

Use of Exome Sequencing for Infants in Intensive Care

JAMA Pediatrics

171, e173438

DOI: [10.1001/jamapediatrics.2017.3438](https://doi.org/10.1001/jamapediatrics.2017.3438)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Genetic disorders and mortality in infancy and early childhood: delayed diagnoses and missed opportunities. <i>Genetics in Medicine</i> , 2018, 20, 1396-1404.	1.1	58
2	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. <i>Npj Genomic Medicine</i> , 2018, 3, 6.	1.7	156
3	Exome and genome sequencing in reproductive medicine. <i>Fertility and Sterility</i> , 2018, 109, 213-220.	0.5	22
4	Paediatric genomics: diagnosing rare disease in children. <i>Nature Reviews Genetics</i> , 2018, 19, 253-268.	7.7	369
5	Exome sequencing helps diagnose infants in the ICU. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 8-9.	0.7	1
6	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. <i>Genetics in Medicine</i> , 2018, 20, 1554-1563.	1.1	125
7	Exome sequencing in neonates: diagnostic rates, characteristics, and time to diagnosis. <i>Genetics in Medicine</i> , 2018, 20, 1468-1471.	1.1	31
8	Applications of Pharmacogenomics in Oncology. <i>Advances in Molecular Pathology</i> , 2018, 1, 115-124.	0.2	1
9	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. <i>Advances in Molecular Pathology</i> , 2018, 1, 1-8.	0.2	0
10	The author replies. <i>Pediatric Critical Care Medicine</i> , 2018, 19, 694-695.	0.2	0
11	Metabolomics in the clinic: A review of the shared and unique features of untargeted metabolomics for clinical research and clinical testing. <i>Journal of Mass Spectrometry</i> , 2018, 53, 1143-1154.	0.7	69
12	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. <i>Genome Medicine</i> , 2018, 10, 74.	3.6	105
13	Next-Generation Sequencing to Diagnose Suspected Genetic Disorders. <i>New England Journal of Medicine</i> , 2018, 379, 1353-1362.	13.9	181
14	Classification, Ontology, and Precision Medicine. <i>New England Journal of Medicine</i> , 2018, 379, 1452-1462.	13.9	220
17	Clinical whole-exome sequencing results impact medical management. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 1068-1078.	0.6	33
18	Exome Sequencing in the Evaluation of the Fetus With Structural Anomalies. , 2018, , 289-305.		0
19	Clinical exome sequencing in France and Quebec: what are the challenges? What does the future hold?. <i>Life Sciences, Society and Policy</i> , 2018, 14, 17.	3.1	2
20	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1688-1691.	0.7	28

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21	Peri-mortem evaluation of infants who die without a diagnosis: focus on advances in genomic technology. <i>Journal of Perinatology</i> , 2018, 38, 1125-1134.	0.9	9
22	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
23	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. <i>Journal of Medical Genetics</i> , 2018, 55, 721-728.	1.5	98
24	SpainUDP: The Spanish Undiagnosed Rare Diseases Program. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1746.	1.2	19
25	Genome-wide sequencing in acutely ill infants: genomic medicine's critical application?. <i>Genetics in Medicine</i> , 2019, 21, 498-504.	1.1	42
26	Clinical Application of Genome and Exome Sequencing as a Diagnostic Tool for Pediatric Patients: a Scoping Review of the Literature. <i>Genetics in Medicine</i> , 2019, 21, 3-16.	1.1	96
27	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
28	Clinical genome sequencing in an unbiased pediatric cohort. <i>Genetics in Medicine</i> , 2019, 21, 303-310.	1.1	36
29	Genomic medicine for undiagnosed diseases. <i>Lancet</i> , The, 2019, 394, 533-540.	6.3	82
30	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
31	What is the role of next generation sequencing in status epilepticus?. <i>Epilepsy and Behavior</i> , 2019, 101, 106373.	0.9	5
32	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. <i>Frontiers in Genetics</i> , 2019, 10, 425.	1.1	33
33	Prenatally diagnosed developmental abnormalities of the central nervous system and genetic syndromes: A practical review. <i>Prenatal Diagnosis</i> , 2019, 39, 666-678.	1.1	27
34	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1821-1826.	1.4	19
35	Clinical genetic testing in endocrinology: Current concepts and contemporary challenges. <i>Clinical Endocrinology</i> , 2019, 91, 587-607.	1.2	17
36	Making a Genetic Diagnosis in a Level IV Neonatal Intensive Care Unit Population: Who, When, How, and at What Cost?. <i>Journal of Pediatrics</i> , 2019, 213, 211-217.e4.	0.9	26
37	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. <i>Genetics in Medicine</i> , 2019, 21, 2723-2733.	1.1	48
38	Exome and Genome Sequencing. , 2019, , 137-148.		0

