D-Z Li

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4589907/publications.pdf

Version: 2024-02-01

222	1,452	17 h-index	29
papers	citations		g-index
222	222	222	1533
all docs	docs citations	times ranked	citing authors

#	Article	IF	CITATIONS
1	A novel splicing mutation of <i>ARHGAP29</i> is associated with nonsyndromic cleft lip with or without cleft palate. Journal of Maternal-Fetal and Neonatal Medicine, 2022, 35, 2499-2506.	1.5	7
2	Can perinatal outcomes of fetal omphalocele be improved at a tertiary center in South China?. Journal of Maternal-Fetal and Neonatal Medicine, 2022, 35, 8409-8411.	1.5	1
3	Idiopathic polyhydramnios and foetal macrosomia in the absence of maternal diabetes: clinical vigilance for costello syndrome. Journal of Obstetrics and Gynaecology, 2022, 42, 704-706.	0.9	2
4	Fetal Pyruvate Kinase Deficiency Identified Incidentally in a Chinese Family at Risk for \hat{l}_{\pm} -Thalassemia. Indian Journal of Hematology and Blood Transfusion, 2022, 38, 424-426.	0.6	0
5	The Application of Crownâ€Chin Length to Crownâ€Rump Length Ratio in Predicting Fetal Skeletal Dysplasia at First Trimester. Journal of Ultrasound in Medicine, 2022, , .	1.7	O
6	Influence of fibroids on cellâ€free DNA screening accuracy: what we need to know. Ultrasound in Obstetrics and Gynecology, 2022, 59, 127-128.	1.7	0
7	Prenatal exome sequencing in fetuses with callosal anomalies. Prenatal Diagnosis, 2022, 42, 744-752.	2.3	10
8	Single nucleotide polymorphism–based cell-free DNA prenatal screening for 22q11.2 deletion syndrome. American Journal of Obstetrics and Gynecology, 2022, 227, 123-124.	1.3	0
9	Does chorionic villus sampling protect against fetal loss in twin pregnancy at high background risk of spontaneous miscarriage?. Ultrasound in Obstetrics and Gynecology, 2022, 59, 273-274.	1.7	O
10	How can cell-free DNA screening best be incorporated into current prenatal screening algorithm?. American Journal of Obstetrics and Gynecology, 2022, 227, 126-127.	1.3	0
11	The 16-week sonographic findings in fetuses with increased nuchal translucency and a normal array. Journal of Maternal-Fetal and Neonatal Medicine, 2022, , 1-5.	1.5	O
12	Contribution of maternal mosaicism to false-positive chromosome X loss associated with noninvasive prenatal testing. Journal of Maternal-Fetal and Neonatal Medicine, 2022, 35, 9647-9653.	1.5	4
13	Prenatal microcephaly: Exome sequencing aids rapid determination of causative etiologies. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, , .	1.1	1
14	Residual risk for clinically significant copy number variants in low-risk pregnancies with a normal noninvasive prenatal screening result: does it have clinical value?. American Journal of Obstetrics and Gynecology, 2022, 226, 599-600.	1.3	0
15	Detection of rare thalassemia mutations using long-read single-molecule real-time sequencing. Gene, 2022, 825, 146438.	2.2	19
16	Chorioamnionitis and risk of long-term neurodevelopmental impairment in offspring. American Journal of Obstetrics and Gynecology, 2022, , .	1.3	0
17	Identification of differential microRNAs and messenger RNAs resulting from ASXL transcriptional regulator 3 knockdown during during heart development. Bioengineered, 2022, 13, 9948-9961.	3.2	2
18	Case Report: Two Novel L1CAM Mutations in Two Unrelated Chinese Families With X-Linked Hydrocephalus. Frontiers in Genetics, 2022, 13, 810853.	2.3	0

