Mark D Pertile

List of Publications by Year
in descending order

53 papers	1,649 citations	304602 22 h-index	302012 39 g-index
55 all docs	55 docs citations	55 times ranked	2076 citing authors

#	Article	IF	Citations
1	Chromosome translocations in couples with in-vitro fertilization implantation failure. Human Reproduction, 1999, 14, 2097-2101.	0.4	144
2	Rare autosomal trisomies, revealed by maternal plasma DNA sequencing, suggest increased risk of feto-placental disease. Science Translational Medicine, 2017, 9, .	5.8	122
3	Case Report: Delivery of normal twins following the intracytoplasmic injection of spermatozoa from a patient with 47, XXY Klinefelter's syndrome. Human Reproduction, 1997, 12, 2447-2450.	0.4	115
4	Rapid, high throughput prenatal detection of aneuploidy using a novel quantitative method (MLPA). Journal of Medical Genetics, 2003, 40, 907-912.	1.5	108
5	Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. Genetics in Medicine, 2018, 20, 513-523.	1.1	80
6	Pathogenic aberrations revealed exclusively by single nucleotide polymorphism (SNP) genotyping data in 5000 samples tested by molecular karyotyping. Journal of Medical Genetics, 2011, 48, 831-839.	1.5	71
7	Abnormal plasma DNA profiles in early ovarian cancer using a non-invasive prenatal testing platform: implications for cancer screening. BMC Medicine, 2016, 14, 126.	2.3	69
8	Methylation of the adenomatous polyposis coli (APC) gene in human placenta and hypermethylation in choriocarcinoma cells. Cancer Letters, 2008, 268, 56-62.	3.2	66
9	Harmonizing Cell-Free DNA Collection and Processing Practices through Evidence-Based Guidance. Clinical Cancer Research, 2020, 26, 3104-3109.	3.2	66
10	Rapid evolution of mouse Y centromere repeat DNA belies recent sequence stability. Genome Research, 2009, 19, 2202-2213.	2.4	51
11	High-throughput analysis of chromosome abnormality in spontaneous miscarriage using an MLPA subtelomere assay with an ancillary FISH test for polyploidy. American Journal of Medical Genetics, Part A, 2006, 140A, 2786-2793.	0.7	45
12	Monozygotic twins with discordant karyotypes: a case report. Journal of Pediatrics, 2004, 145, 406-408.	0.9	43
13	Mesenchymal Stem/Stromal Cells Derived From a Reproductive Tissue Niche Under Oxidative Stress Have High Aldehyde Dehydrogenase Activity. Stem Cell Reviews and Reports, 2016, 12, 285-297.	5.6	41
14	International Society for Prenatal Diagnosis Position Statement: cell free (cf)<scp>DNA</scp> screening for Down syndrome in multiple pregnancies. Prenatal Diagnosis, 2021, 41, 1222-1232.	1.1	41
15	Establishment and characterization of fetal and maternal mesenchymal stem/stromal cell lines from the human term placenta. Placenta, 2016, 39, 134-146.	0.7	38
16	â€˜Is it better not to know certain things?â€™: views of women who have undergone non-invasive prenatal testing on its possible future applications. Journal of Medical Ethics, 2019, 45, 231-238.	1.0	37
17	Ectopic Bone Formation by Mesenchymal Stem Cells Derived from Human Term Placenta and the Decidua. PLoS ONE, 2015, 10, e0141246.	1.1	36
18	Health and developmental outcome of children following prenatal diagnosis of confined placental mosaicism. Prenatal Diagnosis, 2006, 26, 443-448.	1.1	34