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39	Genomic Sequencing for Infants and Children in Intensive Care Units. <i>Current Pediatrics Reports</i> , 2019, 7, 78-82.	1.7	1
40	A novel high-throughput molecular counting method with single base-pair resolution enables accurate single-gene NIPT. <i>Scientific Reports</i> , 2019, 9, 14382.	1.6	34
41	Toward Clinical Implementation of Next-Generation Sequencing-Based Genetic Testing in Rare Diseases: Where Are We?. <i>Trends in Genetics</i> , 2019, 35, 852-867.	2.9	65
42	Diagnostik seltener Erkrankungen mit "next generation sequencing" angekommen oder abgewehrt?. <i>Medizinische Genetik</i> , 2019, 31, 335-343.	0.1	1
43	DNA extraction from placental, fetal and neonatal tissue at autopsy: what organ to sample for DNA in the genomic era?. <i>Pathology</i> , 2019, 51, 705-710.	0.3	2
44	Infant mortality: the contribution of genetic disorders. <i>Journal of Perinatology</i> , 2019, 39, 1611-1619.	0.9	47
45	A Randomized, Controlled Trial of the Analytic and Diagnostic Performance of Singleton and Trio, Rapid Genome and Exome Sequencing in Ill Infants. <i>American Journal of Human Genetics</i> , 2019, 105, 719-733.	2.6	238
46	Introduction to Human Genetics. , 2019, , 1-17.		1
47	Many newborns in level IV NICUs are eligible for rapid DNA sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 280-284.	0.7	13
48	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. <i>Lancet, The</i> , 2019, 393, 758-767.	6.3	368
49	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
50	Comparison of medical management and genetic counseling options pre- and post-whole exome sequencing for patients with positive and negative results. <i>Journal of Genetic Counseling</i> , 2019, 28, 182-193.	0.9	13
51	Exome sequencing in the assessment of congenital malformations in the fetus and neonate. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2019, 104, fetalneonatal-2018-316352.	1.4	11
52	Targeted gene panel sequencing for the rapid diagnosis of acutely ill infants. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00796.	0.6	34
53	Molecular and phenotypic spectrum of Noonan syndrome in Chinese patients. <i>Clinical Genetics</i> , 2019, 96, 290-299.	1.0	29
54	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit" successes and challenges. <i>European Journal of Pediatrics</i> , 2019, 178, 1207-1218.	1.3	59
55	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
56	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. <i>Genome Medicine</i> , 2019, 11, 30.	3.6	42

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57	Whole-Exome Sequencing of Adult and Pediatric Cohorts of the Rare Vascular Disorder Systemic Capillary Leak Syndrome. <i>Shock</i> , 2019, 52, 183-190.	1.0	9
58	What do parents expect from a genetic diagnosis of their child with intellectual disability?. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2019, 32, 1129-1137.	1.3	8
59	<i>WNT10B</i> variants in split hand/foot malformation: Report of three novel families and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1351-1356.	0.7	4
60	Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	203
61	Case for genome sequencing in infants and children with rare, undiagnosed or genetic diseases. <i>Journal of Medical Genetics</i> , 2019, 56, 783-791.	1.5	93
62	Clinical utility in infants with suspected monogenic conditions through next-generation sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e684.	0.6	11
63	Incidentally identified genetic variants in arrhythmogenic right ventricular cardiomyopathy-associated genes among children undergoing exome sequencing reflect healthy population variation. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e593.	0.6	13
64	Assessing the Psychological Impact of Next-Generation Sequencing Information in the Clinic: An Attempt to Map Terra Incognita?. , 2019, , 103-123.		0
65	Use of whole exome sequencing in the NICU: Case of an extremely low birth weight infant with syndromic features. <i>Molecular and Cellular Probes</i> , 2019, 45, 89-93.	0.9	10
66	Genomic Medicine—Progress, Pitfalls, and Promise. <i>Cell</i> , 2019, 177, 45-57.	13.5	143
67	Recent developments in genetic/genomic medicine. <i>Clinical Science</i> , 2019, 133, 697-708.	1.8	80
68	Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. <i>Intensive Care Medicine</i> , 2019, 45, 627-636.	3.9	183
69	A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. <i>Journal of Genetic Counseling</i> , 2019, 28, 213-228.	0.9	11
70	Bayesian-based noninvasive prenatal diagnosis of single-gene disorders. <i>Genome Research</i> , 2019, 29, 428-438.	2.4	31
71	Determining the Likelihood of Variant Pathogenicity Using Amino Acid-level Signal-to-Noise Analysis of Genetic Variation. <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	10
72	Factors complicating the informed consent process for whole exome sequencing in neonatal and pediatric intensive care units. <i>Journal of Genetic Counseling</i> , 2019, 28, 256-262.	0.9	15
73	Genetic counseling considerations with rapid genome-wide sequencing in a neonatal intensive care unit. <i>Journal of Genetic Counseling</i> , 2019, 28, 263-272.	0.9	25
74	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