#	Article	IF	CITATIONS
19	Congenital extrahepatic portosystemic shunt: An unusual feature in cardio-facio-cutaneous syndrome. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, 273, 107-108.	1.1	2
20	The Genetic and Clinical Outcomes in Fetuses With Isolated Fetal Growth Restriction: A Chinese Single-Center Retrospective Study. Frontiers in Genetics, 2022, 13, 856522.	2.3	4
21	Beyond diagnostic yield: use of exome sequencing in prenatal diagnosis. Ultrasound in Obstetrics and Gynecology, 2022, 59, 697-698.	1.7	0
22	Fetal phenotype of <scp>SLC35A2 DG</scp> : Enlarged cisterna magna on ultrasound. Congenital Anomalies (discontinued), 2022, 62, 217-219.	0.6	0
23	Prenatal diagnosis of Miller-Dieker syndrome/PAFAH1B1-related lissencephaly: Ultrasonography and genetically investigative results. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, 274, 28-32.	1.1	5
24	Ongoing reanalysis of prenatal exome sequencing data leads to higher diagnostic yield. Ultrasound in Obstetrics and Gynecology, 2022, 59, 833-834.	1.7	0
25	Early prenatal detection of triploidy: a 9-year experience in mainland China. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 4072-4076.	1.5	2
26	First-trimester detection of micrognathia as a presentation of mandibulofacial dysostosis with microcephaly. Journal of Obstetrics and Gynaecology, 2021, 41, 821-823.	0.9	2
27	First trimester prenatal detection of mosaic trisomy 8. Journal of Obstetrics and Gynaecology, 2021, 41, 484-486.	0.9	0
28	Chromosomal microarray analysis in pregnancies at risk for a molecular disorder. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 159-162.	1.5	2
29	Can cell-free DNA testing be used in pregnancies with isolated fetal omphalocele? Preliminary evidence from cytogenetic results of prenatal cases. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 624-628.	1.5	3
30	Compound heterozygous mutation of the ASXL3 gene causes autosomal recessive congenital heart disease. Human Genetics, 2021, 140, 333-348.	3.8	17
31	Prenatal phenotypic discordance in monozygotic twins due to a postzygotic <scp><i>TSC2</i></scp> variant. Prenatal Diagnosis, 2021, 41, 207-209.	2.3	0
32	Microarray analysis in fetuses with duodenal obstruction: It is not just trisomy 21. Prenatal Diagnosis, 2021, 41, 316-322.	2.3	10
33	The role of midtrimester soft markers for aneuploidy in the era of cell-free DNA screening. American Journal of Obstetrics and Gynecology, 2021, 224, 546-547.	1.3	0
34	Prenatal detection of $1p36$ deletion syndrome: ultrasound findings and microarray testing results. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 2180-2184.	1.5	2
35	Identification of thalassemia gene cluster deletion by longâ€read wholeâ€genome sequencing (LRâ€WGS). International Journal of Laboratory Hematology, 2021, 43, 859-865.	1.3	8
36	Unmasking a recessive allele by a deletion: Early prenatal diagnosis of <scp>Bardetâ€Biedl</scp> syndrome in a Chinese family. Congenital Anomalies (discontinued), 2021, 61, 138-139.	0.6	2

#	Article	IF	Citations
37	Mild α-Thalassemia Caused by a Mosaic α-Globin Gene Mutation. Hemoglobin, 2021, 45, 1-4.	0.8	O
38	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
39	A novel deletion of the major regulatory element flanking the αâ€globin gene cluster as a cause of α ⁰ â€thalassemia. International Journal of Laboratory Hematology, 2021, 43, O190-O192.	1.3	3
40	Micromelic upper limbs and cardiac defect: A fetal case of Holt-Oram syndrome identified in the first trimester. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 258, 471-473.	1.1	0
41	Prenatal diagnosis and postnatal management of congenital mesoblastic nephroma: Experience at a single center in China. Prenatal Diagnosis, 2021, 41, 766-771.	2.3	4
42	Germline mosaicism in a collagen <scp>VI</scp> â€related myopathy family: A cause of autosomal recessive inheritance. Congenital Anomalies (discontinued), 2021, 61, 197-198.	0.6	0
43	Fetal akinesia: The application of clinical exome sequencing in cases with decreased fetal movement. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 260, 59-63.	1.1	5
44	Sonographic detection of monochorionic monozygotic twins discordant for sex: Implications for prenatal genetic counseling. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 260, 236-238.	1.1	0
45	A novel SPTB frameshift deletion causing hereditary spherocytosis identified by nextâ€generation sequencing in a Chinese family. International Journal of Laboratory Hematology, 2021, 43, e294-e297.	1.3	0
46	Fetal Crown–Chin Length to Crown–Rump Length Ratio as a Prenatal Sonographic Marker for Triploidy at First Trimester. Journal of Ultrasound in Medicine, 2021, , .	1.7	1
47	First Report of Nondeletional Hb H Disease Caused by an α2-Globin Gene Mutation: <i>HBA2</i> : c.184A>T. Hemoglobin, 2021, 45, 210-211.	0.8	0
48	Fetal akinesia: The need for clinical vigilance in first trimester with decreased fetal movements. Taiwanese Journal of Obstetrics and Gynecology, 2021, 60, 559-562.	1.3	4
49	GATA zinc finger domainâ€containing protein 2A (<i>GATAD2A</i>) deficiency reactivates fetal haemoglobin in patients with βâ€thalassaemia through impaired formation of methylâ€binding domain protein 2 (MBD2)â€containing nucleosome remodelling and deacetylation (NuRD) complex. British lournal of Haematology, 2021, 193, 1220-1227.	2.5	5
50	Pregnancies with trisomy 2 cells in chorionic villi: Ultrasound determines the outcome. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 261, 247-248.	1.1	2
51	Is there a role for nuchal translucency in the detection of rare chromosomal abnormalities inÂtheÂera of noninvasive prenatal testing?. American Journal of Obstetrics and Gynecology, 2021, 225, 463-464.	1.3	2
52	Insufficient fetal fraction of cell-free DNA in non-invasive prenatal testing: Not always true. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 261, 245-247.	1.1	0
53	Noninvasive prenatal testing for aneuploidy in twin pregnancies with maternal plasma DNA sequencing. American Journal of Obstetrics and Gynecology, 2021, 224, 638-639.	1.3	0
54	Hb Lepore-Hong Kong: First Report of a Novel δ∫β-Globin Gene Fusion in a Chinese Family. Hemoglobin, 2021, 45, 1-5.	0.8	1

