Prenatal exome sequencing analysis in fetal structural a ultrasonography (PAGE): a cohort study

Lancet, The 393, 747-757 DOI: 10.1016/s0140-6736(18)31940-8

Citation Report

#	Article	IF	CITATIONS
1	Ultrasound examination: The key to maximising the benefits of advances in molecular diagnostic technologies. Prenatal Diagnosis, 2019, 39, 663-665.	2.3	2
2	Prenatal chromosomal microarray testing of fetuses with ultrasound structural anomalies: A prospective cohort study of over 1000 consecutive cases. Prenatal Diagnosis, 2019, 39, 1064-1069.	2.3	16
4	Fetal arthrogryposis: Challenges and perspectives for prenatal detection and management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 327-336.	1.6	29
5	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. Frontiers in Genetics, 2019, 10, 425.	2.3	33
6	Prenatally diagnosed developmental abnormalities of the central nervous system and genetic syndromes: A practical review. Prenatal Diagnosis, 2019, 39, 666-678.	2.3	27
7	Fetal phenotypes emerge as genetic technologies become robust. Prenatal Diagnosis, 2019, 39, 811-817.	2.3	37
8	Prenatal genetic considerations of congenital anomalies of the kidney and urinary tract (CAKUT). Prenatal Diagnosis, 2019, 39, 679-692.	2.3	39
9	Looking Back at Fetal Medicine in India in 2018, and Looking Forward to 2019. Journal of Fetal Medicine, 2019, 06, 47-50.	0.1	2
10	Is it feasible to select fetuses for prenatal WES based on the prenatal phenotype?. Prenatal Diagnosis, 2019, 39, 1039-1040.	2.3	14
11	Human Genetics and Fetal Disease: Assessment of the Fetal Genome. , 2019, , 36-47.		0
12	A comprehensive biomedical variant catalogue based on whole genome sequences of 582 dogs and eight wolves. Animal Genetics, 2019, 50, 695-704.	1.7	138
13	Prenatal Diagnosis of Fetuses With Increased Nuchal Translucency by Genome Sequencing Analysis. Frontiers in Genetics, 2019, 10, 761.	2.3	52
14	Introduction of genomics into prenatal diagnostics. Lancet, The, 2019, 393, 719-721.	13.7	13
15	Exome sequencing in the assessment of congenital malformations in the fetus and neonate. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2019, 104, fetalneonatal-2018-316352.	2.8	11
16	A systemâ€based approach to the genetic etiologies of nonâ€immune hydrops fetalis. Prenatal Diagnosis, 2019, 39, 732-750.	2.3	34
17	From diagnostic yield to clinical impact: a pilot study on the implementation of prenatal exome sequencing in routine care. Genetics in Medicine, 2019, 21, 2303-2310.	2.4	41
18	Antenatal Fetal Assessment: 75 Years Later (1945-2019). Journal of Obstetrics and Gynaecology Canada, 2019, 41, S276-S280.	0.7	0
19	Relationships between the clinical phenotypes and genetic variants associated with the immunological mechanism in childhood idiopathic nephrotic syndrome: protocol for a prospective observational single-centre cohort study. BMJ Open, 2019, 9, e028717.	1.9	0

#	Article	IF	CITATIONS
20	Évaluation fÅ"tale prénataleÂ: 75Âans plus tard (1945-2019). Journal of Obstetrics and Gynaecology Canada, 2019, 41, S281-S286.	0.7	0
21	Chromosomal copy number variations in products of conception from spontaneous abortion by next-generation sequencing technology. Medicine (United States), 2019, 98, e18041.	1.0	17
22	From sub-microscopic variants to the resolution of a single base pair: Exome sequencing in prenatal diagnosis. European Journal of Medical Genetics, 2020, 63, 103779.	1.3	0
23	Perspectives of US private payers on insurance coverage for pediatric and prenatal exome sequencing: Results of a study from the Program in Prenatal and Pediatric Genomic Sequencing (P3EGS). Genetics in Medicine, 2020, 22, 283-291.	2.4	41
24	Contribution of singleâ€gene defects to congenital cardiac leftâ€sided lesions in the prenatal setting. Ultrasound in Obstetrics and Gynecology, 2020, 56, 225-232.	1.7	32
25	Two novel mutations of <i>COL1A1</i> in fetal genetic skeletal dysplasia of Chinese. Molecular Genetics & Genomic Medicine, 2020, 8, e1105.	1.2	8
26	Case report: targeted whole exome sequencing enables the first prenatal diagnosis of the lethal skeletal dysplasia Osteocraniostenosis. BMC Medical Genetics, 2020, 21, 7.	2.1	5
27	In case you missed it: The <i>Prenatal Diagnosis</i> editors bring you the most significant advances of 2019. Prenatal Diagnosis, 2020, 40, 287-293.	2.3	6
28	Diagnosis of fetal abnormalities using exome sequencing: translating research into practice. Ultrasound in Obstetrics and Gynecology, 2020, 56, 779-779.	1.7	2
29	Genetic syndromes associated with isolated fetal growth restriction. Prenatal Diagnosis, 2020, 40, 432-446.	2.3	42
30	Exome sequencing for perinatal phenotypes: The significance of deep phenotyping. Prenatal Diagnosis, 2020, 40, 260-273.	2.3	20
31	The use of fetal exome sequencing in prenatal diagnosis: a points to consider document of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 675-680.	2.4	128
32	Recurrent hypoplasia of corpus callosum as a prenatal phenotype of Xia-Gibbs syndrome caused by maternal germline mosaicism of an AHDC1 variant. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 244, 208-210.	1.1	11
33	A prenatally diagnosed case of Donnaiâ€Barrow syndrome: Highlighting the importance of whole exome sequencing in cases of consanguinity. American Journal of Medical Genetics, Part A, 2020, 182, 289-292.	1.2	9
34	The current and future impact of genome-wide sequencing on fetal precision medicine. Human Genetics, 2020, 139, 1121-1130.	3.8	20
35	Accuracy of in-utero MRI to detect fetal brain abnormalities and prognosticate developmental outcome: postnatal follow-up of the MERIDIAN cohort. The Lancet Child and Adolescent Health, 2020, 4, 131-140.	5.6	25
36	Positive Rate of Noninvasive Prenatal Screening for Pregnancies with Fetal Congenital Heart Disease and Its Impact on Pregnancy Outcome. Maternal-Fetal Medicine, 2020, 2, 84-88.	0.8	0
37	Nonimmune Hydrops Fetalis — More Than Meets the Eye?. New England Journal of Medicine, 2020, 383, 1785-1786.	27.0	3

#	Article	IF	CITATIONS
38	Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis. New England Journal of Medicine, 2020, 383, 1746-1756.	27.0	114
39	Genome-wide noninvasive prenatal diagnosis of monogenic disorders: Current and future trends. Computational and Structural Biotechnology Journal, 2020, 18, 2463-2470.	4.1	22
40	Single nucleotide polymorphism array analysis of 102 patients with developmental delay and/or intellectual disability from Fujian, China. Clinica Chimica Acta, 2020, 510, 638-643.	1.1	2
41	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
42	Noninvasive prenatal sequencing for multiple Mendelian monogenic disorders among fetuses with skeletal dysplasia or increased nuchal translucency. Prenatal Diagnosis, 2020, 40, 1459-1465.	2.3	11
43	Exome sequencing improves genetic diagnosis of fetal increased nuchal translucency. Prenatal Diagnosis, 2020, 40, 1426-1431.	2.3	22
44	Rapid prenatal diagnosis of skeletal dysplasia using medical trio exome sequencing: Benefit for prenatal counseling and pregnancy management. Prenatal Diagnosis, 2020, 40, 577-584.	2.3	42
45	Simultaneous Detection of CNVs and SNVs Improves the Diagnostic Yield of Fetuses with Ultrasound Anomalies and Normal Karyotypes. Genes, 2020, 11, 1397.	2.4	30
46	Clinical and genetic characteristics and prenatal diagnosis of patients presented GDD/ID with rare monogenic causes. Orphanet Journal of Rare Diseases, 2020, 15, 317.	2.7	5
47	Stillbirth. Obstetrics and Gynecology Clinics of North America, 2020, 47, 439-451.	1.9	10
48	Application of exome sequencing for prenatal diagnosis: a rapid scoping review. Genetics in Medicine, 2020, 22, 1925-1934.	2.4	25
49	Causal Genetic Variants in Stillbirth. New England Journal of Medicine, 2020, 383, 1107-1116.	27.0	67
50	Prenatal clinical manifestations in individuals with <i>COL4A1/2</i> variants. Journal of Medical Genetics, 2021, 58, 505-513.	3.2	22
51	Molecular diagnostic in fetuses with isolated congenital anomalies of the kidney and urinary tract by wholeâ€exome sequencing. Journal of Clinical Laboratory Analysis, 2020, 34, e23480.	2.1	15
52	Gene and Stem Cell Therapies for Fetal Care. JAMA Pediatrics, 2020, 174, 985.	6.2	11
53	Novel and recurrent variants identified in fetuses with central nervous system abnormalities by trios-medical exome sequencing. Clinica Chimica Acta, 2020, 510, 599-604.	1.1	14
54	Genetic diagnosis and clinical evaluation of severe fetal akinesia syndrome. Prenatal Diagnosis, 2020, 40, 1532-1539.	2.3	15
55	Comprehensive evaluation of genetic variants using chromosomal microarray analysis and exome sequencing in fetuses with congenital heart defect. Ultrasound in Obstetrics and Gynecology, 2021, 58, 377-387.	1.7	31