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19	Meeting the challenge of interpreting highâ€resolution single nucleotide polymorphism array data in prenatal diagnosis: does increased diagnostic power outweigh the dilemma of rare variants?. BJOG: an International Journal of Obstetrics and Gynaecology, 2013, 120, 594-606.	1.1	34
20	Mesenchymal stem cells reside in a vascular niche in the decidua basalis and are absent in remodelled spiral arterioles. Placenta, 2015, 36, 312-321.	0.7	34
21	Prospective ranking of the sonographic markers for aneuploidy: Data of 2143 prenatal cytogenetic diagnoses referred for abnormalities on ultrasound. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2003, 43, 16-26.	0.4	28
22	â€˜Small cost to pay for peace of mindâ€™: Women's experiences with nonâ€invasive prenatal testing. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2019, 59, 649-655.	0.4	26
23	Performance of a Paired-End Sequencing-Based Noninvasive Prenatal Screening Test in the Detection of Genome-Wide Fetal Chromosomal Anomalies. Clinical Chemistry, 2021, 67, 1210-1219.	1.5	25
24	PRENATAL DIAGNOSIS OF PRADERâ€“WILLI SYNDROME USING PW71 METHYLATION ANALYSISâ€”UNIPARENTAL DISOMY AND THE SIGNIFICANCE OF RESIDUAL TRISOMY 15. , 1997, 17, 109-113.		21
25	Offering pregnant women different levels of genetic information from prenatal chromosome microarray: a prospective study. European Journal of Human Genetics, 2018, 26, 485-494.	1.4	19
26	Cancer Diagnoses Following Abnormal Noninvasive Prenatal Testing: A Case Series, Literature Review, and Proposed Management Model. JCO Precision Oncology, 2021, 5, 1001-1012.	1.5	19
27	Trisomy 13 mosaicism at prenatal diagnosis: dilemmas in interpretation. , 1998, 18, 45-49.		18
28	Monosomy 21 Seen in Live Born Is Unlikely to Represent True Monosomy 21: A Case Report and Review of the Literature. Case Reports in Genetics, 2014, 2014, 1-6.	0.1	17
29	Stateâ€wide utilization and performance of traditional and cellâ€free DNAâ€based prenatal testing pathways: the Victorian Perinatal Record Linkage (PeRL) study. Ultrasound in Obstetrics and Gynecology, 2020, 56, 215-224.	0.9	17
30	Isochromosome 21q is overrepresented among false-negative cell-free DNA prenatal screening results involving Down syndrome. European Journal of Human Genetics, 2018, 26, 1490-1496.	1.4	16
31	Genome-wide noninvasive prenatal screening for carriers of balanced reciprocal translocations. Genetics in Medicine, 2020, 22, 1944-1955.	1.1	16
32	Trisomy 16 Mosaicism at Chorionic Villus Sampling and Amniocentesis with a Normal Physical and Intellectual Outcome. Fetal Diagnosis and Therapy, 2010, 28, 117-118.	0.6	15
33	Maternal mosaicism for a large segmental duplication of 18q as a secondary finding following nonâ€invasive prenatal testing and implications for test accuracy. Prenatal Diagnosis, 2015, 35, 986-989.	1.1	15
34	Reexamining the optimal nuchal translucency cutoff for diagnostic testing in the cell-free DNA and microarray era: results from the Victorian Perinatal Record Linkage study. American Journal of Obstetrics and Gynecology, 2021, 225, 527.e1-527.e12.	0.7	15
35	Questionable pathogenicity of FOXG1 duplication. European Journal of Human Genetics, 2012, 20, 595-596.	1.4	14
36	Isolation and Characterization of Mesenchymal Stem/Stromal Cells Derived from Human Third Trimester Placental Chorionic Villi and Decidua Basalis. Methods in Molecular Biology, 2018, 1710, 247-266.	0.4	13

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37	A Novel Combination of Homeobox Genes Is Expressed in Mesenchymal Chorionic Stem/Stromal Cells in First Trimester and Term Pregnancies. Reproductive Sciences, 2014, 21, 1382-1394.	1.1	12
38	First trimester detection of trisomy 16 using combined biochemical and ultrasound screening. Prenatal Diagnosis, 2014, 34, 291-295.	1.1	10
39	Genome-Wide Cell-Free DNA-Based Prenatal Testing for Rare Autosomal Trisomies and Subchromosomal Abnormalities. , 2018, , 97-123.		9
40	Confirmation of trisomy 22 in two cases using chromosome painting: Comparison with t(11;22). American Journal of Medical Genetics Part A, 1993, 46, 434-437.	2.4	8
41	A Novel Mechanism for Human Cardiac Ankyrin-B Syndrome due to Reciprocal Chromosomal Translocation. Heart Lung and Circulation, 2017, 26, 612-618.	0.2	8
42	A minimum estimate of the prevalence of 22q11 deletion syndrome and other chromosome abnormalities in a combined prenatal and postnatal cohort. Human Reproduction, 2020, 35, 694-704.	0.4	7
43	Haplotyping the human leukocyte antigen system from single chromosomes. Scientific Reports, 2016, 6, 30381.	1.6	6
44	The clinical benefit of genome-wide cfDNA testing cannot be extrapolated from CVS data. Genetics in Medicine, 2020, 22, 657-658.	1.1	4
45	Chromosome Microarrays in Diagnostic Testing: Interpreting the Genomic Data. Methods in Molecular Biology, 2014, 1168, 117-155.	0.4	4
46	Bilateral Congenital Adrenal Agenesis: A Rare Disease Entity and Not a Result of Poor Autopsy Technique. Pediatric and Developmental Pathology, 2014, 17, 308-311.	0.5	3
47	A fetus coexisting with a complete hydatidiform mole with trisomy 9 of maternal origin. Journal of Obstetrics and Gynaecology Research, 2018, 44, 955-959.	0.6	3
48	Unstable Robertsonian translocations der(13;15)(q10;q10): Heritable chromosome fission without phenotypic effect in two kindreds. American Journal of Medical Genetics, Part A, 2005, 136A, 25-30.	0.7	1
49	Minimal impact of maternal intravenous immunoglobulin infusion on cell-free DNA sequencing for fetal aneuploidy. Ultrasound in Obstetrics and Gynecology, 2016, 48, 250-250.	0.9	1
50	Is chorionic villus sampling as reliable as amniocentesis?. Pathology, 1992, 24, 30.	0.3	0
51	Exceptional Complex Chromosomal Rearrangements in Three Generations. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	0
52	Chromosome microarray proficiency testing and analysis of quality metric data trends through an external quality assessment program for Australasian laboratories. Pathology, 2016, 48, 586-596.	0.3	0
53	Association between timing of diagnosis of trisomy 21, 18, and 13 and maternal socioâ€economic status in Victoria, Australia: A populationâ€based cohort study from 2015 to 2016. Prenatal Diagnosis, 2019, 39, 1254-1261.	1.1	0