#	Article	IF	CITATIONS
55	Outcome of survivors with hemoglobin Bart's hydrops fetalis syndrome: The most severe form of αâ€thalassemia. Pediatric Transplantation, 2021, 25, e14090.	1.0	1
56	A genetic approach to the etiologic investigation of isolated intrauterine growth restriction. American Journal of Obstetrics and Gynecology, 2021, 225, 695-696.	1.3	2
57	Fetal micrognathia in the first trimester: An ominous finding even after a normal array. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 263, 176-180.	1.1	8
58	Tumor markers in cord blood: A predictor of fetal malignant neoplasm?. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 263, 280-281.	1.1	0
59	A New Hemoglobin Variant: Hb Jiujiang [α18(A16)Glyâ†'Cys, HBA2: c.55G>T]. Hemoglobin, 2021, 45, 1-2.	0.8	0
60	Exomeâ€based preconception carrier testing for consanguineous couples in China. Prenatal Diagnosis, 2021, 41, 1425-1429.	2.3	1
61	Is there an optimal gestational age for cell-free DNA testing in maternal obesity?. American Journal of Obstetrics and Gynecology, 2021, 225, 350.	1.3	1
62	Cellâ€free DNA screening for fetal 22q11.2 deletion: a targeted test or genomeâ€wide methodology?. Ultrasound in Obstetrics and Gynecology, 2021, 58, 644-646.	1.7	0
63	Early prenatal diagnosis of cleft lip and palate in a Chinese woman with a mosaic CDH1 variant. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 266, 45-47.	1.1	0
64	A Rare Case of Hb H Disease and Systemic Lupus Erythematosus. Hemoglobin, 2021, 45, 66-68.	0.8	1
65	Parental germline mosaic transmission of $5p13.2$ microduplication in two siblings of a Chinese family. Journal of Obstetrics and Gynaecology, 2021 , , $1-3$.	0.9	0
66	Further genetic testing in prenatal cases of nonimmune hydrops fetalis with a normal array: a targeted panel or exome?. American Journal of Obstetrics and Gynecology, 2021, , .	1.3	1
67	Dominant β-Thalassemia Phenotype Caused by Hb Dieppe (<i>HBB</i> : c.383A>G): Another Case Report. Hemoglobin, 2021, 45, 329-331.	0.8	1
68	The indications for early prenatal diagnosis of trisomy 18: a 7-year experience at mainland China. Journal of Maternal-Fetal and Neonatal Medicine, 2020, 33, 2038-2042.	1.5	0
69	Early prenatal detection of hypertrophic cardiomyopathy in Noonan syndrome: A case to remember. Congenital Anomalies (discontinued), 2020, 60, 68-70.	0.6	0
70	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
71	Short-rib polydactyly syndrome presenting with recurrent severe first-trimester phenotypes: the utility of exome sequencing in deciphering variants of DYNC2H1 gene. Journal of Obstetrics and Gynaecology, 2020, 40, 874-876.	0.9	1
72	First prenatal case of 48,XXYY syndrome detected by maternal cell-free DNA testing. Journal of Obstetrics and Gynaecology, 2020, 40, 270-272.	0.9	0

#	Article	IF	CITATIONS
73	Prenatal diagnosis of trisomy 22 at the first trimester of pregnancy. Journal of Obstetrics and Gynaecology, 2020, 40, 440-442.	0.9	2
74	Carnitine palmitoyltransferase II deficiency in a prenatal case with polycystic kidney diseaseâ€ike phenotype. Congenital Anomalies (discontinued), 2020, 60, 131-132.	0.6	1
75	Confined placental trisomy detection through non-invasive prenatal testing: benefit for pregnancy management. Journal of Obstetrics and Gynaecology, 2020, 40, 1020-1022.	0.9	O
76	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
77	Risk factors associated with fetal pleural effusion in prenatal diagnosis: a retrospective study in a single institute in Southern China. Journal of Obstetrics and Gynaecology, 2020, 40, 443-447.	0.9	5
78	Recurrent Wilms tumor in a Chinese family caused by a novel WT1 variant inherited from a mosaic parent. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 254, 333-334.	1.1	2
79	Use of noninvasive prenatal screening with cell-free DNA in late pregnancy with sonographic soft markers. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 252, 431-433.	1.1	2
80	Coâ€occurrence of two rare genetic diseases: A potential pitfall for prenatal diagnosis in successive pregnancies. Prenatal Diagnosis, 2020, 40, 1606-1609.	2.3	0
81	Exome sequencing improves genetic diagnosis of fetal increased nuchal translucency. Prenatal Diagnosis, 2020, 40, 1426-1431.	2.3	22
82	Hematological Characteristics of β-Globin Gene Mutation –50 (G>A) (HBB: c100G>A) Carriers in Mainland China. Hemoglobin, 2020, 44, 240-243.	0.8	10
83	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
84	Fetal blood sampling in midâ€pregnancy: does it still have a role in prenatal diagnosis?. Ultrasound in Obstetrics and Gynecology, 2020, 56, 791-792.	1.7	0
85	First-trimester cystic hygroma and neurodevelopmental disorders: The association to remember. Taiwanese Journal of Obstetrics and Gynecology, 2020, 59, 960-962.	1.3	10
86	Detection of Parental Contribution to Molar Genome Leads to Diagnosis of Recurrent Hydatidiform Mole in a Family with NLRP7 Variants. Fetal and Pediatric Pathology, 2020, , 1-8.	0.7	1
87	The Trend in Timing of Prenatal Diagnosis for Thalassemia at a Chinese Tertiary Obstetric Center. Hemoglobin, 2020, 44, 325-328.	0.8	2
88	Foetal phenotype of ALG1-CDG caused by paternal uniparental disomy 16. Journal of Obstetrics and Gynaecology, 2020, 41, 1-3.	0.9	3
89	Detection of an α-Globin Fusion Gene Using Real-Time Polymerase Chain Reaction-Based Multicolor Melting Curve. Hemoglobin, 2020, 44, 427-431.	0.8	5
90	Prenatal diagnosis of singleâ€gene disorders: theÂearlier, the better?. Ultrasound in Obstetrics and Gynecology, 2020, 56, 788-790.	1.7	0