#	Article	IF	CITATIONS
56	Wholeâ€exome sequencing in the evaluation of fetal congenital anomalies of the kidney and urinary tract detected by ultrasonography. Prenatal Diagnosis, 2020, 40, 1290-1299.	2.3	24
5 7	Benefits and limitations of noninvasive prenatal aneuploidy screening. JAAPA: Official Journal of the American Academy of Physician Assistants, 2020, 33, 49-53.	0.3	4
58	Prenatal exome sequencing in fetuses with congenital heart defects. Clinical Genetics, 2020, 98, 215-230.	2.0	23
59	Genetic testing for kidney disease of unknown etiology. Kidney International, 2020, 98, 590-600.	5.2	46
60	Prenatal testing. , 2020, , 201-221.		0
61	Clinical application of medical exome sequencing for prenatal diagnosis of fetal structural anomalies. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 251, 119-124.	1.1	24
62	Prenatal exome sequencing in 65 fetuses with abnormality of the corpus callosum: contribution to further diagnostic delineation. Genetics in Medicine, 2020, 22, 1887-1891.	2.4	26
64	Molecular Diagnostics and In Utero Therapeutics for Orofacial Clefts. Journal of Dental Research, 2020, 99, 1221-1227.	5.2	8
65	A homozygous variant in growth and differentiation factor 2 <i>(</i> <scp><i>GDF2</i></scp> <i>)</i> may cause lymphatic dysplasia with hydrothorax and nonimmune hydrops fetalis. American Journal of Medical Genetics, Part A, 2020, 182, 2152-2160.	1.2	8
66	Comparison of Multiple Displacement Amplification (MDA) and Multiple Annealing and Looping-Based Amplification Cycles (MALBAC) in Limited DNA Sequencing Based on Tube and Droplet. Micromachines, 2020, 11, 645.	2.9	18
67	A prospective study on rapid exome sequencing as a diagnostic test for multiple congenital anomalies on fetal ultrasound. Prenatal Diagnosis, 2020, 40, 1300-1309.	2.3	36
68	Comprehensive clinically oriented workflow for nucleotide level resolution and interpretation in prenatal diagnosis of de novo apparently balanced chromosomal translocations in their genomic landscape. Human Genetics, 2020, 139, 531-543.	3.8	9
69	Genetic Examination for Fetuses with Increased Fetal Nuchal Translucency by Genomic Technology. Cytogenetic and Genome Research, 2020, 160, 57-62.	1.1	22
70	Genetic diagnosis in the fetus. Journal of Perinatology, 2020, 40, 997-1006.	2.0	10
71	Maternity health care professionals' views and experiences of fetal genomic uncertainty: A review. Prenatal Diagnosis, 2020, 40, 652-660.	2.3	6
72	Dietary modification, penetrance, and the origins of congenital malformation. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 5097-5099.	7.1	1
73	Increased nuchal translucency: diagnostic value of RASopathyâ€disorder testing. Ultrasound in Obstetrics and Gynecology, 2020, 55, 423-424.	1.7	1
74	Reply. Ultrasound in Obstetrics and Gynecology, 2020, 55, 424-425.	1.7	1

		CITATION REPORT	
#	Article	IF	CITATIONS
75	Evolution of a prenatal genetic clinic—A 10â€year cohort study. Prenatal Diagnosis, 2020, 40, 618	8-625. 2.3	12
76	Next-generation sequencing in prenatal setting: Some examples of unexpected variant association. European Journal of Medical Genetics, 2020, 63, 103875.	1.3	10
77	An approach to integrating exome sequencing for fetal structural anomalies into clinical practice. Genetics in Medicine, 2020, 22, 954-961.	2.4	49
78	The prevalence of genetic diagnoses in fetuses with severe congenital heart defects. Genetics in Medicine, 2020, 22, 1206-1214.	2.4	48
79	De novo damaging variants associated with congenital heart diseases contribute to the connectom Scientific Reports, 2020, 10, 7046.	ie. 3.3	34
80	Nonimmune hydrops fetalis: Genetic analysis and clinical outcome. Prenatal Diagnosis, 2020, 40, 803-812.	2.3	17
81	Dual independent genetic etiologies in a lethal complex malformation phenotype. Ultraschall in Der Medizin, 2020, 41, 112-114.	. 1.5	1
82	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses w congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	vith 2.3	49
83	Implementation of exome sequencing in fetal diagnostics—Data and experiences from a tertiary c in Denmark. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 783-790.	enter 2.8	33
84	Noninvasive prenatal diagnosis in a family at risk for Fraser syndrome. Prenatal Diagnosis, 2020, 40, 905-908.	, 2.3	4
85	Genetic innovations and our understanding of stillbirth. Human Genetics, 2020, 139, 1161-1172.	3.8	18
86	<scp>COngenital</scp> heart disease and the Diagnostic yield with Exome sequencing (<scp>CODE</scp>) study: prospective cohort study and systematic review. Ultrasound in Obstetri and Gynecology, 2021, 57, 43-51.	ics 1.7	46
87	Diagnostic and perinatal outcomes in consanguineous couples with a structural fetal anomaly: A cohort study. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 418-424.	2.8	5
88	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	2.3	18
89	Prenatal and postnatal diagnosis of <scp>Schuursâ€Hoeijmakers</scp> syndrome: Case series and of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 384-389.	review 1.2	15
90	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. Journal of Medical Genetics, 2021, 58, 400-413.	, 3.2	18
91	The role of nextâ€generation sequencing in the investigation of ultrasoundâ€identified fetal structu anomalies. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, 420-429.	ural 2.3	23
92	Earlier detection of hypochondroplasia: A large singleâ€center <scp>UK</scp> case series and systematic review. American Journal of Medical Genetics, Part A, 2021, 185, 73-82.	1.2	3

#	Article	IF	CITATIONS
93	A report on the impact of rapid prenatal exome sequencing on the clinical management of 52 ongoing pregnancies: a retrospective review. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, 1012-1019.	2.3	20
94	The potential diagnostic yield of whole exome sequencing in pregnancies complicated by fetal ultrasound anomalies. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 1106-1115.	2.8	21
95	Delivery technologies for in utero gene therapy. Advanced Drug Delivery Reviews, 2021, 169, 51-62.	13.7	24
96	Impact of prenatal exome sequencing for fetal genetic diagnosis on maternal psychological outcomes and decisional conflict in a prospective cohort. Genetics in Medicine, 2021, 23, 713-719.	2.4	18
97	Postâ€mortem confirmation of fetal brain abnormalities: challenges highlighted by the MERIDIAN cohort study. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, 1174-1182.	2.3	4
98	Risk of Clinically Significant Chromosomal Microarray Analysis Findings in Fetuses With Nuchal Translucency From 3.0 mm Through 3.4 mm. Obstetrics and Gynecology, 2021, 137, 126-131.	2.4	12
99	Selective serotonin reuptake inhibitor or serotoninâ€norepinephrine reuptake inhibitors and epidemiological characteristics associated with prenatal diagnosis of congenital heart disease. Prenatal Diagnosis, 2021, 41, 35-42.	2.3	5
100	Exome sequencing analysis on products of conception: a cohort study to evaluate clinical utility and genetic etiology for pregnancy loss. Genetics in Medicine, 2021, 23, 435-442.	2.4	27
101	Correspondence on "The prevalence of genetic diagnoses in fetuses with severe congenital heart defects―by Nisselrooij et al Genetics in Medicine, 2021, 23, 234-235.	2.4	2
102	Improved noninvasive fetal variant calling using standardized benchmarking approaches. Computational and Structural Biotechnology Journal, 2021, 19, 509-517.	4.1	1
103	Beyond diagnostic yield: prenatal exome sequencing results in maternal, neonatal, and familial clinical management changes. Genetics in Medicine, 2021, 23, 909-917.	2.4	21
104	Role of whole exome sequencing for unidentified genetic syndromes. Current Opinion in Obstetrics and Gynecology, 2021, 33, 112-122.	2.0	10
105	The role of chromosomal microarray and exome sequencing in prenatal diagnosis. Current Opinion in Obstetrics and Gynecology, 2021, 33, 148-155.	2.0	8
106	Prenatal Diagnosis of Genetic Disorders by DNA Profiling. , 2021, , 1-20.		0
107	Genomic disorders in the genomics era. , 2021, , 35-59.		1
108	Prenatal Exome Sequencing: Background, Current Practice and Future Perspectives—A Systematic Review. Diagnostics, 2021, 11, 224.	2.6	16
109	Evidence to Support the Clinical Utility of Prenatal Exome Sequencing in Evaluation of the Fetus with Congenital Anomalies. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, e39-e50.	2.3	23
110	Comparison between Two-Dimensional and Three-Dimensional Assessments of the Fetal Corpus Callosum: Reproducibility of Measurements and Acquisition Time. Journal of Pediatric Neurology, 2021, 19, 312-320.	0.2	2

