

非侵入式胎兒染色體檢測 NIFTY 系列

超過 **50** 篇醫學論文佐證支持

	文獻標題 Title	刊登期刊
1	Genomic Analyses from Non-invasive Prenatal Testing Reveal Genetic Associations, Patterns of Viral Infections, and Chinese Population History	<u>Cell</u> . 2018 Oct 4;175(2):347-359
2	Incidental identification of maternal malignancies in two Asian women underwent noninvasive prenatal test.	bioRxiv preprint
3	Haplotype-assisted accurate non-invasive fetal whole genome recovery through maternal plasma sequencing	<u>Genome Med</u> . 2013 Feb 27; 5(2): 18
4	Integration of targeted sequencing and NIPT into clinical practice in a Chinese family with maple syrup urine disease	<u>Genet Med</u> . 2014 Aug;16(8):594-600
5	Feasibility Study of Semiconductor Sequencing for Noninvasive Prenatal Detection of Fetal Aneuploidy	<u>Clin Chem</u> . 2013 May;59(5):846-9
6	Validation of combinatorial probe–anchor ligation-based sequencing as non-invasive prenatal test for trisomy at a central laboratory	<u>Ultrasound Obstet Gynecol</u> . 2017 Jul;50(1):49-57
7	Targeted sequencing of maternal plasma for haplotype-based non-invasive prenatal testing of spinal muscular atrophy	<u>Ultrasound Obstet Gynecol</u> . 2017 Jun;49(6):799-802
8	Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies.	<u>Ultrasound Obstet Gynecol</u> . 2015 May;45(5):530-8
9	Detection of fetal sex chromosome aneuploidy by massively parallel sequencing of maternal plasma DNA: initial experience in a Chinese hospital	<u>Ultrasound Obstet Gynecol</u> . 2014 Jul;44(1):17-24
10	False-negative trisomy 18 non-invasive prenatal test result due to 48,XXX,+18 placental mosaicism	<u>Ultrasound Obstet Gynecol</u> . 2014 Apr;43(4):477-8
11	Non-invasive prenatal testing for fetal chromosomal abnormalities by low-coverage whole-genome sequencing of maternal plasma DNA: review of 1982 consecutive cases in a single center.	<u>Ultrasound Obstet Gynecol</u> . 2014 Mar;43(3):254-64
12	Genome-wide detection of additional fetal chromosomal abnormalities by cell-free DNA testing of 15626 consecutive pregnant women.	<u>Sci China Life Sci</u> . 2019 Feb;62(2):215-224
13	Non-invasive prenatal assessment of trisomy 21 by multiplexed maternal plasma DNA sequencing: large scale validity study.	<u>BMJ</u> . 2011 Jan 11;342:c7401

14	Haplotype-based approach for noninvasive prenatal tests of Duchenne muscular dystrophy using cell-free fetal DNA in maternal plasma	<u>Genet Med.</u> 2015 Nov;17(11):889-96
15	Noninvasive prenatal testing for autosomal recessive conditions by maternal plasma sequencing in a case of congenital deafness.	<u>Genet Med.</u> 2014 Dec;16(12):972-6
16	Reproductive management through integration of PGD and MPS-based noninvasive prenatal screening/diagnosis for a family with GJB2-associated hearing impairment	<u>Sci China Life Sci.</u> 2015 Sep;58(9):829-38
17	An Advanced Model to Precisely Estimate the Cell-Free Fetal DNA Concentration in Maternal Plasma	<u>PLoS One.</u> 2016 Sep 23;11(9):e0161928
18	Noninvasive Prenatal Paternity Testing (NIPAT) through Maternal Plasma DNA Sequencing:A Pilot Study	<u>PLoS One.</u> 2016 Sep 15;11(9):e0159385
19	Performance Evaluation of NIPT in Detection of Chromosomal Copy Number Variants Using Low-Coverage Whole-Genome Sequencing of Plasma DNA	<u>PLoS One.</u> 2016 Jul 14;11(7):e0159233
20	Effective Noninvasive Zygosity Determination by Maternal Plasma Target Region Sequencing	<u>PLoS One.</u> 2013 Jun 10;8(6):e65050
21	Noninvasive Prenatal Detection for Pathogenic CNVs:The Application in a-Thalassemia	<u>PLoS One.</u> 2013 Jun 28;8(6):e67464
22	A single cell level based method for copy number variation analysis by low coverage massively parallel sequencing.	<u>PLoS One.</u> 2013;8(1):e54236
23	Noninvasive Prenatal Diagnosis of Fetal Trisomy 18 and Trisomy 13 by Maternal Plasma DNA Sequencing	<u>PLoS One.</u> 2011; 6(7): e21791
24	Noninvasive Fetal Trisomy (NIFTY) test: an advanced noninvasive prenatal diagnosis methodology for fetal autosomal and sex chromosomal aneuploidies	<u>BMC Med Genomics.</u> 2012 Dec 1;5:57
25	Effects of Maternal and Fetal Characteristics on Cell-Free Fetal DNA Fraction in Maternal Plasma.	<u>Reprod Sci.</u> 2015 Nov;22(11):1429-35
26	Noninvasive prenatal test for FGFR3-related skeletal dysplasia based on next-generation sequencing and plasma cell-free DNA: Test performance analysis and feasibility exploration.	<u>Prenat Diagn.</u> 2018 Oct;38(11):821-828
27	Isolation and whole genome sequencing of fetal cells from maternal blood towards the ultimate non-invasive prenatal testing	<u>Prenat Diagn.</u> 2017 Dec;37(13):1311-1321
28	Noninvasive prenatal testing (NIPT) in twin pregnancies with treatment of assisted reproductive techniques (ART) in a single center	<u>Prenat Diagn.</u> 2016 Jul;36(7):672-9
29	Clinical experience from Thailand: noninvasive prenatal testing as screening tests for trisomies 21, 18 and 13 in 4736 pregnancies	<u>Prenat Diagn.</u> 2016 Mar;36(3):224-31
30	Noninvasive prenatal testing of trisomies 21 and 18 by massively	<u>Prenat Diagn.</u> 2014