#	Article	IF	Citations
91	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
92	Prenatal exome sequencing in fetuses with congenital heart defects. Clinical Genetics, 2020, 98, 215-230.	2.0	23
93	Prospective ultrasonographic diagnosis of orofacial clefts during the first trimester. Ultrasound in Obstetrics and Gynecology, 2020, 58, 134-137.	1.7	5
94	Hb Westmead (HBA2: c.369C>G): Hematological Characteristics in Heterozygotes with and without α0-Thalassemia. Hemoglobin, 2020, 44, 153-155.	0.8	6
95	Increased nuchal translucency: diagnostic value of RASopathyâ€disorder testing. Ultrasound in Obstetrics and Gynecology, 2020, 55, 423-424.	1.7	1
96	Allâ€transâ€retinoid acid induces the differentiation of P19 cells into neurons involved in the Pl3K/Akt/GSK3β signaling pathway. Journal of Cellular Biochemistry, 2020, 121, 4386-4396.	2.6	12
97	Hematological Characteristics of Hb Constant Spring (HBA2: c.427T>C) Carriers in Mainland China. Hemoglobin, 2020, 44, 86-88.	0.8	5
98	Value of increased nuchal translucency in the era of cellâ€free DNA testing. Ultrasound in Obstetrics and Gynecology, 2020, 55, 697-698.	1.7	0
99	Prenatal genetic diagnosis of cardiac rhabdomyoma: A single-center experience. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 249, 7-10.	1.1	5
100	Impact of cell-free fetal DNA on early invasive prenatal diagnosis at a Chinese reference maternal medicine center. Journal of Maternal-Fetal and Neonatal Medicine, 2020, , 1-5.	1.5	2
101	Neurofibromatosis type 1 due to possible maternal mosaicism in a family with two affected siblings. Congenital Anomalies (discontinued), 2020, 60, 156-157.	0.6	0
102	Early prenatal diagnosis of 49,XXXXY: two case reports. Journal of Obstetrics and Gynaecology, 2019, 39, 275-277.	0.9	1
103	Early prenatal detection of Bardetâ€Biedl syndrome in a case with postaxial polydactyly and hyperechoic kidneys confirmed by next generation sequencing. Congenital Anomalies (discontinued), 2019, 59, 142-144.	0.6	2
104	Unstable Hemoglobin Variants: The Need for Clinical Vigilance in Infants with Congenital Jaundice. Hemoglobin, 2019, 43, 60-62.	0.8	2
105	The role of ultrasound in the choice between chorionic villus sampling and amniocentesis for patients with a positive NIPT result for trisomy $18/13$. Prenatal Diagnosis, 2019, 39, 1155-1158.	2.3	6
106	Coinheritance of Hb City of Hope (HBB: c.208G>A) and \hat{I}^2 -Thalassemia: Compromising the Molecular Diagnosis of the Codons 71/72 (+A) (HBB: c.216_217insA) Mutation by Reverse Dot-Blot Hybridization. Hemoglobin, 2019, 43, 145-147.	0.8	0
107	Regulatory Single Nucleotide Polymorphism rs368698783 (G>A): a Genetic Modifier of Hb F Production Only under Erythropoietic Stress Characteristic for β-Globin Chain Deficiency?. Hemoglobin, 2019, 43, 73-75.	0.8	4
108	Germline mosaicism in an αâ€thalassemia family: Incidental identification by prenatal ultrasound. Prenatal Diagnosis, 2019, 39, 1166-1169.	2.3	2

