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	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
2	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
3	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
4	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
5	The clinical benefit of genome-wide cfDNA testing cannot be extrapolated from CVS data. Genetics in Medicine, 2020, 22, 657-658.	1.1	4
6	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
7	Genome-wide noninvasive prenatal screening for carriers of balanced reciprocal translocations. Genetics in Medicine, 2020, 22, 1944-1955.	1.1	16
8	Harmonizing Cell-Free DNA Collection and Processing Practices through Evidence-Based Guidance. Clinical Cancer Research, 2020, 26, 3104-3109.	3.2	66
9	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
10	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
11	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
12	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
13	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
14	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
15	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
16	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
17	Genome-Wide Cell-Free DNA-Based Prenatal Testing for Rare Autosomal Trisomies and Subchromosomal Abnormalities. , 2018, , 97-123.		9
18	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

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19	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
20	Rare autosomal trisomies, revealed by maternal plasma DNA sequencing, suggest increased risk of feto-placental disease. Science Translational Medicine, 2017, 9, .	5.8	122
21	Haplotyping the human leukocyte antigen system from single chromosomes. Scientific Reports, 2016, 6, 30381.	1.6	6
22	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
23	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
24	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
25	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
26	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
27	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
28	Ectopic Bone Formation by Mesenchymal Stem Cells Derived from Human Term Placenta and the Decidua. PLoS ONE, 2015, 10, e0141246.	1.1	36
29	Exceptional Complex Chromosomal Rearrangements in Three Generations. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	0
30	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
31	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
32	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
33	First trimester detection of trisomy 16 using combined biochemical and ultrasound screening. Prenatal Diagnosis, 2014, 34, 291-295.	1.1	10
34	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
35	Chromosome Microarrays in Diagnostic Testing: Interpreting the Genomic Data. Methods in Molecular Biology, 2014, 1168, 117-155.	0.4	4
36	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

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37	Questionable pathogenicity of FOXG1 duplication. European Journal of Human Genetics, 2012, 20, 595-596.	1.4	14
38	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
39	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
40	Rapid evolution of mouse Y centromere repeat DNA belies recent sequence stability. Genome Research, 2009, 19, 2202-2213.	2.4	51
41	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
42	Health and developmental outcome of children following prenatal diagnosis of confined placental mosaicism. Prenatal Diagnosis, 2006, 26, 443-448.	1.1	34
43	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
44	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
45	Monozygotic twins with discordant karyotypes: a case report. Journal of Pediatrics, 2004, 145, 406-408.	0.9	43
46	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
47	Rapid, high throughput prenatal detection of aneuploidy using a novel quantitative method (MLPA). Journal of Medical Genetics, 2003, 40, 907-912.	1.5	108
48	Chromosome translocations in couples with in-vitro fertilization implantation failure. Human Reproduction, 1999, 14, 2097-2101.	0.4	144
49	Trisomy 13 mosaicism at prenatal diagnosis: dilemmas in interpretation. , 1998, 18, 45-49.		18
50	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
51	PRENATAL DIAGNOSIS OF PRADERâ€“WILLI SYNDROME USING PW71 METHYLATION ANALYSISâ€”UNIPARENTAL DISOMY AND THE SIGNIFICANCE OF RESIDUAL TRISOMY 15. , 1997, 17, 109-113.		21
52	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
53	Is chorionic villus sampling as reliable as amniocentesis?. Pathology, 1992, 24, 30.	0.3	0