#	Article	IF	Citations
111	Mucopolysaccharidosis type VII as a cause of recurrent Non-Immune Hydrops Fetalis: The first Tunisian case confirmed by Next-Generation Sequencing. Clinica Chimica Acta, 2021, 513, 68-70.	1.1	2
112	The Diagnostic Yield of Prenatal Genetic Technologies in Congenital Heart Disease: A Prospective Cohort Study. Fetal Diagnosis and Therapy, 2021, 48, 112-119.	1.4	10
113	Simpson-Golabi-Behmel-Syndrome in Dichorionic-Diamniotic Twin Pregnancy. Clinics and Practice, 2021, 11, 75-80.	1.4	1
114	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. European Journal of Medical Genetics, 2021, 64, 104124.	1.3	6
115	Diagnostic and clinical utility of nextâ€generation sequencing in children born with multiple congenital anomalies in the China neonatal genomes project. Human Mutation, 2021, 42, 434-444.	2.5	15
116	Genetic testing for unexplained perinatal disorders. Current Opinion in Pediatrics, 2021, 33, 195-202.	2.0	15
117	Bi-allelic loss of function variants in SLC30A5 as cause of perinatal lethal cardiomyopathy. European Journal of Human Genetics, 2021, 29, 808-815.	2.8	9
118	Case Report: Prenatal Whole-Exome Sequencing to Identify a Novel Heterozygous Synonymous Variant in NIPBL in a Fetus With Cornelia de Lange Syndrome. Frontiers in Genetics, 2021, 12, 628890.	2.3	3
119	High diagnosis rate for nonimmune hydrops fetalis with prenatal clinical exome from the Hydrops-Yielding Diagnostic Results of Prenatal Sequencing (HYDROPS) Study. Genetics in Medicine, 2021, 23, 1325-1333.	2.4	22
120	Dealing with uncertain results from chromosomal microarray and exome sequencing in the prenatal setting: An international crossâ€sectional study with healthcare professionals. Prenatal Diagnosis, 2021, 41, 720-732.	2.3	13
121	Whole Genome Sequencing in the Evaluation of Fetal Structural Anomalies: A Parallel Test with Chromosomal Microarray Plus Whole Exome Sequencing. Genes, 2021, 12, 376.	2.4	36
122	First prenatal case of Noonan syndrome with SOS2 mutation: Implications of early diagnosis for genetic counseling. American Journal of Medical Genetics, Part A, 2021, 185, 1897-1902.	1.2	1
123	Prenatal evaluation of fetuses with structural anomalies- is it time to shift from microarray to genome sequencing?. Taiwanese Journal of Obstetrics and Gynecology, 2021, 60, 389.	1.3	0
124	Non invasive prenatal testing (NIPT) for common aneuploidies and beyond. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 258, 424-429.	1.1	27
125	Chances and Challenges of New Genetic Screening Technologies (NIPT) in Prenatal Medicine from a Clinical Perspective: A Narrative Review. Genes, 2021, 12, 501.	2.4	37
126	The diagnostic efficacy of exome data analysis using fixed neurodevelopmental gene lists: Implications for prenatal setting. Prenatal Diagnosis, 2021, 41, 701-707.	2.3	3
129	Janus-faced EPHB4-associated disorders: novel pathogenic variants and unreported intrafamilial overlapping phenotypes. Genetics in Medicine, 2021, 23, 1315-1324.	2.4	6
131	Practice patterns of prenatal and perinatal testing in Canadian cytogenetics laboratories. Prenatal Diagnosis, 2021, 41, 843-854.	2.3	1

#	Article	IF	CITATIONS
132	Value of Exome Sequencing in Diagnosis and Management of Recurrent Non-immune Hydrops Fetalis: A Retrospective Analysis. Frontiers in Genetics, 2021, 12, 616392.	2.3	13
133	Beyond Baby Siblings—Expanding the Definition of "High-Risk Infants―in Autism Research. Current Psychiatry Reports, 2021, 23, 34.	4.5	8
135	Assessment of HCMV-encoded microRNAs in plasma as potential biomarkers in pregnant women with adverse pregnancy outcomes. Annals of Translational Medicine, 2021, 9, 638-638.	1.7	5
138	Fetal hydrops and the Incremental yield of Nextâ€generation sequencing over standard prenatal Diagnostic testing (<scp>FIND</scp>) study: prospective cohort study and metaâ€analysis. Ultrasound in Obstetrics and Gynecology, 2021, 58, 509-518.	1.7	27
139	The prenatal exome – a door to prenatal diagnostics?. Expert Review of Molecular Diagnostics, 2021, 21, 465-474.	3.1	7
141	Fuzzy model based on local injection of MTX combined with traditional Chinese medicine guided by ultrasound intervention to treat CSP. Journal of Intelligent and Fuzzy Systems, 2021, , 1-11.	1.4	1
142	Schuurs–Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	2.4	13
143	Combined exome sequencing and deep phenotyping in highly selected fetuses with skeletal dysplasia during the first and second trimesters improves diagnostic yield. Prenatal Diagnosis, 2021, 41, 1401-1413.	2.3	8
144	Postmortem whole-genome sequencing on a dried blood spot identifies a novel homozygous SUOX variant causing isolated sulfite oxidase deficiency. Journal of Physical Education and Sports Management, 2021, 7, a006091.	1.2	3
145	Preference for secondary findings in prenatal and pediatric exome sequencing. Prenatal Diagnosis, 2022, 42, 753-761.	2.3	11
146	Diagnostic yield of rare skeletal dysplasia conditions in the radiogenomics era. BMC Medical Genomics, 2021, 14, 148.	1.5	7
147	Molecular diagnosis for 55 fetuses with skeletal dysplasias by wholeâ€exome sequencing: A retrospective cohort study. Clinical Genetics, 2021, 100, 219-226.	2.0	13
148	Prenatal phenotype of Kabuki syndrome: A case series and literature review. Prenatal Diagnosis, 2021, 41, 1089-1100.	2.3	6
149	How genomics is changing the practice of prenatal testing. Journal of Perinatal Medicine, 2021, 49, 1003-1010.	1.4	5
150	The many etiologies of nonimmune hydrops fetalis diagnosed by exome sequencing. Prenatal Diagnosis, 2022, 42, 881-889.	2.3	11
151	The Added Value of Whole-Exome Sequencing for Anomalous Fetuses With Detailed Prenatal Ultrasound and Postnatal Phenotype. Frontiers in Genetics, 2021, 12, 627204.	2.3	16
152	Prenatal diagnosis of genetic aberrations in fetuses with short femur detected by ultrasound: A prospective cohort study. Prenatal Diagnosis, 2021, 41, 1153-1163.	2.3	6
153	Extended genetic testing in fetuses with sonographic skeletal system abnormalities. Ultrasound in Obstetrics and Gynecology, 2022, 59, 660-667.	1.7	9