	parallel sequencing of maternal plasma DNA in twin pregnancies	Apr;34(4):335-40
31	Clinical application of noninvasive prenatal testing for the detection of trisomies 21, 18, and 13: A hospital experience	<u>Prenat Diagn.</u> 2014 Nov;34(11):1061-5
32	Discordant results between fetal karyotyping and non-invasive prenatal testing by maternal plasma sequencing in a case of uniparental disomy 21 due to trisomic rescue	<u>Prenat Diagn.</u> 2013 Jun;33(6):598-601
33	Secondary findings from non-invasive prenatal testing for common fetal aneuploidies by whole genome sequencing as a clinical service	<u>Prenat Diagn.</u> 2013 Jun;33(6):602-8
34	A method for noninvasive detection of fetal large deletions/duplications by low coverage massively parallel sequencing	<u>Prenat Diagn.</u> 2013 Jun;33(6):584-90
35	Fetal aneuploidy screening by maternal plasma DNA sequencing: 'false positive' due to confined placental mosaicism.	<u>Prenat Diagn.</u> 2013 Feb;33(2):198-200
36	Clinical application of massively parallel sequencing-based prenatal noninvasive fetal trisomy test for trisomies 21 and 18 in 11,105 pregnancies with mixed risk factors.	<u>Prenat Diagn.</u> 2012 Dec;32(13):1225-32
37	Noninvasive prenatal genetic testing for fetal aneuploidy detects maternal trisomy X	<u>Prenat Diagn.</u> 2012 Nov;32(11):1114-6
38	Haplotype-based approach for noninvasive prenatal diagnosis of congenital adrenal hyperplasia by maternal plasma DNA sequencing	<u>Gene.</u> 2014 Jul 10;544(2):252-8
39	Genetic effects of a 13q31.1 microdeletion detected by noninvasive prenatal testing (NIPT)	<u>Int J Clin Exp Pathol.</u> 2014 Sep 15;7(10):7003-11
40	External Quality Assessment for Detection of Fetal Trisomy 21, 18, and 13 by Massively Parallel Sequencing in Clinical Laboratories	<u>J Mol Diagn.</u> 2016 Mar;18(2):244-52
41	Non-invasive fetal sex determination by maternal plasma sequencing and application in X-linked disorder counseling	<u>J Matern Fetal Neonatal Med.</u> 2014 Dec;27(18):1829-33
42	Non-invasive prenatal screening of fetal Down syndrome by maternal plasma DNA sequencing in twin pregnancies.	<u>J Matern Fetal Neonatal Med.</u> 2013 Mar;26(4):434-7
43	Non-invasive prenatal screening of fetal sex chromosomal abnormalities: perspective of pregnant women	<u>J Matern Fetal Neonatal Med.</u> 2012 Dec;25(12):2616-9
44	Clinical utility of noninvasive fetal trisomy (NIFTY) test – early experience	<u>J Matern Fetal Neonatal Med.</u> 2012 Oct;25(10):1856-9
45	Noninvasive prenatal diagnosis of common fetal chromosomal aneuploidies by maternal plasma DNA sequencing.	<u>J Matern Fetal Neonatal Med.</u> 2012

		Aug;25(8):1370-4
46	Detection of trisomies 13, 18 and 21 using non-invasive prenatal testing.	<u>Exp Ther Med.</u> 2017 May;13(5):2304-2310
47	A prenatal case with discrepant findings between non-invasive prenatal testing and fetal genetic testings	<u>Mol Cytogenet.</u> 2014 Jul 16;7:48
48	A Pilot Study of Noninvasive Prenatal Diagnosis of Alpha- and Beta-Thalassemia with Target Capture Sequencing of Cell-Free Fetal DNA in Maternal Blood	<u>Genet Test Mol Biomarkers.</u> 2017 Jul;21(7):433-439
49	Copy number variation profile in noninvasive prenatal testing (NIPT) can identify co-existing maternal malignancies: Case reports and a literature review	<u>Taiwan J Obstet Gynecol.</u> 2018 Dec;57(6):871-877.
50	Primer-Introduced Restriction Analysis Polymerase Chain Reaction Method for Non-Invasive Prenatal Testing of β -Thalassemia	<u>Hemoglobin.</u> 2015;39(1):18-23
51	孕婦血漿中胎兒游離DNA濃度檢測新方法	Medical Journal of Wuhan University
52	Chromosomal aneuploid non-invasive prenatal genetic testing false positive false negative Biological cause analysis	<u>Zhonghua Yi Xue Yi Chuan Xue Za Zhi.</u> 2018 Jun 10;35(3):329-333
53	Efficacy safety and health economic analysis of Down's syndrome screening program in early pregnancy	中國婦產科臨床雜誌
54	Advances in research and application of non-invasive prenatal gene detection for fetal chromosome aneuploidy	中國產前診斷雜誌
55	Application of Illumina sequencing technology in detection of fetal aneuploidy by maternal blood	中國實驗診斷學
56	Application prospect of fetal DNA in pregnant women's peripheral blood in prenatal diagnosis of Down's syndrome	中國婦產科臨床雜誌