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75	Rapid Challenges: Ethics and Genomic Neonatal Intensive Care. <i>Pediatrics</i> , 2019, 143, S14-S21.	1.0	35
76	Diagnostic and clinical utility of whole genome sequencing in a cohort of undiagnosed Chinese families with rare diseases. <i>Scientific Reports</i> , 2019, 9, 19365.	1.6	28
77	Rapid Whole Genome Sequencing and Fulfilling the Promise of Precision Pediatric Critical Care*. <i>Pediatric Critical Care Medicine</i> , 2019, 20, 1085-1086.	0.2	7
78	Genetic causes of surfactant protein abnormalities. <i>Current Opinion in Pediatrics</i> , 2019, 31, 330-339.	1.0	37
79	Rethinking what constitutes a diagnosis in the genomics era: a critical illness perspective. <i>Current Opinion in Pediatrics</i> , 2019, 31, 317-321.	1.0	3
80	Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses*. <i>Pediatric Critical Care Medicine</i> , 2019, 20, 1021-1026.	0.2	29
81	Can precision medicine help achieve the goal of reducing care when the risks exceed the benefits?. <i>Personalized Medicine</i> , 2019, 16, 365-367.	0.8	0
82	Clinical utility of genomic sequencing. <i>Current Opinion in Pediatrics</i> , 2019, 31, 732-738.	1.0	14
83	Paving the way for precision medicine v2.0 in intensive care by profiling necroinflammation in biofluids. <i>Cell Death and Differentiation</i> , 2019, 26, 83-98.	5.0	10
84	Diagnostic Utility of Exome Sequencing for Kidney Disease. <i>New England Journal of Medicine</i> , 2019, 380, 142-151.	13.9	456
85	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. <i>American Journal of Human Genetics</i> , 2019, 104, 76-93.	2.6	176
86	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. <i>Annals of Internal Medicine</i> , 2019, 170, 11.	2.0	60
87	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 201-215.	3.0	110
88	Panel-Based Next-Generation Sequencing for the Diagnosis of Cholestatic Genetic Liver Diseases: Clinical Utility and Challenges. <i>Journal of Pediatrics</i> , 2019, 205, 153-159.e6.	0.9	40
89	Chromosomal Microarrays and Exome Sequencing for Diagnosis of Fetal Abnormalities. , 2019, , 577-595.		1
90	Putting genome-wide sequencing in neonates into perspective. <i>Genetics in Medicine</i> , 2019, 21, 1074-1082.	1.1	15
91	Next-Generation Sequencing for Gene Panels and Clinical Exomes. , 2019, , 553-575.		1
92	Genetic aetiology of early infant deaths in a neonatal intensive care unit. <i>Journal of Medical Genetics</i> , 2020, 57, 169-177.	1.5	22

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93	Clinical utility of exome sequencing in infantile heart failure. <i>Genetics in Medicine</i> , 2020, 22, 423-426.	1.1	17
94	A pediatric perspective on genomics and prevention in the twenty-first century. <i>Pediatric Research</i> , 2020, 87, 338-344.	1.1	3
95	Parents of newborns in the NICU enrolled in genome sequencing research: hopeful, but not naïve. <i>Genetics in Medicine</i> , 2020, 22, 416-422.	1.1	26
96	Translating genomic testing results for pediatric critical care: Opportunities for genetic counselors. <i>Journal of Genetic Counseling</i> , 2020, 29, 78-87.	0.9	13
97	Considerations for whole exome sequencing unique to prenatal care. <i>Human Genetics</i> , 2020, 139, 1149-1159.	1.8	18
98	Impact of Emerging Technologies in Prenatal Genetic Counseling. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036517.	2.9	3
99	When moments matter: Finding answers with rapid exome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1027.	0.6	12
100	Genetic Counseling and Genome Sequencing in Pediatric Rare Disease. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036632.	2.9	18
101	Clinical Exome Studies Have Inconsistent Coverage. <i>Clinical Chemistry</i> , 2020, 66, 199-206.	1.5	12
102	The current and future impact of genome-wide sequencing on fetal precision medicine. <i>Human Genetics</i> , 2020, 139, 1121-1130.	1.8	20
103	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. <i>Genetics in Medicine</i> , 2020, 22, 736-744.	1.1	83
104	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
105	The Impact of Rapid Exome Sequencing on Medical Management of Critically Ill Children. <i>Journal of Pediatrics</i> , 2020, 226, 202-212.e1.	0.9	35
106	Novel LRPPRC compound heterozygous mutation in a child with early-onset Leigh syndrome French-Canadian type: case report of an Italian patient. <i>Italian Journal of Pediatrics</i> , 2020, 46, 140.	1.0	18
107	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. <i>JAMA Network Open</i> , 2020, 3, e2018109.	2.8	47
108	Mitochondrial Diseases: A Diagnostic Revolution. <i>Trends in Genetics</i> , 2020, 36, 702-717.	2.9	73
109	Rapid Whole-Exome Sequencing as a Diagnostic Tool in a Neonatal/Pediatric Intensive Care Unit. <i>Journal of Clinical Medicine</i> , 2020, 9, 2220.	1.0	48
110	Children's rare disease cohorts: an integrative research and clinical genomics initiative. <i>Npj Genomic Medicine</i> , 2020, 5, 29.	1.7	38

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111	Simultaneous Detection of CNVs and SNVs Improves the Diagnostic Yield of Fetuses with Ultrasound Anomalies and Normal Karyotypes. <i>Genes</i> , 2020, 11, 1397.	1.0	30
112	Diagnostic power and clinical impact of exome sequencing in a cohort of 500 patients with rare diseases. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 955-964.	0.7	22
113	Measurement of genetic diseases as a cause of mortality in infants receiving whole genome sequencing. <i>Npj Genomic Medicine</i> , 2020, 5, 49.	1.7	29
114	Application of Next-Generation Sequencing for Genetic Diagnosis in Neonatal Intensive Care Units: Results of a Multicenter Study in China. <i>Frontiers in Genetics</i> , 2020, 11, 565078.	1.1	11
115	Causal Genetic Variants in Stillbirth. <i>New England Journal of Medicine</i> , 2020, 383, 1107-1116.	13.9	67
116	Rapid Phenotype-Driven Gene Sequencing with the NeoSeq Panel: A Diagnostic Tool for Critically Ill Newborns with Suspected Genetic Disease. <i>Journal of Clinical Medicine</i> , 2020, 9, 2362.	1.0	8
117	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. <i>Npj Genomic Medicine</i> , 2020, 5, 47.	1.7	67
118	CAPICE: a computational method for Consequence-Agnostic Pathogenicity Interpretation of Clinical Exome variations. <i>Genome Medicine</i> , 2020, 12, 75.	3.6	30
119	A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis. <i>Npj Genomic Medicine</i> , 2020, 5, 37.	1.7	54
120	Precision Child Health: an Emerging Paradigm for Paediatric Quality and Safety. <i>Current Treatment Options in Pediatrics</i> , 2020, 6, 317-324.	0.2	6
121	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. <i>Italian Journal of Pediatrics</i> , 2020, 46, 130.	1.0	14
122	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
123	Rapid whole-exome sequencing facilitates precision medicine in paediatric rare disease patients and reduces healthcare costs. <i>The Lancet Regional Health - Western Pacific</i> , 2020, 1, 100001.	1.3	40
124	An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. <i>American Journal of Human Genetics</i> , 2020, 107, 942-952.	2.6	110
125	A pilot study of expanded newborn screening for 573 genes related to severe inherited disorders in China: results from 1,127 newborns. <i>Annals of Translational Medicine</i> , 2020, 8, 1058-1058.	0.7	21
126	Discouraging Elective Genetic Testing of Minors: A Norm under Siege in a New Era of Genomic Medicine. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036657.	2.9	8
127	Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1601-1607.	0.7	10
128	Actin Mutations and Their Role in Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3371.	1.8	44