#	Article	IF	CITATIONS
154	A Novel Multi-Exon Deletion of PACS1 in a Three-Generation Pedigree: Supplements to PACS1 Neurodevelopmental Disorder Spectrum. Frontiers in Genetics, 2021, 12, 690216.	2.3	9
155	Exome sequencing vs targeted gene panels for the evaluation of nonimmune hydrops fetalis. American Journal of Obstetrics and Gynecology, 2022, 226, 128.e1-128.e11.	1.3	14
156	Diagnosis and classification prediction model of pituitary tumor based on machine learning. Neural Computing and Applications, 2022, 34, 9257-9272.	5.6	3
157	Diagnostic yield of nextâ€generation sequencing in fetuses with isolated increased nuchal translucency: systematic review and metaâ€analysis. Ultrasound in Obstetrics and Gynecology, 2022, 59, 26-32.	1.7	14
158	The fetus in the age of the genome. Human Genetics, 2022, 141, 1017-1026.	3.8	3
159	Comprehensive non-invasive prenatal screening for pregnancies with elevated risks of genetic disorders: protocol for a prospective, multicentre study. BMJ Open, 2021, 11, e053617.	1.9	3
160	Utility of fetal whole exome sequencing in the etiological evaluation and outcome of nonimmune hydrops fetalis. Prenatal Diagnosis, 2021, 41, 1414-1424.	2.3	7
161	Preâ€natal and postâ€natal diagnosis of congenital upper limb differences: The first 3 years of the Australian Hand Difference Register. Journal of Paediatrics and Child Health, 2021, , .	0.8	0
162	Increased nuchal translucency after lowâ€risk noninvasive prenatal testing: What should we tell prospective parents?. Prenatal Diagnosis, 2021, 41, 1305-1315.	2.3	10
163	The current state of prenatal detection of genetic conditions in congenital heart defects. Translational Pediatrics, 2021, 10, 2157-2170.	1.2	6
164	Genetic Testing for Heritable Cardiovascular Diseases in Pediatric Patients: A Scientific Statement From the American Heart Association. Circulation Genomic and Precision Medicine, 2021, 14, e000086.	3.6	43
165	Trio-Based Low-Pass Genome Sequencing Reveals Characteristics and Significance of Rare Copy Number Variants in Prenatal Diagnosis. Frontiers in Genetics, 2021, 12, 742325.	2.3	9
166	Whole Exome Sequencing Analysis in Fetal Skeletal Dysplasia Detected by Ultrasonography: An Analysis of 38 Cases. Frontiers in Genetics, 2021, 12, 728544.	2.3	15
167	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	2.0	7
168	Exosomes—a potential indicator and mediator of cleft lip and palate: a narrative review. Annals of Translational Medicine, 2021, 9, 1485-1485.	1.7	3
169	The Current State and Future of Fetal Therapies. Clinical Obstetrics and Gynecology, 2021, Publish Ahead of Print, 926-932.	1.1	0
170	A twoâ€year prospective study assessing the performance of fetal chromosomal microarray analysis and nextâ€generation sequencing in highâ€risk pregnancies. Molecular Genetics & Genomic Medicine, 2021, 9, e1787.	1.2	2
171	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. Human Genetics, 2022, 141, 1269-1278.	3.8	10

#	Article	IF	CITATIONS
172	Whole-exome sequencing increases the diagnostic rate for prenatal fetal structural anomalies. European Journal of Medical Genetics, 2021, 64, 104288.	1.3	12
173	Genetic counseling for congenital heart disease $\hat{a} \in \hat{P}$ Practice resource of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2022, 31, 9-33.	1.6	9
174	Gestational exposure to perfluoroalkyl substances and congenital heart defects: A nested case-control pilot study. Environment International, 2021, 154, 106567.	10.0	19
175	Fetal exome sequencing for isolated increased nuchal translucency: should we be doing it?. BJOG: an International Journal of Obstetrics and Gynaecology, 2022, 129, 52-61.	2.3	32
176	Antenatal counselling for prospective parents whose fetus has a neurological anomaly: part 2, risks of adverse outcome in common anomalies. Developmental Medicine and Child Neurology, 2021, , .	2.1	8
177	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2022, 59, 931-937.	3.2	13
178	Prenatal genetic diagnosis of omphalocele by karyotyping, chromosomal microarray analysis and exome sequencing. Annals of Medicine, 2021, 53, 1286-1292.	3.8	8
179	Prenatal diagnosis of skeletal dysplasias using whole exome sequencing in China. Clinica Chimica Acta, 2020, 507, 187-193.	1.1	17
180	Human and mouse essentiality screens as a resource for disease gene discovery. Nature Communications, 2020, 11, 655.	12.8	64
185	Exome Sequencing for Prenatal Detection of Genetic Abnormalities in Fetal Ultrasound Anomalies: An Economic Evaluation. Fetal Diagnosis and Therapy, 2020, 47, 554-564.	1.4	11
186	Dual Myelomeningoceles in Twins: Case Report, Review, and Insights for Etiology. Pediatric Neurosurgery, 2020, 55, 363-373.	0.7	3
187	The Fetal Phenotype of Noonan Syndrome Caused by Severe, Cancer-Related PTPN11 Variants. American Journal of Case Reports, 2020, 21, e922468.	0.8	3
188	Prenatal exome sequencing: A useful tool for the fetal neurologist. Clinical Genetics, 2022, 101, 65-77.	2.0	14
189	An Introduction: Prenatal Screening, Diagnosis, and Treatment of Single Gene Disorders. Clinical Obstetrics and Gynecology, 2021, 64, 852-860.	1.1	2
190	Isolated Increased Nuchal Translucency in First Trimester Ultrasound Scan: Diagnostic Yield of Prenatal Microarray and Outcome of Pregnancy. Frontiers in Medicine, 2021, 8, 737936.	2.6	8
192	The Promise of Whole-exome Sequencing for Prenatal Genetic Diagnosis. Current Pharmacogenomics and Personalized Medicine, 2020, 17, 25-31.	0.2	0
193	Exome Sequencing and Its Emerging Role in Prenatal Genetic Diagnosis. Obstetrical and Gynecological Survey, 2020, 75, 317-320.	0.4	7
194	Nouvelles techniques génétiques de dépistage et diagnostic anténatals : quels enjeux ?. Périnatalité 2020, 12, 63-69.	'0 . 2	0

#	Article	IF	CITATIONS
198	Deciphering congenital anomalies for the next generation. Journal of Physical Education and Sports Management, 2020, 6, a005504.	1.2	5
199	Embryonic lethal genetic variants and chromosomally normal pregnancy loss. Fertility and Sterility, 2021, 116, 1351-1358.	1.0	5
200	Fetal Anomaly and Genetic Counseling. Comprehensive Gynecology and Obstetrics, 2021, , 219-229.	0.0	0
201	Whole-exome sequencing: A changing landscape of prenatal counseling. , 2022, , 39-67.		0
202	Early Sonographic Findings for Suspecting de novo Single-gene Mutation. Donald School Journal of Ultrasound in Obstetrics and Gynecology, 2020, 14, 125-130.	0.3	1
204	Amniotic fluid stabilized lipid nanoparticles for in utero intra-amniotic mRNA delivery. Journal of Controlled Release, 2022, 341, 616-633.	9.9	29
206	Developing and testing an algorithm for automatic segmentation of the fetal face from three-dimensional ultrasound images. Royal Society Open Science, 2020, 7, 201342.	2.4	7
207	Utilization of Whole Exome Sequencing in Lethal Form of Multiple Pterygium Syndrome: Identification of Mutations in Embryonal Subunit of Acetylcholine Receptor. International Journal of Molecular and Cellular Medicine, 2019, 8, 258-269.	1.1	2
208	Benefits of the incorporation of genomic medicine in clinical practice. , 2022, , 271-308.		0
209	Expansion of use of genome analyses and sequencing in diagnosis of genetic diseases. , 2022, , 65-93.		0
210	Fetal Cardiac Intervention. , 2021, , 103-118.		0
211	Next-Generation Sequencing Gene Panels and "Solo―Clinical Exome Sequencing Applied in Structurally Abnormal Fetuses. Fetal Diagnosis and Therapy, 2021, 48, 746-756.	1.4	6
212	Implementation of fetal clinical exome sequencing: Comparing prospective and retrospective cohorts. Genetics in Medicine, 2022, 24, 344-363.	2.4	13
213	Prenatal exome sequencing and chromosomal microarray analysis in fetal structural anomalies in a highly consanguineous population reveals a propensity of ciliopathy genes causing multisystem phenotypes. Human Genetics, 2022, 141, 101-126.	3.8	6
214	Clinical efficiency of simultaneous CNV-seq and whole-exome sequencing for testing fetal structural anomalies. Journal of Translational Medicine, 2022, 20, 10.	4.4	13
216	Challenges in variant interpretation in prenatal exome sequencing. European Journal of Medical Genetics, 2022, 65, 104410.	1.3	3
217	The prevalence of prenatal sonographic findings in postnatal diagnostic exome sequencing performed for neurocognitive phenotypes: A cohort study. Prenatal Diagnosis, 2022, , .	2.3	4
218	Diagnostic yield of exome sequencing in fetuses with multisystem malformations: systematic review and metaâ€analysis. Ultrasound in Obstetrics and Gynecology, 2022, 59, 715-722.	1.7	19