#	Article	IF	CITATIONS
109	Fetal phenotype of Galloway-Mowat syndrome 3 caused by a specific OSGEP variant. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 242, 182-184.	1.1	1
110	Prenatal Ultrasound Presentations in Late Pregnancies Affected With Alpha Thalassemia Major. Pediatric and Developmental Pathology, 2019, 22, 603-604.	1.0	1
111	A Î ² -Thalassemia Trait with Two Mutations in Cis in a Chinese Family. Hemoglobin, 2019, 43, 289-291.	0.8	2
112	Congenital Nonspherocytic Hemolytic Anemia Caused by <i>Kr$\tilde{A}^{1}/4$ppel-Like Factor 1</i> Gene Variants: Another Case Report. Hemoglobin, 2019, 43, 292-295.	0.8	7
113	Clinical utility of noninvasive prenatal screening for pathogenic copy number variants. American Journal of Obstetrics and Gynecology, 2019, 221, 660.	1.3	1
114	Confined placental trisomy detection through cell-free DNA in the maternal circulation: Benefit forÂpregnancy management. American Journal of Obstetrics and Gynecology, 2019, 221, 286.	1.3	1
115	A <i>Krýppel-Like Factor 1</i> Gene Mutation Ameliorates the Severity of <b<math>\hat{l}^2-Thalassemia: A Case Report. Hemoglobin, 2019, 43, 137-139.</b<math>	0.8	4
116	Cystic hygroma and micromelic lower limbs: First-trimester sonographic markers of campomelic dysplasia. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 238, 191-193.	1.1	0
117	Congenital Cystic Diaphragm with Diaphragmatic Eventration in a Fetus: A Case Presentation. Fetal and Pediatric Pathology, 2019, 38, 335-339.	0.7	1
118	Rare autosomal trisomies on nonâ€invasive prenatal testing: not as adverse as expected. Ultrasound in Obstetrics and Gynecology, 2019, 54, 838-839.	1.7	15
119	Apparent germline mosaicism for a 15q11-q13 deletion causing recurrent Angelman syndrome in a Chinese family. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 236, 255-257.	1.1	3
120	Fetal phenotypes of congenital disorder of glycosylation: A case presentation. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 236, 257-258.	1.1	1
121	Diagnostic value of chromosomal microarray in fetuses with increased nuchal translucency. Ultrasound in Obstetrics and Gynecology, 2019, 53, 554-555.	1.7	2
122	The role of ultrasound in women with a positive NIPT result for trisomy 18 and 13. Taiwanese Journal of Obstetrics and Gynecology, 2019, 58, 798-800.	1.3	6
123	Detection of confined placental trisomy 16 using non-invasive prenatal testing in a pregnancy associated with intrauterine growth restriction and normal karyotype. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 233, 81-83.	1.1	6
124	Early prenatal detection of shortâ€rib polydactyly syndrome in a monochorionic diamniotic twin pregnancy. Congenital Anomalies (discontinued), 2019, 59, 181-182.	0.6	0
125	Prenatal diagnosis of 17q12 deletion syndrome: a retrospective case series. Journal of Obstetrics and Gynaecology, 2019, 39, 323-327.	0.9	22
126	Osteogenesis imperfecta type VIII: Association with increased nuchal translucency and prenatal diagnosis by targeted exome sequencing. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 235, 128-129.	1.1	1

#	Article	IF	Citations
127	Pitfall in genetic screening in a pregnancy involving an allogeneic hematopoietic stem cell transplantation recipient. Prenatal Diagnosis, 2019, 39, 52-53.	2.3	1
128	Chromosomal microarray analysis detects trisomy 9 mosaicism in a prenatal case not revealed by conventional cytogenetic analysis of cord blood. Journal of Obstetrics and Gynaecology, 2019, 39, 123-125.	0.9	4
129	Prenatal diagnosis of Wolf-Hirschhorn syndrome at the first trimester using chromosomal microarray analysis. Journal of Obstetrics and Gynaecology, 2019, 39, 268-270.	0.9	1
130	Application of chromosome microarray analysis in patients with unexplained developmental delay/intellectual disability in South China. Pediatrics and Neonatology, 2019, 60, 35-42.	0.9	21
131	Application of noninvasive prenatal testing in pregnancies with fetal double bubble sign: Is it feasible?. Prenatal Diagnosis, 2018, 38, 402-405.	2.3	2
132	Complex interactions between thalassemia defective alleles compromise screening and cause severe anemia in a Chinese family. International Journal of Laboratory Hematology, 2018, 40, e55-e58.	1.3	2
133	Prenatal diagnosis of Wolf-Hirschhorn syndrome: Ultrasonography and molecular karyotyping results. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 225, 19-21.	1.1	8
134	A <i>><scp>KLF</scp>1</i> yene mutation causes βâ€thalassemia minor in a Chinese family. International Journal of Laboratory Hematology, 2018, 40, e35-e37.	1.3	2
135	What would be missed in the first trimester if nuchal translucency measurement is replaced by cell free DNA foetal aneuploidy screening?. Journal of Obstetrics and Gynaecology, 2018, 38, 498-501.	0.9	11
136	Germline mosaicism in a DMD family: incidental identification in prenatal diagnosis. Journal of Obstetrics and Gynaecology, 2018, 38, 1166-1168.	0.9	1
137	αâ€Haemoglobin pool measurement: a useful biomarker for evaluation of βâ€thalassaemia intermedia?. British Journal of Haematology, 2018, 183, 673-674.	2.5	0
138	Whole exome sequencing as a diagnostic adjunct to clinical testing in fetuses with structural abnormalities. Ultrasound in Obstetrics and Gynecology, 2018, 51, 493-502.	1.7	113
139	Clinical outcome of pregnancies with the prenatal double bubble sign – a five-year experience from one single centre in mainland China. Journal of Obstetrics and Gynaecology, 2018, 38, 206-209.	0.9	11
140	Fetal-onset congenital dyserythropoietic anemia type 1 due to <i>CDAN1</i> mutations presenting as hydrops fetalis. Pediatric Hematology and Oncology, 2018, 35, 447-450.	0.8	4
141	First Report of a Case with Nondeletional Hb H Disease Caused by IVS-I-116 (A>G) of the α2-Globin Gene. Hemoglobin, 2018, 42, 344-346.	0.8	1
142	Results of Coexistence of \hat{I}^2 -Thalassemia Minor in Hb H Disease Patients. Hemoglobin, 2018, 42, 306-309.	0.8	6
143	<i>KFL1</i> Gene Variants in α-Thalassemia Individuals with Increased Fetal Hemoglobin in a Chinese Population. Hemoglobin, 2018, 42, 161-165.	0.8	5
144	Early prenatal diagnosis of lysosomal storage disorders by enzymatic and molecular analysis. Prenatal Diagnosis, 2018, 38, 779-787.	2.3	6