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129	Clinical utility of 24-h rapid trio-exome sequencing for critically ill infants. <i>Npj Genomic Medicine</i> , 2020, 5, 20.	1.7	41
130	Disease-associated mosaic variation in clinical exome sequencing: a two-year pediatric tertiary care experience. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005231.	0.5	15
131	Genetic testing strategies in the newborn. <i>Journal of Perinatology</i> , 2020, 40, 1007-1016.	0.9	9
132	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
133	Delivering genome sequencing for rapid genetic diagnosis in critically ill children: parent and professional views, experiences and challenges. <i>European Journal of Human Genetics</i> , 2020, 28, 1529-1540.	1.4	29
134	Whole-exome Sequencing for the Identification of Rare Variants in Primary Immunodeficiency Genes in Children With Sepsis: A Prospective, Population-based Cohort Study. <i>Clinical Infectious Diseases</i> , 2020, 71, e614-e623.	2.9	12
135	Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. <i>Genetics in Medicine</i> , 2020, 22, 986-1004.	1.1	53
136	Neonatal respiratory failure due to novel compound heterozygous mutations in the ABCA3 lipid transporter. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005074.	0.5	4
137	Use of ultra-rapid whole-exome sequencing to diagnose congenital central hypoventilation syndrome. <i>Pediatric Pulmonology</i> , 2020, 55, 855-857.	1.0	2
138	Optimized trio genome sequencing (OTGS) as a first-tier genetic test in critically ill infants: practice in China. <i>Human Genetics</i> , 2020, 139, 473-482.	1.8	51
139	Exome sequencing compared with standard genetic tests for critically ill infants with suspected genetic conditions. <i>Genetics in Medicine</i> , 2020, 22, 1303-1310.	1.1	21
140	Nonimmune hydrops fetalis: Genetic analysis and clinical outcome. <i>Prenatal Diagnosis</i> , 2020, 40, 803-812.	1.1	17
141	Rapid exome sequencing in PICU patients with new-onset metabolic or neurological disorders. <i>Pediatric Research</i> , 2020, 88, 761-768.	1.1	19
142	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. <i>Prenatal Diagnosis</i> , 2020, 40, 972-983.	1.1	49
143	Developmental Support for Infants With Genetic Disorders. <i>Pediatrics</i> , 2020, 145, e20190629.	1.0	5
144	Bridging the Gap between Scientific Advancement and Real-World Application: Pediatric Genetic Counseling for Common Syndromes and Single-Gene Disorders. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036640.	2.9	4
145	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. <i>Canadian Journal of Neurological Sciences</i> , 2021, 48, 233-244.	0.3	4
146	The role of next-generation sequencing in the investigation of ultrasound-identified fetal structural anomalies. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2021, 128, 420-429.	1.1	23

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147	The role of clinical response to treatment in determining pathogenicity of genomic variants. <i>Genetics in Medicine</i> , 2021, 23, 581-585.	1.1	18
148	Ultra-rapid emergency genomic diagnosis of Donahue syndrome in a preterm infant within 17 hours. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 90-96.	0.7	14
149	Amino Acid-Level Signal-to-Noise Analysis Aids in Pathogenicity Prediction of Incidentally Identified <i>TTN</i> -Encoded Titin Truncating Variants. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003131.	1.6	7
150	Rapid acute care genomics: Challenges and opportunities for genetic counselors. <i>Journal of Genetic Counseling</i> , 2021, 30, 30-41.	0.9	15
151	The influence of social determinants of health on the genetic diagnostic odyssey: who remains undiagnosed, why, and to what effect?. <i>Pediatric Research</i> , 2021, 89, 295-300.	1.1	47
152	Rapid genome-wide sequencing in a neonatal intensive care unit: A retrospective qualitative exploration of parental experiences. <i>Journal of Genetic Counseling</i> , 2021, 30, 616-629.	0.9	17
153	Evidence that <i>FGFRL1</i> contributes to congenital diaphragmatic hernia development in humans. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 836-840.	0.7	8
154	Improved noninvasive fetal variant calling using standardized benchmarking approaches. <i>Computational and Structural Biotechnology Journal</i> , 2021, 19, 509-517.	1.9	1
155	High-Resolution Cryo-EM Structure of the Cardiac Actomyosin Complex. <i>Structure</i> , 2021, 29, 50-60.e4.	1.6	41
156	Application of Next Generation Sequencing in Laboratory Medicine. <i>Annals of Laboratory Medicine</i> , 2021, 41, 25-43.	1.2	99
157	Primary immunodeficiency-related genes in neonatal intensive care unit patients with various genetic immune abnormalities: a multicentre study in China. <i>Clinical and Translational Immunology</i> , 2021, 10, e1266.	1.7	6
158	Improving diagnostics of rare genetic diseases with NGS approaches. <i>Journal of Community Genetics</i> , 2021, 12, 247-256.	0.5	25
159	Rare versus common diseases: a false dichotomy in precision medicine. <i>Npj Genomic Medicine</i> , 2021, 6, 19.	1.7	14
160	Diagnostic and clinical utility of next-generation sequencing in children born with multiple congenital anomalies in the China neonatal genomes project. <i>Human Mutation</i> , 2021, 42, 434-444.	1.1	15
161	Genetic testing for unexplained perinatal disorders. <i>Current Opinion in Pediatrics</i> , 2021, 33, 195-202.	1.0	15
162	<i>ABCA3</i> gene mutations shape the clinical profiles of severe unexplained respiratory distress syndrome in late preterm and term infants. <i>Translational Pediatrics</i> , 2021, 10, 350-358.	0.5	6
163	Genetic Testing for Neonatal Respiratory Disease. <i>Children</i> , 2021, 8, 216.	0.6	5
164	Leigh syndrome associated with <i>TRMU</i> gene mutations. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100690.	0.4	3