#	Article	IF	Citations
219	Singleton exome sequencing of 90 fetuses with ultrasound anomalies revealing novel disease-causing variants and genotype–phenotype correlations. European Journal of Human Genetics, 2022, 30, 428-438.	2.8	6
220	Assessing women's preferences towards tests that may reveal uncertain results from prenatal genomic testing: Development of attributes for a discrete choice experiment, using a mixed-methods design. PLoS ONE, 2022, 17, e0261898.	2.5	4
221	Prenatal exome sequencing in fetuses with callosal anomalies. Prenatal Diagnosis, 2022, 42, 744-752.	2.3	10
222	Diagnostic yield of genome sequencing for prenatal diagnosis of fetal structural anomalies. Prenatal Diagnosis, 2022, 42, 822-830.	2.3	12
223	Prenatal phenotype of 22q11 micro-duplications: A systematic review and report on 12 new cases. European Journal of Medical Genetics, 2022, 65, 104422.	1.3	3
224	How can prenatal exome sequencing inform future pregnancies?. American Journal of Obstetrics and Gynecology, 2022, 227, 98-99.	1.3	1
225	Parental motivations for and adaptation to trioâ€exome sequencing in a prospective prenatal testing cohort: Beyond the diagnosis. Prenatal Diagnosis, 2022, 42, 775-782.	2.3	8
226	Optimising Exome Prenatal Sequencing Services (EXPRESS): a study protocol to evaluate rapid prenatal exome sequencing in the NHS Genomic Medicine Service. NIHR Open Research, 0, 2, 10.	0.0	1
227	Retrospective identification of prenatal fetal anomalies associated with diagnostic neonatal genomic sequencing results. Prenatal Diagnosis, 2022, 42, 705-716.	2.3	2
228	Evolving fetal phenotypes and clinical impact of progressive prenatal exome sequencing pathways: cohort study. Ultrasound in Obstetrics and Gynecology, 2022, 59, 723-730.	1.7	23
229	High rate of abnormal findings in Prenatal Exome Trio in low risk pregnancies and apparently normal fetuses. Prenatal Diagnosis, 2022, 42, 725-735.	2.3	10
231	Molecular Approaches in Fetal Malformations, Dynamic Anomalies and Soft Markers: Diagnostic Rates and Challenges—Systematic Review of the Literature and Meta-Analysis. Diagnostics, 2022, 12, 575.	2.6	11
232	A 6.3ÂMb maternally derived microduplication of 20p13p12.2 in a fetus with Brachydactyly type D and related literature review. Molecular Cytogenetics, 2022, 15, 6.	0.9	2
233	Identification of novel heterozygous missense variant in the <i>COL11A1</i> causing fetal craniofacial anomalies. International Journal of Transgender Health, 2022, 15, 240-246.	2.3	0
234	Diagnostic yield of exome sequencing for prenatal diagnosis of fetal structural anomalies: A systematic review and metaâ€analysis. Prenatal Diagnosis, 2022, 42, 662-685.	2.3	75
235	A case of Costello syndrome diagnosed by trio whole exome sequencing. Journal of Obstetrics and Gynaecology, 2022, , 1-4.	0.9	0
236	Pathologic whole exome sequencing analysis in fetuses with minor sonographic abnormal findings and normal chromosomal microarray analysis: case series. Journal of Maternal-Fetal and Neonatal Medicine, 2022, , 1-6.	1.5	0
237	Case Report: Prenatal Diagnosis of a Novel Variant c.251dupT (p.N87Kfs*6) in BCOR Resulting in Oculofaciocardiodental Syndrome Using Whole-Exome Sequencing. Frontiers in Genetics, 2022, 13, 829613	2.3	Ο

ARTICLE IF CITATIONS # Exome sequencing as firstâ€tier test for fetuses with severe central nervous system structural 238 1.7 24 anomalies. Ultrasound in Obstetrics and Gynecology, 2022, 60, 59-67. Beyond AOPs: A Mechanistic Evaluation of NAMs in DART Testing. Frontiers in Toxicology, 2022, 4, 3.1 838466. Implementing a rapid fetal exome sequencing service: What do parents and health professionals think?. 240 2.35 Prenatal Diagnosis, 2022, 42, 783-795. Case Report: A Novel Mutation Identified in CHST14 Gene in a Fetus With Structural Abnormalities. 241 Frontiers in Genetics, 2022, 13, 853907. Parental mosaicism for apparent de novo genetic variants: Scope, detection, and counseling 242 2.3 8 challenges. Prenatal Diagnosis, 2022, 42, 811-821. In Vitro Fertilization Using Preimplantation Genetic Testing in a Romanian Couple Carrier of Mutations in the TTN Gene: A Case Report and Literature Review. Diagnostics, 2021, 11, 2328. 244 2.6 245 Trio exome sequencing is highly relevant in prenatal diagnostics. Prenatal Diagnosis, 2022, 42, 845-851. 2.3 18 Reappraising the Value of Fetal First-Trimester Ultrasonography. Maternal-Fetal Medicine, 2021, 246 0.8 Publish Ahead of Print, . 247 Prenatal Diagnosis of Genetic Disorders by DNA Profiling., 2022, , 625-644. 0 Exploring the diagnostic utility of genome sequencing for fetal congenital heart defects. Prenatal 248 2.3 Diagnosis, 2022, 42, 862-872 A single center experience in 90 cases with nonimmune hydrops fetalis: diagnostic categories â€' mostly 249 1.4 1 aneuploidy and still often idiopathic. Journal of Perinatal Medicine, 2022, 50, 985-992. Diagnostic yield of whole exome data in fetuses aborted for conotruncal malformations. Prenatal 2.3 Diagnosis, 2022, 42, 852-861. Fetal central nervous system anomalies: When should we offer exome sequencing?. Prenatal 251 2.3 16 Diagnosis, 2022, 42, 736-743. Genetic and Clinical Features of Heterotaxy in a Prenatal Cohort. Frontiers in Genetics, 2022, 13, 2.3 818241. Emerging technologies for prenatal diagnosis: The application of whole genome and RNA sequencing. 253 2.36 Prenatal Diagnosis, 2022, 42, 686-696. Implementation of Exome Sequencing in Prenatal Diagnosis and Impact on Genetic Counseling: The 2.4 Polish Experience. Genes, 2022, 13, 724. Prenatal Genetic Testing and Screening: A Focused Review. Seminars in Pediatric Neurology, 2022, 42, 258 2.05 100976. Factors that impact on women's decisionâ€making around prenatal genomic tests: An international 2.3 discrete choice survey. Prenatal Diagnosis, 2022, 42, 934-946.

#	Article	IF	CITATIONS
260	Beyond diagnostic yield: use of exome sequencing in prenatal diagnosis. Ultrasound in Obstetrics and Gynecology, 2022, 59, 697-698.	1.7	0
261	Prenatal diagnosis of acrania/exencephaly/anencephaly sequence (AEAS): additional structural and genetic anomalies. Archives of Gynecology and Obstetrics, 2023, 307, 293-299.	1.7	3
262	Information is power: The experiences, attitudes and needs of individuals who chose to have prenatal genomic sequencing for fetal anomalies. Prenatal Diagnosis, 2022, 42, 947-954.	2.3	3
263	Lessons learnt from prenatal exome sequencing. Prenatal Diagnosis, 2022, 42, 831-844.	2.3	22
264	Genomic architecture of fetal central nervous system anomalies using whole-genome sequencing. Npj Genomic Medicine, 2022, 7, 31.	3.8	6
265	Prenatal case of RIT1 mutation associated Noonan syndrome by whole exome sequencing (WES) and review of the literature. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 535-538.	1.3	3
266	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. European Journal of Human Genetics, 2022, , .	2.8	1
267	A single center experience of prenatal parentâ€fetus trio exome sequencing for pregnancies with congenital anomalies. Prenatal Diagnosis, 2022, 42, 901-910.	2.3	4
268	Prenatal exomes and genomes – so much new and so much more to learn. Prenatal Diagnosis, 2022, 42, 659-661.	2.3	0
269	International Society for Prenatal Diagnosis Updated Position Statement on the use of genomeâ€wide sequencing for prenatal diagnosis. Prenatal Diagnosis, 2022, 42, 796-803.	2.3	37
270	Prenatal diagnosis for fetuses with isolated and nonâ€isolated congenital heart defects using chromosomal microarray and exome sequencing. Prenatal Diagnosis, 2022, 42, 873-880.	2.3	3
271	Family history is key to the interpretation of exome sequencing in the prenatal context: unexpected diagnosis of Basal Cell Nevus Syndrome. Prenatal Diagnosis, 2022, , .	2.3	1
272	Prenatal trio-based whole exome sequencing in fetuses with abnormalities of the skeletal system. Molecular Genetics and Genomics, 2022, 297, 1017-1026.	2.1	4
273	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
274	"Let's Just Wait Until She's Born― Temporal Factors That Shape Decision-Making for Prenatal Genom Sequencing Amongst Families Underrepresented in Genomic Research. Frontiers in Genetics, 2022, 13, .	nic 2.3	3
275	Case Report: Identification Pathogenic Abnormal Splicing of BBS1 Causing Bardet–Biedl Syndrome Type I (BBS1) due to Missense Mutation. Frontiers in Genetics, 0, 13, .	2.3	2
276	Chromosomal Microarray Analysis in Fetuses Detected with Isolated Cardiovascular Malformation: A Multicenter Study, Systematic Review of the Literature and Meta-Analysis. Diagnostics, 2022, 12, 1328.	2.6	2
278	Fetal Aberrant Right Subclavian Artery: Associated Anomalies, Genetic Etiology, and Postnatal Outcomes in a Retrospective Cohort Study. Frontiers in Pediatrics, 2022, 10, .	1.9	5