#	Article	IF	Citations
145	Pregnancy outcome of autosomal aneuploidies other than common trisomies detected by noninvasive prenatal testing in routine clinical practice. Prenatal Diagnosis, 2018, 38, 849-857.	2.3	24
146	\hat{l} -Thalassemia with Complete Absence of Hb A ₂ in a Chinese Family. Hemoglobin, 2018, 42, 135-137.	0.8	2
147	A cost-effectiveness analysis comparing two different strategies in advanced maternal age: Combined first-trimester screening and maternal blood cell-free DNA testing. Taiwanese Journal of Obstetrics and Gynecology, 2018, 57, 536-540.	1.3	8
148	How to make an accurate diagnosis of fetal pyriform sinus fistula in utero: experience at a single medical center in mainland China. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 228, 76-81.	1.1	6
149	Whole-exome sequencing identifies compound heterozygous LHX4 mutations in a fetus with early-onset growth restriction. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2017, 211, 225-227.	1.1	0
150	A Novel Frameshift Mutation at Codons 138/139 ($<$ i> $>$ HBB $<$ /i $>:$ c.417_418insT) on the \hat{I}^2 -Globin Gene Leads to \hat{I}^2 -Thalassemia. Hemoglobin, 2017, 41, 59-60.	0.8	2
151	Combination of Hb Heze [\hat{l}^2 144(HC1)Lysâ†'Arg;HBB: c.434A>G] and \hat{l}^2 O-Thalassemia in a Chinese Patient with \hat{l}^2 -Thalassemia Intermedia. Hemoglobin, 2017, 41, 47-49.	0.8	0
152	Prenatal diagnosis of Ectrodactyly–Ectodermal dysplasia–Cleft (EEC) syndrome in a Chinese woman with a TP63 mutation. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2017, 213, 146-147.	1.1	4
153	Outcome of isolated enlarged cisterna magna identified in utero: experience at a single medical center in mainland China. Prenatal Diagnosis, 2017, 37, 575-582.	2.3	9
154	Novel FREM1 mutations are associated with severe hydrocephalus and shortened limbs in a prenatal case. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2017, 215, 262-264.	1.1	5
155	Whole-exome sequencing for prenatal diagnosis of fetuses with congenital anomalies of the kidney and urinary tract. Nephrology Dialysis Transplantation, 2017, 32, 1665-1675.	0.7	54
156	A de novo ankyrin mutation (ANK1 Q109X) causing severe hereditary spherocytosis from preterm neonatal period. Annals of Hematology, 2017, 96, 1067-1068.	1.8	5
157	A prenatal case of osteogenesis imperfecta diagnosed with next-generation sequencing. Journal of Obstetrics and Gynaecology, 2017, 37, 809-810.	0.9	1
158	Increased first-trimester nuchal translucency associated with a dicentric chromosome and 9q34.3 microdeletion syndrome. Journal of Obstetrics and Gynaecology, 2017, 37, 327-329.	0.9	6
159	Characterization of a novel βâ€globin gene cluster deletion causing (^A γÎβ) ^O â€thalassemia by nextâ€generation sequencing. International Journal of Laboratory Hematology, 2017, 39, e19-e22.	1.3	1
160	Generation of Induced Pluripotent Stem Cells from Amniotic Fluid Cells of a Fetus with Hb Bart's Disease. Hemoglobin, 2017, 41, 198-202.	0.8	3
161	Pre Gestational Thalassemia Screening in Mainland China: The First Two Years of a Preventive Program. Hemoglobin, 2017, 41, 248-253.	0.8	26
162	Two α1-Globin Gene Point Mutations Causing Severe Hb H Disease. Hemoglobin, 2017, 41, 293-296.	0.8	5