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165	Characterization of genetic counselor practices in inpatient care settings. <i>Journal of Genetic Counseling</i> , 2021, 30, 1181-1190.	0.9	3
166	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. <i>Genetics in Medicine</i> , 2021, 23, 1372-1375.	1.1	47
167	Hematological findings associated with tubulinâ€folding cofactors Dâ€related encephalopathy: Expanding the phenotype. <i>Clinical Genetics</i> , 2021, 99, 724-731.	1.0	0
168	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
170	Exome sequencing as the firstâ€tier test for pediatric respiratory diseases: A singleâ€center study. <i>Human Mutation</i> , 2021, 42, 891-900.	1.1	2
171	La tecnologia genetica: ciÃ² che ogni pediatra dovrebbe sapere. <i>Medico E Bambino</i> , 2021, 40, 291-301.	0.1	2
172	Diagnosing newborns with suspected mitochondrial disorders: an economic evaluation comparing early exome sequencing to current typical care. <i>Genetics in Medicine</i> , 2021, 23, 1854-1863.	1.1	4
173	Prevalence of monogenic disease in paediatric patients with a predominant respiratory phenotype. <i>Archives of Disease in Childhood</i> , 2021, , archdischild-2021-322058.	1.0	0
174	Enrichment of low abundance DNA/RNA by oligonucleotide-clicked iron oxide nanoparticles. <i>Scientific Reports</i> , 2021, 11, 13053.	1.6	7
175	Towards improved genetic diagnosis of human differences of sex development. <i>Nature Reviews Genetics</i> , 2021, 22, 588-602.	7.7	35
176	Screening of Candidate Pathogenic Genes for Spontaneous Abortion using Whole Exome Sequencing. <i>Combinatorial Chemistry and High Throughput Screening</i> , 2021, 24, .	0.6	0
177	Diagnostic yield of rare skeletal dysplasia conditions in the radiogenomics era. <i>BMC Medical Genomics</i> , 2021, 14, 148.	0.7	7
178	Project Baby Bear: Rapid precision care incorporating rWGS in 5 California childrenâ€™s hospitals demonstrates improved clinical outcomes and reduced costs of care. <i>American Journal of Human Genetics</i> , 2021, 108, 1231-1238.	2.6	140
179	Utility of clinical exome sequencing in the evaluation of neonates with suspected genetic condition â€“ An observational study from tertiary neonatal care unit in South India. <i>European Journal of Medical Genetics</i> , 2021, 64, 104247.	0.7	4
180	5G Edge Computing Enabled Directional Data Collection for Medical Community Electronic Health Records. <i>Journal of Healthcare Engineering</i> , 2021, 2021, 1-12.	1.1	4
181	The Unrecognized Mortality Burden of Genetic Disorders in Infancy. <i>American Journal of Public Health</i> , 2021, 111, S156-S162.	1.5	5
182	Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 2029-2037.	1.1	229
183	Expanding the genotypes and phenotypes for 19 rare diseases by exome sequencing performed in pediatric intensive care unit. <i>Human Mutation</i> , 2021, 42, 1443-1460.	1.1	4