#	Article	IF	CITATIONS
279	The fetal sequencing consortium: The value of multidisciplinary dialog and collaboration. Prenatal Diagnosis, 2022, 42, 807-810.	2.3	10
280	Ongoing reanalysis of prenatal exome sequencing data leads to higher diagnostic yield. Ultrasound in Obstetrics and Gynecology, 2022, 59, 833-834.	1.7	0
281	Utility of trioâ€based prenatal exome sequencing incorporating spliceâ€site and mitochondrial genome assessment in pregnancies with fetal ultrasound anomalies: prospective cohort study. Ultrasound in Obstetrics and Gynecology, 2022, 60, 780-792.	1.7	3
282	The utility of pathologic examination and comprehensive phenotyping for accurate diagnosis with perinatal exome sequencing. Prenatal Diagnosis, 0, , .	2.3	0
283	Prenatal Detection of Novel Compound Heterozygous Splice Site Variants of the KIAA0825 Gene in a Fetus with Postaxial Polydactyly Type A. Genes, 2022, 13, 1230.	2.4	3
284	Advances in studying human gametogenesis and embryonic development in China. Biology of Reproduction, 0, , .	2.7	0
285	Prenatal diagnosis and pregnancy outcome of major structural anomalies detectable in the first trimester: A populationâ€based cohort study in the Netherlands. Paediatric and Perinatal Epidemiology, 2022, 36, 804-814.	1.7	5
286	Parental Hopes and Understandings of the Value of Prenatal Diagnostic Genomic Sequencing: A Qualitative Analysis. Frontiers in Genetics, 0, 13, .	2.3	3
287	Prenatal ultrasound finding of atypical genitalia: Counseling, genetic testing and outcomes. Prenatal Diagnosis, 0, , .	2.3	4
288	Optimising Exome Prenatal Sequencing Services (EXPRESS): a study protocol to evaluate rapid prenatal exome sequencing in the NHS Genomic Medicine Service. NIHR Open Research, 0, 2, 10.	0.0	1
289	Rapid exome sequencing in critically ill children impacts acute and long-term management of patients and their families: A retrospective regional evaluation. European Journal of Medical Genetics, 2022, 65, 104571.	1.3	5
290	Prenatal phenotyping: A community effort to enhance the Human Phenotype Ontology. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 231-242.	1.6	14
291	Case Report: Novel compound heterozygous variants in CHRNA1 gene leading to lethal multiple pterygium syndrome: A case report. Frontiers in Genetics, 0, 13, .	2.3	0
292	Cost-Effectiveness of Exome Sequencing versus Targeted Gene Panels for Prenatal Diagnosis of Fetal Effusions and Non-Immune Hydrops Fetalis. American Journal of Obstetrics & Gynecology MFM, 2022, 4, 100724.	2.6	2
293	Drug delivery technologies for fetal, neonatal, and maternal therapy. Advanced Drug Delivery Reviews, 2022, , 114523.	13.7	0
294	Familial Aggregation of a Novel Missense Variant of COL2A1 Gene Associated with Short Extremities: Case Report and Review of the Literature. Children, 2022, 9, 1229.	1.5	0
295	Prenatal exome and genome sequencing for fetal structural abnormalities. American Journal of Obstetrics and Gynecology, 2023, 228, 140-149.	1.3	6
296	Next Generation Sequencing after Invasive Prenatal Testing in Fetuses with Congenital Malformations: Prenatal or Neonatal Investigation. Genes, 2022, 13, 1517.	2.4	10

#	Article	IF	Citations
297	How to choose a test for prenatal genetic diagnosis: a practical overview. American Journal of Obstetrics and Gynecology, 2023, 228, 178-186.	1.3	2
298	Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. Nature Genetics, 2022, 54, 1320-1331.	21.4	155
299	Cardiovascular Anomalies among 1005 Fetuses Referred to Invasive Prenatal Testing—A Comprehensive Cohort Study of Associated Chromosomal Aberrations. International Journal of Environmental Research and Public Health, 2022, 19, 10019.	2.6	0
300	Prenatal Somatic Cell Gene Therapies: Charting a Path Toward Clinical Applications (Proceedings of) Tj ETQq1 1	0.784314 2.0	rg&T /Overlo
302	Impact of variation in practice in the prenatal reporting of variants of uncertain significance by commercial laboratories: Need for greater adherence to published guidelines. Prenatal Diagnosis, 2022, 42, 1514-1524.	2.3	5
303	Prenatal Diagnosis and Outcomes in Fetuses with Hemivertebra. Genes, 2022, 13, 1623.	2.4	Ο
304	Rapid genome sequencing for pediatrics. Human Mutation, 2022, 43, 1507-1518.	2.5	9
305	A Central Role of Telomere Dysfunction in the Formation of a Unique Translocation within the Sub-Telomere Region Resulting in Duplication and Partial Trisomy. Genes, 2022, 13, 1762.	2.4	1
306	Exome sequencing for structurally normal fetuses—yields and ethical issues. European Journal of Human Genetics, 2023, 31, 164-168.	2.8	3
307	Genetic abnormalities in fetal congenital heart disease with aberrant right subclavian artery. Scientific Reports, 2022, 12, .	3.3	0
308	Should we offer prenatal exome sequencing for intrauterine growth restriction or short long bones? A systematic review and meta-analysis. American Journal of Obstetrics and Gynecology, 2023, 228, 409-417.e4.	1.3	7
309	Application of exome sequencing for prenatal diagnosis of fetal structural anomalies: clinical experience and lessons learned from a cohort of 1618 fetuses. Genome Medicine, 2022, 14, .	8.2	15
311	International Society for Prenatal Diagnosis 2022 debate 3—Fetal genome sequencing should be offered to all pregnant patients. Prenatal Diagnosis, 2023, 43, 428-434.	2.3	2
312	Comment on: Disease gene identification strategies for exome sequencing by Gilissen et al. 2012. European Journal of Human Genetics, 2022, 30, 1100-1101.	2.8	0
313	Genetic examination for fetuses with increased nuchal translucency by exome sequencing. Journal of Obstetrics and Gynaecology Research, 0, , .	1.3	0
314	Facilitating variant curation sharing for fetal precision genomics: A new venture for prenatal diagnosis. Prenatal Diagnosis, 2022, 42, 1479-1480.	2.3	1
315	Prenatal isolated clubfoot increases the risk for clinically significant exome sequencing results. Prenatal Diagnosis, 2022, 42, 1622-1626.	2.3	1
317	Implementation of Public Funded Genome Sequencing in Evaluation of Fetal Structural Anomalies. Genes, 2022, 13, 2088.	2.4	3

#	Article	IF	Citations
318	Genetic analysis of 55 cases with fetal skeletal dysplasia. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	2
319	Fetal cerebral ventriculomegaly: What do we tell the prospective parents?. Prenatal Diagnosis, 2022, 42, 1674-1681.	2.3	7
320	Prenatal Genetic Testing in the Era of Next Generation Sequencing: A One-Center Canadian Experience. Genes, 2022, 13, 2019.	2.4	1
321	Case report and a brief review: Analysis and challenges of prenatal imaging phenotypes and genotypes in Joubert syndrome. Frontiers in Genetics, 0, 13, .	2.3	2
322	Prenatal diagnosis of ALPL gene mutations in recurrent fetal skeletal dysplasia. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 1065-1068.	1.3	1
323	Fetal Neurology: From Prenatal Counseling to Postnatal Follow-Up. Diagnostics, 2022, 12, 3083.	2.6	2
324	Evaluating Genetic Disorders in the Neonate: The Role of Exome Sequencing in the NICU. NeoReviews, 2022, 23, e829-e840.	0.8	1
325	Compound heterozygous splicing variants in <scp><i>KIAA0586</i></scp> cause fetal shortâ€rib thoracic dysplasia and cerebellar malformation: Use of exome sequencing in prenatal diagnosis. Molecular Genetics & Genomic Medicine, 2023, 11, .	1.2	2
326	Genetic causes of isolated and severe fetal growth restriction in normal chromosomal microarray analysis. International Journal of Gynecology and Obstetrics, 0, , .	2.3	0
327	Genetic aetiology distribution of 398 foetuses with congenital heart disease in the prenatal setting. ESC Heart Failure, 2023, 10, 917-930.	3.1	4
328	Application of Prenatal Whole Exome Sequencing for Structural Congenital Anomalies—Experience from a Local Prenatal Diagnostic Laboratory. Healthcare (Switzerland), 2022, 10, 2521.	2.0	1
329	Pathogenic/likely pathogenic copy number variations and regions of homozygosity in fetal central nervous system malformations. Archives of Gynecology and Obstetrics, 0, , .	1.7	1
330	Whole exome sequencing improves genetic diagnosis of fetal clubfoot. Human Genetics, 2023, 142, 407-418.	3.8	4
333	Prenatal Diagnosis of PPP2R1A-Related Neurodevelopmental Disorders Using Whole Exome Sequencing: Clinical Report and Review of Literature. Genes, 2023, 14, 126.	2.4	2
334	Allâ€inâ€one whole exome sequencing strategy with simultaneous copy number variant, single nucleotide variant and absenceâ€ofâ€heterozygosity analysis in fetuses with structural ultrasound anomalies: A 1â€year experience. Prenatal Diagnosis, 2023, 43, 527-543.	2.3	1
335	Diagnostic potential of the amniotic fluid cells transcriptome in deciphering mendelian disease: a proof-of-concept. Npj Genomic Medicine, 2022, 7, .	3.8	7
336	Genetics in prenatal diagnosis. Singapore Medical Journal, 2023, 64, 27.	0.6	2
337	Parental mosaicism detection and preimplantation genetic testing in families with multiple transmissions of de novo mutations. Journal of Medical Genetics, 2023, 60, 910-917.	3.2	2