#	Article	IF	CITATIONS
163	A novel selective deletion of the major αâ€globin regulatory element (MCSâ€R2) causing αâ€thalassaemia. British Journal of Haematology, 2017, 176, 984-986.	2.5	17
164	Treatment of fetal congenital chylothorax: Report of eight cases at a mainland Chinese medical center. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 867-869.	1.3	4
165	Hb A ₂ -Tianhe (<i>HBD</i> : c.323G>A): First Report in a Chinese Family with Normal Hb A ₂ -β-Thalassemia Trait. Hemoglobin, 2017, 41, 291-292.	0.8	4
166	First Report of the Rare IVS-II-705 (T>G) \hat{I}^2 -Thalassemia Mutation in a Chinese Family. Hemoglobin, 2017, 41, 286-287.	0.8	3
167	Hb Hornchurch [Î ² 43(CD2)Gluâ†'Lys; HBB: c.130G>A] Compromises the Molecular Diagnosis of Î ² -Thalassemia in a Chinese Family. Hemoglobin, 2017, 41, 274-277.	0.8	0
168	A Program on Noninvasive Prenatal Diagnosis of α-Thalassemia in Mainland China: A Cost–Benefit Analysis. Hemoglobin, 2016, 40, 247-249.	0.8	2
169	First Report of a Chinese Family Carrying a Double Heterozygosity for Hb Q-Thailand and Hb J-Bangkok. Hemoglobin, 2016, 40, 425-427.	0.8	4
170	Hb Alesha [β67(E11)Valâ†'Met (<i>G</i> TG> <i>A</i> TG); <i>HBB</i> : c.202G > A] Found in a Chin Hemoglobin, 2016, 40, 420-421.	ese Girl.	5
171	The Codon 35 (A > G) (<i>HBB</i> : c.107A > G) at the α - β Chain Interfac the β -Globin Gene: A Silent Mutation?. Hemoglobin, 2016, 40, 56-58.	e of 0.8	1
172	Prenatal diagnosis of fetal multicystic dysplastic kidney via high-resolution whole-genome array. Nephrology Dialysis Transplantation, 2016, 31, 1693-1698.	0.7	31
173	Prenatal diagnosis of thalassemia in twin pregnancies in mainland China. Journal of Obstetrics and Gynaecology, 2016, 36, 731-734.	0.9	0
174	Incidental prenatal detection of DMD gene deletion using array comparative genomic hybridization. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 203, 338-340.	1.1	1
175	Prenatal DNA diagnosis of Noonan syndrome in a fetus with increased nuchal translucency using next-generation sequencing. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 201, 229-230.	1.1	4
176	Consequences of Delayed Prenatal Diagnosis of \hat{l}^2 -Thalassemia in Mainland China. Hemoglobin, 2016, 40, 191-193.	0.8	3
177	A New Î'-Globin Gene Variant: Hb A ₂ -Fengshun [δ121(GH4)Glu→Lys (<i>HBD</i> : c.364G >â Hemoglobin, 2016, 40, 213-214.	€‰A)].	1
178	Diagnostic Dilemma of Hb Perth [î²32(B14)Leuâ†'Pro; <i>HBB</i> : c.98T > C] in Mainland China. Hem 2016, 40, 202-205.	oglobin,	1
179	Maternal serum PIGF (placental growth factor) in Chinese women in the first trimester undergoing screening for Down syndrome. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 201, 166-170.	1.1	11
180	The Frequency of \hat{l}_{\pm} -Globin Gene Triplication in a Southern Chinese Population. Indian Journal of Hematology and Blood Transfusion, 2016, 32, 320-322.	0.6	6

#	Article	IF	CITATIONS
181	Fetal Anemia and Hydrops Fetalis Associated with Homozygous Hb Constant Spring (HBA2: c.427T > C). Hemoglobin, 2016, 40, 97-101.	0.8	11
182	Noninvasive prenatal testing: impact on invasive prenatal diagnosis at a mainland Chinese tertiary medical center. Journal of Maternal-Fetal and Neonatal Medicine, 2016, 29, 1-12.	1.5	12
183	First Case of a Compound Heterozygosity for Two Nondeletional $\begin{subarray}{c} \begin{subarray}{c} \b$	0.8	4
184	First Detection of the â^27 (A > G) (HBB: c77A > G) Mutation of theβ-Globin Gene in a Chinese Hemoglobin, 2016, 40, 59-60.	e Family. 0.8	1
185	Detection of fetal copy number variants by nonâ€invasive prenatal testing for common aneuploidies. Ultrasound in Obstetrics and Gynecology, 2016, 47, 53-57.	1.7	61
186	A Novel Mutation of Ribosomal Protein S19 Gene in a Chinese Child with Diamond-Blackfan Anemia. Indian Journal of Hematology and Blood Transfusion, 2016, 32, 233-234.	0.6	0
187	Heterozygous \hat{l}^2 -thalassemia with complete absence of hemoglobin A2in a Chinese adult. International Journal of Laboratory Hematology, 2015, 37, e147-e149.	1.3	2
188	Compound Heterozygosity for HK $<$ b $>$ Î \pm Î $\pm <$ /b $>$ and an $<$ i $>$ in Cis $<$ /i $>Deletion of DoubleÎ\pm Genes Presents asÎ\pm -Thalassemia Trait. Hemoglobin, 2015, 39, 256-259.$	0.8	6
189	Rapid prenatal diagnosis of complete mole with co-existing twin by QF-PCR analysis. Journal of Obstetrics and Gynaecology, 2015, 35, 526-544.	0.9	O
190	Implementation of Newborn Screening for Hemoglobin H Disease in Mainland China. Indian Journal of Hematology and Blood Transfusion, 2015, 31, 242-246.	0.6	2
191	Association of an α-Globin Gene Cluster Duplication and Heterozygous β-Thalassemia in a Patient with a Severe Thalassemia Syndrome. Hemoglobin, 2015, 39, 102-106.	0.8	15
192	Identification of Nondeletional $<$ b $>$ Î $\pm <$ /b $>$ -Thalassemia in a Prenatal Screening Program by Reverse Dot-Blot in Southern China. Hemoglobin, 2015, 39, 42-45.	0.8	6
193	Thalassemia Intermedia Caused by $16p13.3$ Sectional Duplication in a \hat{I}^2 -Thalassemia Heterozygous Child. Pediatric Hematology and Oncology, 2015, 32, 349-353.	0.8	14
194	Neonatal screening for αâ€thalassemia by cord hemoglobin Barts: how effective is it?. International Journal of Laboratory Hematology, 2015, 37, 649-653.	1.3	5
195	Screening for common βâ€globin gene cluster deletions in Chinese individuals with increased hemoglobin F. International Journal of Laboratory Hematology, 2015, 37, 752-757.	1.3	15
196	Non-invasive prenatal detection of haemoglobin Bart's disease by cardiothoracic ratio during the first trimester. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2015, 193, 92-95.	1.1	9
197	Evidence of Selection for the $\langle b \rangle \hat{l} \pm \langle b \rangle$ -Globin Gene Deletions and Triplications in a Southern Chinese Population. Hemoglobin, 2015, 39, 442-444.	0.8	10
198	First-trimester combined screening for trisomy 21 in women at risk for \hat{I}_{\pm} -thalassemia. Journal of Maternal-Fetal and Neonatal Medicine, 2015, 29, 1-3.	1.5	0