#	ARTICLE	IF	CITATIONS
184	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
185	Congenital anomalies and genetic disorders in neonates and infants: a single-center observational cohort study. <i>European Journal of Pediatrics</i> , 2022, 181, 359-367.	1.3	7
186	Next-Generation Sequencing Reveals Novel Genetic Variants for Dilated Cardiomyopathy in Pediatric Chinese Patients. <i>Pediatric Cardiology</i> , 2022, 43, 110-120.	0.6	7
187	Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease. <i>JAMA Pediatrics</i> , 2021, 175, 1218.	3.3	83
188	Diagnostic yield of clinical exome sequencing as a first-tier genetic test for the diagnosis of genetic disorders in pediatric patients: results from a referral center study. <i>Human Genetics</i> , 2022, 141, 1269-1278.	1.8	10
189	Amplification of a minimally biased antibody repertoire for in vitro display using a universal primer-based amplification method. <i>Journal of Immunological Methods</i> , 2021, 496, 113089.	0.6	2
190	Clinical Application of Whole Exome Sequencing for Monogenic Disorders in PICU of China. <i>Frontiers in Genetics</i> , 2021, 12, 677699.	1.1	6
191	Implementing Rapid Whole-Genome Sequencing in Critical Care: A Qualitative Study of Facilitators and Barriers to New Technology Adoption. <i>Journal of Pediatrics</i> , 2021, 237, 237-243.e2.	0.9	31
192	Multilocus inheritance and variable disease expressivity in rare disease. , 2021, , 185-204.		0
193	Genomic sequencing of rare diseases. , 2021, , 61-95.		6
195	Exome sequencing in genetic disease: recent advances and considerations. <i>F1000Research</i> , 2020, 9, 336.	0.8	22
196	Neonatal Presentations of Metabolic Disorders. <i>NeoReviews</i> , 2020, 21, e649-e662.	0.4	6
197	Use of medical exome sequencing for identification of underlying genetic defects in <scp>NICU</scp>: Experience in a cohort of 2303 neonates in China. <i>Clinical Genetics</i> , 2022, 101, 101-109.	1.0	16
198	Dâ€bifunctional protein deficiency caused by splicing variants in a neonate with severe peroxisomal dysfunction and persistent hypoglycemia. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	0.7	2
199	Neonatal Metabolic Acidosis in the Neonatal Intensive Care Unit: What Are the Genetic Causes?. <i>Frontiers in Pediatrics</i> , 2021, 9, 727301.	0.9	0
201	Genomic Applications in Inherited Genetic Disorders. , 2019, , 543-560.		0
202	A Case of Early Diagnosis of Pyruvate Dehydrogenase Complex Deficiency: The Use of Next-Generation Sequencing. <i>Iranian Journal of Pediatrics</i> , 2019, In Press, .	0.1	0
204	Deciphering congenital anomalies for the next generation. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005504.	0.5	5

#	ARTICLE	IF	CITATIONS
205	Somatic variation as an incidental finding in the pediatric next-generation sequencing era. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006135.	0.5	3
206	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 916-922.	0.7	1
207	Ethical Issues in Clinical Child Neurology. , 2020, , 1393-1404.		0
208	Genomics for the Neonatologist. , 2020, , 545-557.		0
209	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
210	Select Ethical Aspects of Next-Generation Sequencing Tests for Newborn Screening and Diagnostic Evaluation of Critically Ill Newborns. <i>International Journal of Neonatal Screening</i> , 2021, 7, 76.	1.2	5
211	Genome-Wide Sequencing for Unexplained Developmental Disabilities or Multiple Congenital Anomalies: A Health Technology Assessment. <i>Ontario Health Technology Assessment Series</i> , 2020, 20, 1-178.	3.0	11
215	How neonatologists use genetic testing: findings from a national survey. <i>Journal of Perinatology</i> , 2022, 42, 260-261.	0.9	5
216	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. <i>European Journal of Human Genetics</i> , 2022, 30, 567-576.	1.4	12
218	Case Report: An Infant With Kabuki Syndrome, Alobar Holoprosencephaly and Truncus Arteriosus: A Case for Whole Exome Sequencing in Neonates With Congenital Anomalies. <i>Frontiers in Genetics</i> , 2021, 12, 766316.	1.1	1
219	Approach to the Child With Dysmorphism. , 2021, , .		0
220	Exome and Whole Genome Sequencing in the Neonatal Intensive Care Unit. <i>Clinics in Perinatology</i> , 2022, 49, 167-179.	0.8	6
221	Singleton exome sequencing of 90 fetuses with ultrasound anomalies revealing novel disease-causing variants and genotypeâ€“phenotype correlations. <i>European Journal of Human Genetics</i> , 2022, 30, 428-438.	1.4	6
222	PhenoApt leverages clinical expertise to prioritize candidate genes via machine learning. <i>American Journal of Human Genetics</i> , 2022, 109, 270-281.	2.6	5
223	Genetic diagnosis of basal ganglia disease in childhood. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 743-752.	1.1	0
224	Molecular Diagnostic Outcomes from 700 Cases. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 274-286.	1.2	7
225	Cost Efficacy of Rapid Whole Genome Sequencing in the Pediatric Intensive Care Unit. <i>Frontiers in Pediatrics</i> , 2021, 9, 809536.	0.9	18
226	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. <i>Journal of Pediatrics</i> , 2022, 244, 38-48.e1.	0.9	8