#	Article	IF	CITATIONS
338	Genomic autopsy to identify underlying causes of pregnancy loss and perinatal death. Nature Medicine, 2023, 29, 180-189.	30.7	11
339	Exome sequencing in fetuses with short long bones detected by ultrasonography: A retrospective cohort study. Frontiers in Genetics, 0, 14, .	2.3	5
340	Whole exome sequencing in fetuses with isolated increased nuchal translucency: a systematic review and meta-analysis. Journal of Maternal-Fetal and Neonatal Medicine, 2023, 36, .	1.5	1
341	Identification of biâ€ellelic <i>LFNG</i> variants in three patients and further clinical and molecular refinement of spondylocostal dysostosis 3. Clinical Genetics, 2023, 104, 230-237.	2.0	0
342	Prenatal next-generation sequencing in the fetus with congenital malformations: how can we improve clinical utility?. American Journal of Obstetrics & Gynecology MFM, 2023, 5, 100923.	2.6	2
343	Prenatal diagnosis of euploid increased nuchal translucency on fetal ultrasound (I): Noonan syndrome: Prenatal diagnosis and genetic testing. Journal of Medical Ultrasound, 2022, 30, 257.	0.4	1
344	Diagnostic yield and psychological outcomes among women pursuing trioâ€exome sequencing: Do women with recurrent anomalous fetal phenotypes experience more negative psychological outcomes?. Prenatal Diagnosis, 0, , .	2.3	0
345	Fetal anomaly diagnosis and termination of pregnancy. Developmental Medicine and Child Neurology, 2023, 65, 900-907.	2.1	7
347	Piloting a multidisciplinary approach to improve outcomes of fetal whole exome sequencing: An overview of workflow and case example. Prenatal Diagnosis, 2023, 43, 544-552.	2.3	1
349	The human periconceptional maternal-embryonic space in health and disease. Physiological Reviews, 2023, 103, 1965-2038.	28.8	3
350	Whole-exome sequencing in deceased fetuses with ultrasound anomalies: a retrospective analysis. BMC Medical Genomics, 2023, 16, .	1.5	1
351	Diagnóstico genético prenatal de enfermedades monogénicas. Advances in Laboratory Medicine / Avances En Medicina De Laboratorio, 2023, 4, 40-51.	0.2	0
352	Association between aberrant amino acid metabolism and nonchromosomal modifications fetal structural anomalies: A cohort study. Frontiers in Endocrinology, 0, 14, .	3.5	1
353	Anwendung genetischer Untersuchungen in der PrÃ ¤ ataldiagnostik. Springer Reference Medizin, 2023, , 1-18.	0.0	0
354	Diagnostic yield of exome sequencing in isolated fetal growth restriction: Systematic review and metaâ€analysis. Prenatal Diagnosis, 2023, 43, 596-604.	2.3	4
355	Essential genes: a cross-species perspective. Mammalian Genome, 2023, 34, 357-363.	2.2	2
356	Whole genome sequencing vs chromosomal microarray analysis in prenatal diagnosis. American Journal of Obstetrics and Gynecology, 2023, 229, 302.e1-302.e18.	1.3	2
357	Prenatal genomic testing for ultrasoundâ€detected fetal structural anomalies. The Obstetrician and Gynaecologist, 2023, 25, 121-130.	0.4	1

#	Article	IF	CITATIONS
358	Genetic analysis and prenatal diagnosis of short-rib thoracic dysplasia 3 with or without polydactyly caused by compound heterozygous variants of DYNC2H1 gene in four Chinese families. Frontiers in Genetics, 0, 14, .	2.3	0
359	Prenatal genetic diagnosis of monogenic diseases. Advances in Laboratory Medicine / Avances En Medicina De Laboratorio, 2023, 4, 28-39.	0.2	0
361	Comparison of Genetic Profiles of Neonates in Intensive Care Units Conceived With or Without Assisted Reproductive Technology. JAMA Network Open, 2023, 6, e236537.	5.9	1
362	Invasive Procedures in the First Trimester. , 2023, , 457-473.		0
363	Screening for Fetal Chromosome Abnormalities. , 2023, , 139-150.		0
364	Molecular cytogenomics of human genetic disorders. , 2023, , 721-741.		0
365	Chromosomal microarrays and next-generation sequencing for diagnosis of fetal abnormalities. , 2023, , 767-787.		0
366	Genetics and genomics of recurrent pregnancy loss. , 2023, , 565-598.		0
367	Next-generation sequencing for gene panels, clinical exome, and whole-genome analysis. , 2023, , 743-766.		0
368	Prenatal diagnosis in the fetal hyperechogenic kidneys: assessment using chromosomal microarray analysis and exome sequencing. Human Genetics, 2023, 142, 835-847.	3.8	2
369	Prenatal exome sequencing analysis in fetuses with central nervous system anomalies. Ultrasound in Obstetrics and Gynecology, 2023, 62, 721-726.	1.7	2
370	The new frontier: a case for whole exome sequencing with multiple fetal anomalies. Case Reports in Perinatal Medicine, 2023, 12, .	0.1	0
371	Searching for a sense of closure: parental experiences of recontacting after a terminated pregnancy for congenital malformations. European Journal of Human Genetics, 0, , .	2.8	0
372	Clinical utility of chromosomal microarray analysis and whole exome sequencing in foetuses with oligohydramnios. Annals of Medicine, 2023, 55, .	3.8	0
373	Diagnostic Yield of Exome Sequencing in Fetuses with Sonographic Features of Skeletal Dysplasias but Normal Karyotype or Chromosomal Microarray Analysis: A Systematic Review. Genes, 2023, 14, 1203.	2.4	0
374	Gastrointestinal congenital anomalies requiring surgery: diagnosis, counselling, and management. The Obstetrician and Gynaecologist, 0, , .	0.4	0
375	Case Report: A prenatal diagnosis of osteogenesis imperfecta in a patient with a novel pathogenic variant in COL1A2. F1000Research, 0, 12, 603.	1.6	0
376	Diagnostic yield with exome sequencing in prenatal severe bilateral ventriculomegaly: a systematic review and meta-analysis. American Journal of Obstetrics & Gynecology MFM, 2023, 5, 101048.	2.6	1