#	Article	IF	CITATIONS
199	Prenatal diagnosis of 17q12 duplication and deletion syndrome in two fetuses with congenital anomalies. Taiwanese Journal of Obstetrics and Gynecology, 2014, 53, 579-582.	1.3	22
200	Chorionic villus sampling for early prenatal diagnosis: Experience at a mainland Chinese hospital. Journal of Obstetrics and Gynaecology, 2014, 34, 669-672.	0.9	6
201	Noninvasive prenatal diagnosis of common aneuploidies by semiconductor sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 7415-7420.	7.1	110
202	Prenatal diagnosis of foetuses with congenital abnormalities and duplication of the MECP2 region. Gene, 2014, 546, 222-225.	2.2	17
203	Delineation of the molecular basis of borderline hemoglobin A2 in Chinese individuals. Blood Cells, Molecules, and Diseases, 2014, 53, 261-264.	1.4	18
204	Prenatal diagnosis of sex chromosome aneuploidies: experience at a mainland Chinese hospital. Journal of Obstetrics and Gynaecology, 2013, 33, 827-829.	0.9	4
205	Association of Hb New York with Hb E and $\hat{l}\pm < sup > 0 < sup > -Thalassemia in a Chinese Woman Identified by Sebia Capillarys 2 System. Hemoglobin, 2012, 36, 157-160.$	0.8	2
206	α0-Thalassemia Trait with Normal Red Cell Indices: A Report of Two Cases. Hemoglobin, 2012, 36, 589-591.	0.8	2
207	<pre><scp>H</scp>b <scp>A</scp>₂ levels in patients with nondeletional <scp>H</scp>b <scp>H</scp> disease. International Journal of Laboratory Hematology, 2012, 34, 663-664.</pre>	1.3	2
208	Homozygous α-Thalassemia-1 Presenting in a Fetus without Anemia. Acta Haematologica, 2010, 123, 207-209.	1.4	2
209	Monozygotic Twins Discordant for Dandy-Walker Malformation. Fetal Diagnosis and Therapy, 2009, 25, 141-143.	1.4	3
210	A case of transfusionâ€dependent nondeletional Hb H disease undiagnosed during prenatal screening for thalassemia. Prenatal Diagnosis, 2008, 28, 165-166.	2.3	11
211	Association of Hb Q-Thailand with Heterozygous Hb E in a Chinese Patient. Hemoglobin, 2008, 32, 319-321.	0.8	2
212	Incidental Discovery of Nonpaternity during Prenatal Testing of Genetic Disease. Fetal Diagnosis and Therapy, 2008, 24, 39-41.	1.4	7
213	Hydrops Fetalis Caused by Homozygous Alpha-Thalassemia and Kidd Antigen Alloimmunization in a Chinese Woman. Fetal Diagnosis and Therapy, 2008, 24, 331-333.	1.4	3
214	Misdiagnosis of Hb Constant Spring ($\hat{l}\pm 142$, Term→Gln,TAA→CAA in $\hat{l}\pm 2$) in a Hb H (\hat{l}^24) Disease Child. Hemoglo 2007, 31, 105-108.	bin, 0:8	7
215	Four Cases of Hb Q-H Disease Found in Southern China. Hemoglobin, 2007, 31, 109-111.	0.8	7
216	Prenatal Diagnosis of \hat{l}^2 -Thalassemia by Reverse Dot-Blot Hybridization in Southern China. Hemoglobin, 2006, 30, 365-370.	0.8	42

#	Article	IF	CITATION
217	The Codon 37 (TGG→TAG)βO-Thalassemia Mutation Found in a Chinese Family. Hemoglobin, 2006, 30, 171-173.	0.8	7
218	Prenatal diagnosis of \hat{I}^2 -thalassemia in Southern China. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2006, 128, 81-85.	1.1	14
219	Efficacy and safety of cordocentesis for prenatal diagnosis. International Journal of Gynecology and Obstetrics, 2006, 93, 13-17.	2.3	64
220	Detection of alpha-thalassemia in beta-thalassemia carriers and prevention of Hb Bart's hydrops fetalis through prenatal screening. Haematologica, 2006, 91, 649-51.	3.5	22
221	Prenatal diagnosis and molecular analysis of type 1 thanatophoric dysplasia. International Journal of Gynecology and Obstetrics, 2005, 91, 268-270.	2.3	6
222	Missed Diagnosis of $\hat{l}\pm$ -Globin Gene Cluster Duplication in Prenatal Screening: Identified Incidentally by Invasive Testing. Indian Journal of Hematology and Blood Transfusion, $0, 1$.	0.6	0