#	ARTICLE	IF	CITATIONS
227	Evaluating use of changing technologies for rapid next-generation sequencing in pediatrics. <i>Pediatric Research</i> , 2022, 92, 1364-1369.	1.1	8
228	Towards Next-Generation Sequencing (NGS)-Based Newborn Screening: A Technical Study to Prepare for the Challenges Ahead. <i>International Journal of Neonatal Screening</i> , 2022, 8, 17.	1.2	15
229	Influence of Genetic Information on Neonatologists'™ Decisions: A Psychological Experiment. <i>Pediatrics</i> , 2022, 149, .	1.0	12
230	Detection and impact of genetic disease in a level IV neonatal intensive care unit. <i>Journal of Perinatology</i> , 2022, 42, 580-588.	0.9	6
231	Patterns of multiple congenital anomalies in the National Birth Defect Prevention Study: Challenges and insights. <i>Birth Defects Research</i> , 2023, 115, 43-55.	0.8	4
232	Implications of Dosage Deficiencies in CTCF and Cohesin on Genome Organization, Gene Expression, and Human Neurodevelopment. <i>Genes</i> , 2022, 13, 583.	1.0	10
233	Perspectives of United States neonatologists on genetic testing practices. <i>Genetics in Medicine</i> , 2022, 24, 1372-1377.	1.1	6
234	Platelet VPS16B is dependent on VPS33B expression, as determined in two siblings with arthrogyriposis, renal dysfunction and cholestasis (ARC) syndrome. <i>Journal of Thrombosis and Haemostasis</i> , 2022, , .	1.9	1
235	Healthcare Professionals'™ Attitudes toward Rapid Whole Genome Sequencing in Pediatric Acute Care. <i>Children</i> , 2022, 9, 357.	0.6	4
236	Ethical Considerations for Equitable Access to Genomic Sequencing for Critically Ill Neonates in the United States. <i>International Journal of Neonatal Screening</i> , 2022, 8, 22.	1.2	7
237	Cost-effectiveness of exome and genome sequencing for children with rare and undiagnosed conditions. <i>Genetics in Medicine</i> , 2022, 24, 1349-1361.	1.1	25
238	Rapid Exome and Genome Sequencing in the Intensive Care Unit. <i>Critical Care Clinics</i> , 2022, 38, 173-184.	1.0	5
239	Lessons learned: next-generation sequencing applied to undiagnosed genetic diseases. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	11
240	Parental mosaicism for apparent de novo genetic variants: Scope, detection, and counseling challenges. <i>Prenatal Diagnosis</i> , 2022, 42, 811-821.	1.1	8
242	Inborn Errors of Immunity in the Premature Infant: Challenges in Recognition and Diagnosis. <i>Frontiers in Immunology</i> , 2021, 12, 758373.	2.2	7
243	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. <i>Genetics in Medicine</i> , 2022, , .	1.1	4
247	From newborn screening to genomic medicine: challenges and suggestions on how to incorporate genomic newborn screening in public health programs. <i>Medizinische Genetik</i> , 2022, 34, 13-20.	0.1	2
248	Genomic architecture of fetal central nervous system anomalies using whole-genome sequencing. <i>Npj Genomic Medicine</i> , 2022, 7, 31.	1.7	6

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249	Case Report: Novel Biallelic Null Variants of SMPD4 Confirm Its Involvement in Neurodevelopmental Disorder With Microcephaly, Arthrogyrosis, and Structural Brain Anomalies. <i>Frontiers in Genetics</i> , 2022, 13, .	1.1	3
250	Comparing genome sequencing technologies to improve rare disease diagnostics: a protocol for the evaluation of a pilot project, <i>Genome-wide Sequencing Ontario. CMAJ Open</i> , 2022, 10, E460-E465.	1.1	3
251	The effect of rapid exome sequencing on downstream health care utilization for infants with suspected genetic disorders in an intensive care unit. <i>Genetics in Medicine</i> , 2022, 24, 1675-1683.	1.1	3
252	“Doctors shouldn’t have to cheat the system”: Clinicians’ real-world experiences of the utility of genomic sequencing. <i>Genetics in Medicine</i> , 2022, , .	1.1	5
253	The Role of Genome Sequencing in Neonatal Intensive Care Units. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, 427-448.	2.5	23
254	Implementing genomics in the neonatal period: An assessment of parental decision making and anxiety. <i>Journal of Genetic Counseling</i> , 2022, 31, 1306-1316.	0.9	3
255	A model to implement genomic medicine in the neonatal intensive care unit. <i>Journal of Perinatology</i> , 2023, 43, 248-252.	0.9	6
256	Better and faster is cheaper. <i>Human Mutation</i> , 2022, 43, 1495-1506.	1.1	2
257	Rapid exome sequencing in critically ill infants: implementation in routine care from French regional hospital’s perspective. <i>European Journal of Human Genetics</i> , 2022, 30, 1076-1082.	1.4	5
258	Evidence for an association between Coffinâ€širis syndrome and congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2718-2723.	0.7	3
259	Rapid exome sequencing in critically ill children impacts acute and long-term management of patients and their families: A retrospective regional evaluation. <i>European Journal of Medical Genetics</i> , 2022, 65, 104571.	0.7	5
260	A retrospective cohort analysis of the Yale pediatric genomics discovery program. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2869-2878.	0.7	2
261	Prenatal phenotyping: A community effort to enhance the Human Phenotype Ontology. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, 190, 231-242.	0.7	14
262	Prenatal exome and genome sequencing for fetal structural abnormalities. <i>American Journal of Obstetrics and Gynecology</i> , 2023, 228, 140-149.	0.7	6
263	Next Generation Sequencing after Invasive Prenatal Testing in Fetuses with Congenital Malformations: Prenatal or Neonatal Investigation. <i>Genes</i> , 2022, 13, 1517.	1.0	10
264	Integrating rapid exome sequencing into NICU clinical care after a pilot research study. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	10
265	From diagnostic testing to precision medicine: the evolving role of genomics in cardiac channelopathies and cardiomyopathies in children. <i>Current Opinion in Genetics and Development</i> , 2022, 76, 101978.	1.5	1
266	Diagnostic utility of rapid sequencing in critically ill infants: a systematic review and meta-analysis. <i>Expert Review of Molecular Diagnostics</i> , 2022, 22, 833-840.	1.5	4