#	Article	IF	Citations
377	Respiratory features of centronuclear myopathy in the Netherlands. Neuromuscular Disorders, 2023, 33, 580-588.	0.6	0
378	Association of deep phenotyping with diagnostic yield of prenatal exome sequencing for fetal brain abnormalities. Genetics in Medicine, 2023, , 100915.	2.4	0
379	In Utero Pediatrics in Maternal-Fetal Medicine. , 2023, , 21-28.		0
380	Identification and characterization of the largest deletion in the PCCA gene causing severe acute early-onset form of propionic acidemia. Molecular Genetics and Genomics, 2023, 298, 905-917.	2.1	1
381	Identifying the genetic causes of developmental disorders and intellectual disability in Africa: a systematic literature review. Frontiers in Genetics, 0, 14, .	2.3	2
382	Challenges and Pragmatic Solutions in Pre-Test and Post-Test Genetic Counseling for Prenatal Exome Sequencing. The Application of Clinical Genetics, 0, Volume 16, 89-97.	3.0	1
383	Noninvasive prenatal testing, ultrasonographic findings and poor prenatal diagnosis rates for twin pregnancies: a retrospective study. BMC Pregnancy and Childbirth, 2023, 23, .	2.4	0
384	Whole-exome sequencing applications in prenatal diagnosis of fetal bowel dilatation. Open Life Sciences, 2023, 18, .	1.4	0
386	Diagnostic yield of pediatric and prenatal exome sequencing in a diverse population. Npj Genomic Medicine, 2023, 8, .	3.8	7
388	Association analysis between chromosomal abnormalities and fetal ultrasonographic soft markers based on 15,263 fetuses. American Journal of Obstetrics & Gynecology MFM, 2023, 5, 101072.	2.6	3
391	Counseling in a Changing World of Genetics. , 2023, , 321-332.		0
392	Prenatal Genome-Wide Sequencing for the Investigation of Fetal Structural Anomalies: Is There a Role for Noninvasive Prenatal Diagnosis?. , 2023, , 357-377.		0
393	Prenatal whole exome sequencing identified two rare compound heterozygous variants in <i>EVC2</i> causing Ellisâ€van Creveld syndrome. Molecular Genetics & Genomic Medicine, 0, , .	1.2	0
394	Diagnostic yield of exome sequencing in prenatal agenesis of corpus callosum: systematic review and metaâ€analysis. Ultrasound in Obstetrics and Gynecology, 2024, 63, 312-320.	1.7	2
395	Spectrum of congenital anomalies of the kidney and urinary tract (CAKUT) including renal parenchymal malformations during fetal life and the implementation of prenatal exome sequencing (WES). Archives of Gynecology and Obstetrics, 0, , .	1.7	2
396	Human Exome Sequencing and Prospects for Predictive Medicine: Analysis of International Data and Own Experience. Journal of Personalized Medicine, 2023, 13, 1236.	2.5	2
397	Fetal hyperechoic kidney cohort study and a meta-analysis. Frontiers in Genetics, 0, 14, .	2.3	1
398	Systematic evaluation of genome sequencing for the diagnostic assessment of autism spectrum disorder and fetal structural anomalies. American Journal of Human Genetics, 2023, 110, 1454-1469.	6.2	7

		CITATION REPORT		
#	ARTICLE	2022 275 204	IF	CITATIONS
399	Whole-genome sequencing as a method of prenatal genetic diagnosis: Ethical issues. ,	2023, , 275-304.		0
401	When NIPT meets WES, prenatal diagnosticians face the dilemma: genetic etiological a cases of NT thickening and follow-up of pregnancy outcomes. Frontiers in Genetics, 0,		2.3	1
403	Genetic Spectrum of Congenital Anomalies of the Kidney and Urinary Tract in Chinese Genome Project. Kidney International Reports, 2023, , .	Newborn	0.8	0
404	Fetal congenital heart diseases: Diagnosis by anatomical scans, echocardiography and Annals of the Academy of Medicine, Singapore, 2023, 52, 420-431.	genetic tests.	0.4	0
405	The role of a multidisciplinary team in managing variants of uncertain clinical significar genetic diagnosis. European Journal of Medical Genetics, 2023, 66, 104844.	nce in prenatal	1.3	2
406	Incremental yield of wholeâ€genome sequencing over chromosomal microarray analys sequencing for congenital anomalies in prenatal period and infancy: systematic review metaâ€analysis. Ultrasound in Obstetrics and Gynecology, 2024, 63, 15-23.	is and exome and	1.7	1
407	Fetal Brain Imaging: A Composite Neural Network Approach for Keyframe Detection in Videos. , 2023, , .	Ultrasound		0
408	Chromosome Microarray Analysis and Exome Sequencing: Implementation in Prenatal Fetuses with Digestive System Malformations. Genes, 2023, 14, 1872.	Diagnosis of	2.4	0
409	Prenatal wholeâ€exome sequencing in fetuses with increased nuchal translucency. Mc & Genomic Medicine, 2023, 11, .	ecular Genetics	1.2	0
411	Case Report: A prenatal diagnosis of osteogenesis imperfecta in a patient with a novel variant in COL1A2. F1000Research, 0, 12, 603.	pathogenic	1.6	0
412	Uncovering the Genetic Basis of Congenital Heart Disease: Recent Advancements and Clinical Management. , 2023, 2, 464-480.	Implications for		0
413	Prenatal whole-exome sequencing for fetal structural anomalies: a retrospective analys Chinese cases. BMC Medical Genomics, 2023, 16, .	sis of 145	1.5	0
415	Prenatal diagnosis of fetal digestive system malformations and pregnancy outcomes a referral center in Fujian, China: A retrospective study. Heliyon, 2023, 9, e21546.	t a tertiary	3.2	0
416	Prenatal genetic diagnosis associated with fetal ventricular septal defect: an assessme chromosomal microarray analysis and exome sequencing. Frontiers in Genetics, 0, 14,		2.3	0
417	Towards personalized genome-scale modeling of inborn errors of metabolism for syste applications. Metabolism: Clinical and Experimental, 2024, 150, 155738.	ems medicine	3.4	0
418	Enhancing Fetal Anomaly Detection in Ultrasonography Images: A Review of Machine I Approaches. Biomimetics, 2023, 8, 519.	_earning-Based	3.3	0
419	The expansion of genomic precision medicine to prenatal care. , 2024, , 196-216.			0
420	Case Report: A prenatal diagnosis of osteogenesis imperfecta in a patient with a novel variant in COL1A2. F1000Research, 0, 12, 603.	pathogenic	1.6	0

#	Article	IF	CITATIONS
421	Diagnostic yield of prenatal exome sequencing in the genetic screening of fetuses with brain anomalies detected by <scp>MRI</scp> and ultrasonography: A systematic review and metaâ€analysis. BJOG: an International Journal of Obstetrics and Gynaecology, 0, , .	2.3	0
422	Molecular Diagnostic Yield of Exome Sequencing in Patients With Congenital Hydrocephalus. JAMA Network Open, 2023, 6, e2343384.	5.9	1
424	Clinical and molecular analysis of nine fetal cases with clinically significant variants causing nemaline myopathy. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2024, 292, 263-266.	1.1	1
425	Genomics of Stillbirth. Seminars in Perinatology, 2023, , 151866.	2.5	0
426	Prenatal Exome Sequencing Analysis in Fetuses with Various Ultrasound Findings. Journal of Clinical Medicine, 2024, 13, 181.	2.4	0
427	Lessons learned from rapid exome sequencing for 575 critically ill patients across the broad spectrum of rare disease. Frontiers in Genetics, 0, 14, .	2.3	1
428	Contribution of uniparental disomy to fetal growth restriction: a whole-exome sequencing series in a prenatal setting. Scientific Reports, 2024, 14, .	3.3	1
429	Contribution of genetic variants to congenital heart defects in both singleton and twin fetuses: a Chinese cohort study. Molecular Cytogenetics, 2024, 17, .	0.9	0
430	Multidisciplinary Workup for Stillbirth at a Tertiary-Care Hospital in Northeast Mexico: Findings, Challenges and Perspectives. Maternal and Child Health Journal, 2024, 28, 1072-1079.	1.5	0
432	Importance and application of WES in fetal genetic diagnostics: Identification of novel ASPM mutation in a fetus with microcephaly. Molecular Genetics and Metabolism Reports, 2024, 38, 101056.	1.1	0
433	Prospective prenatal cell-free DNA screening for genetic conditions of heterogenous etiologies. Nature Medicine, 2024, 30, 470-479.	30.7	0
434	De novo heterozygous missense variants in <i>CELSR1</i> as cause of fetal pleural effusions and progressive fetal hydrops. Journal of Medical Genetics, 0, , jmg-2023-109698.	3.2	0
435	Next-generation variant exon screening: Moving forward in routine genetic disease investigations. , 2024, 2, 101816.		0
436	Expanding the phenotypic spectrum of LIG4 pathogenic variations: neuro-histopathological description of 4 fetuses with stenosis of the aqueduct. European Journal of Human Genetics, 2024, 32, 545-549.	2.8	0
437	Ultrasound signs of fetal chromosomal abnormalities at 11–14 weeks of pregnancy and its value in the era of modern genetic testing. Ulʹtrazvukovaâ I Funkcionalʹnaâ Diagnostika, 2024, , 67-95.	0.0	0
438	Analysis of automatic news segmentation combining with conditional random field knowledge recognition algorithm. Signal, Image and Video Processing, 2024, 18, 3867-3875.	2.7	0
439	Prenatal Genetic Testing. , 2023, , 197-210.		0
440	Single-cell guided prenatal derivation of primary fetal epithelial organoids from human amniotic and tracheal fluids. Nature Medicine, 2024, 30, 875-887.	30.7	0

#	Article	IF	CITATIONS
441	Prenatal Genome-Wide Sequencing analysis (Exome or Genome) in detecting pathogenic Single Nucleotide Variants in fetal Central Nervous System Anomalies: systematic review and meta-analysis. European Journal of Human Genetics, 0, , .	2.8	0
442	Spotlight onâ \in antenatal genetic testing. The Obstetrician and Gynaecologist, 2024, 26, 64-65.	0.4	0
443	Prospective Investigation of Optical Genome Mapping for Prenatal Genetic Diagnosis. Clinical Chemistry, 0, , .	3.2	0