined by the absence, or reduced expression levels, of KIFBP. Human Mutation, 2020, 41, 1906-1917.	2.5	6
95	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. International Journal of Molecular Sciences, 2022, 23, 986.	4.1	5
96	Consent for rapid genomic sequencing for critically ill children: legal and ethical issues. Monash Bioethics Review, 2021, 39, 117-129.	0.8	5
97	Predictive genetic testing for neurodegenerative conditions: how should conflicting interests within families be managed?. Journal of Medical Ethics, 2016, 42, 640-642.	1.8	4
98	Novel missense mutations in a conserved loop between ERCC6 (CSB) helicase motifs V and VI: Insights into Cockayne syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 773-776.	1.2	4
99	Clinical and Molecular Differences between 4-Year-Old Monozygous Male Twins Mosaic for Normal, Premutation and Fragile X Full Mutation Alleles. Genes, 2019, 10, 279.	2.4	4
100	Clinical versus research genomics in kidney disease. Nature Reviews Nephrology, 2021, 17, 570-571.	9.6	4
101	Neonatal Bartter syndrome diagnosed by rapid genomics following low risk preâ€conception carrier screening. Journal of Paediatrics and Child Health, 2022, 58, 758-761.	0.8	4
102	Ethylmalonic encephalopathy masquerading as meningococcemia. Journal of Physical Education and Sports Management, 2022, , mcs.a006193.	1.2	3
103	Can Rapid Nanopore Sequencing Bring Genomic Testing to the Bedside?. Clinical Chemistry, 2022, 68, 1484-1485.	3.2	3
104	Use of ultraâ€rapid wholeâ€exome sequencing to diagnose congenital central hypoventilation syndrome. Pediatric Pulmonology, 2020, 55, 855-857.	2.0	2
105	Distinct diagnostic trajectories in <scp>NBAS</scp>â€associated acute liver failure highlights the need for timely functional studies. JIMD Reports, 2022, 63, 240-249.	1.5	2
106	Methylâ€CpG binding domain 4, DNA glycosylase (<scp>MBD4</scp>)â€associated neoplasia syndrome associated with a homozygous missense variant in MBD4 : Expansion of an emerging phenotype. British Journal of Haematology, 2022, , .	2.5	2
107	The HIDDEN Protocol: An Australian Prospective Cohort Study to Determine the Utility of Whole Genome Sequencing in Kidney Failure of Unknown Aetiology. Frontiers in Medicine, 0, 9, .	2.6	2
108	Response to Ferket et al.. Genetics in Medicine, 2020, 22, 1910.	2.4	0