#	ARTICLE	IF	CITATIONS
267	Using the Sankey diagram to visualize article features on the topics of whole-exome sequencing (WES) and whole-genome sequencing (WGS) since 2012: Bibliometric analysis. <i>Medicine (United States)</i> , 2022, 101, e30682.	0.4	7
268	Genetic testing of sperm donors at a human sperm bank in China. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	0
269	Exome sequencing efficacy and phenotypic expansions involving esophageal atresia/tracheoesophageal fistula plus. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	2
270	Rapid Genetic Testing in Pediatric and Neonatal Critical Care: A Scoping Review of Emerging Ethical Issues. <i>Hospital Pediatrics</i> , 2022, 12, e347-e359.	0.6	2
271	Current Practices for Genetic Testing in Neonatal Extracorporeal Membrane Oxygenation: Findings from a National survey. <i>Perfusion (United Kingdom)</i> , 2024, 39, 116-123.	0.5	2
272	Rapid Genome Sequencing: Consent for New Technologies in the Neonatal Intensive Care Context. <i>Pediatrics</i> , 2022, 150, .	1.0	2
273	Rapid Targeted Genomic Testing: A Powerful Tool for Diagnostic Evaluation of Critically Ill Neonates and Infants With Suspected Genetic Diseases. <i>Annals of Laboratory Medicine</i> , 2023, 43, 223-224.	1.2	0
274	A robust pipeline for ranking carrier frequencies of autosomal recessive and X-linked Mendelian disorders. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	3
275	Pitfalls of whole exome sequencing in undefined clinical conditions with a suspected genetic etiology. <i>Genes and Genomics</i> , 0, , .	0.5	1
276	Expansion of the genotypic and phenotypic spectrum of <sc><i>CTCF</i></sc>-related disorder guides clinical management: 43 new subjects and a comprehensive literature review. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 718-729.	0.7	6
277	High molecular diagnostic yields and novel phenotypic expansions involving syndromic anorectal malformations. <i>European Journal of Human Genetics</i> , 0, , .	1.4	4
279	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. <i>Annals of Laboratory Medicine</i> , 2023, 43, 280-289.	1.2	4
280	Clinical impact of exome sequencing in the setting of a general pediatric ward for hospitalized children with suspected genetic disorders. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	0
281	Insights into the perinatal phenotype of Kabuki syndrome in infants identified by genome-wide sequencing. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
282	A Survey on Genetic Disorder Prediction of Fetus from Ultrasound-Based Computer-Aided Diagnosis. , 2022, , .		0
283	Precision diagnostics in children. , 2023, 1, .		0
285	Role of genomic medicine and implementing equitable access for critically ill infants in neonatal intensive care units. <i>Journal of Perinatology</i> , 2023, 43, 963-967.	0.9	3
286	Evaluation of an automated genome interpretation model for rare disease routinely used in a clinical genetic laboratory. <i>Genetics in Medicine</i> , 2023, 25, 100830.	1.1	4

#	ARTICLE	IF	CITATIONS
287	A Retrospective Analysis of Clinically Focused Exome Sequencing Results of 372 Infants with Suspected Monogenic Disorders in China. <i>Pharmacogenomics and Personalized Medicine</i> , 0, Volume 16, 81-97.	0.4	1
288	Predictors of the utility of clinical exome sequencing as a first-tier genetic test in patients with Mendelian phenotypes: results from a referral center study on 603 consecutive cases. <i>Human Genomics</i> , 2023, 17, .	1.4	2
289	Esophageal Atresia With or Without Tracheoesophageal Fistula: Comorbidities, Genetic Evaluations, and Neonatal Outcomes. <i>Cureus</i> , 2023, , .	0.2	0
290	Reclassification of the Etiology of Infant Mortality With Whole-Genome Sequencing. <i>JAMA Network Open</i> , 2023, 6, e2254069.	2.8	15
291	<i>KCNA1</i> gain-of-function epileptic encephalopathy treated with 4-aminopyridine. <i>Annals of Clinical and Translational Neurology</i> , 2023, 10, 656-663.	1.7	8
292	Rapid Whole Genome Sequencing Diagnoses and Guides Treatment in Critically Ill Children in Belgium in Less than 40 Hours. <i>International Journal of Molecular Sciences</i> , 2023, 24, 4003.	1.8	7
295	We've opened Pandora's box, haven't we?—clinical geneticists' views on ethical aspects of genomic testing in neonatal intensive care. <i>Balkan Journal of Medical Genetics</i> , 2023, 25, 5-12.	0.5	0
296	Implications of Genomic Newborn Screening for Infant Mortality. <i>International Journal of Neonatal Screening</i> , 2023, 9, 12.	1.2	2
298	Novel mutation causing Zellweger syndrome. <i>BMJ Case Reports</i> , 2023, 16, e252014.	0.2	0
299	Hepatology Genome Rounds: An interdisciplinary approach to integrate genomic data into clinical practice. <i>Journal of Hepatology</i> , 2023, 79, 1065-1071.	1.8	2
300	Chromosomal microarrays and next-generation sequencing for diagnosis of fetal abnormalities. , 2023, , 767-787.		0
301	Next-generation sequencing for gene panels, clinical exome, and whole-genome analysis. , 2023, , 743-766.		0
307	A precision environmental health approach to prevention of human disease. <i>Nature Communications</i> , 2023, 14, .	5.8	12
322	Genomic medicine in neonatal care: progress and challenges. <i>European Journal of Human Genetics</i> , 0, , .	1.4	2
329	The clock is ticking — combination of genome sequencing and omics analyses for the diagnosis of critically ill children. <i>DGNeurologie</i> , 0, , .	0.0	0