

Ignatia B Van Den Veyver

List of Publications by Year in descending order

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Version: 2024-02-01

148
papers

9,964
citations

71061

41
h-index

36008

97
g-index

193
all docs

193
docs citations

193
times ranked

10528
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 1 | eP462: Detection of clinically relevant exonic copy number changes in fetuses by chromosomal microarray analysis. <i>Genetics in Medicine</i> , 2022, 24, S291. | 1.1 | 0 |
| 2 | eP474: Prenatal exome sequencing analysis in the clinical setting of fetuses with structural anomalies or increased nuchal translucency. <i>Genetics in Medicine</i> , 2022, 24, S301. | 1.1 | 0 |
| 3 | OP054: Chromosomal microarray analysis for open neural tube defect: The prevalence of significant results and implications on in utero repair. <i>Genetics in Medicine</i> , 2022, 24, S378-S381. | 1.1 | 0 |
| 4 | Parental mosaicism for apparent de novo genetic variants: Scope, detection, and counseling challenges. <i>Prenatal Diagnosis</i> , 2022, 42, 811-821. | 1.1 | 8 |
| 5 | Information is power: The experiences, attitudes and needs of individuals who chose to have prenatal genomic sequencing for fetal anomalies. <i>Prenatal Diagnosis</i> , 2022, 42, 947-954. | 1.1 | 3 |
| 6 | Prenatal exomes and genomes – so much new and so much more to learn. <i>Prenatal Diagnosis</i> , 2022, 42, 659-661. | 1.1 | 0 |
| 7 | International Society for Prenatal Diagnosis Updated Position Statement on the use of genome-wide sequencing for prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2022, 42, 796-803. | 1.1 | 37 |
| 8 | Reproductive Outcomes from Maternal Loss of Nlrp2 Are Not Improved by IVF or Embryo Transfer Consistent with Oocyte-Specific Defect. <i>Reproductive Sciences</i> , 2021, 28, 1850-1865. | 1.1 | 3 |
| 9 | Use of amplicon-based sequencing for testing fetal identity and monogenic traits with Single Circulating Trophoblast (SCT) as one form of cell-based NIPT. <i>PLoS ONE</i> , 2021, 16, e0249695. | 1.1 | 5 |
| 10 | Improving the prenatal diagnosis of Beckwith-Wiedemann syndrome. <i>Prenatal Diagnosis</i> , 2021, 41, 795-797. | 1.1 | 4 |
| 11 | Overview and recent developments in cell-based noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2021, 41, 1202-1214. | 1.1 | 22 |
| 12 | DNA Methylation Dynamics in the Female Germline and Maternal-Effect Mutations That Disrupt Genomic Imprinting. <i>Genes</i> , 2021, 12, 1214. | 1.0 | 24 |
| 13 | Advances in Molecular Genetics Including Fetal Sequencing. , 2020, , 247-253.e1. | | 0 |
| 14 | The current and future impact of genome-wide sequencing on fetal precision medicine. <i>Human Genetics</i> , 2020, 139, 1121-1130. | 1.8 | 20 |
| 15 | 575: The effect of fetal fraction on the development of adverse obstetrical outcomes. <i>American Journal of Obstetrics and Gynecology</i> , 2020, 222, S368-S369. | 0.7 | 0 |
| 16 | The effect of maternal body mass index and gestational age on circulating trophoblast yield in cell-based noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2020, 40, 1383-1389. | 1.1 | 7 |
| 17 | Current controversies in prenatal diagnosis 2: The 59 genes ACMG recommends reporting as secondary findings when sequencing postnatally should be reported when detected on fetal (and) Tj ETQq1 1 0.784314 rgBTj/Overlook | | |
| 18 | Prenatal testing in pregnancies conceived by in vitro fertilization with pre-implantation genetic testing. <i>Prenatal Diagnosis</i> , 2020, 40, 846-851. | 1.1 | 2 |

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|----|--|------|-----------|
| 19 | Prenatally diagnosed developmental abnormalities of the central nervous system and genetic syndromes: A practical review. <i>Prenatal Diagnosis</i> , 2019, 39, 666-678. | 1.1 | 27 |
| 20 | Exome and Genome Sequencing. , 2019, , 137-148. | | 0 |
| 21 | Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316. | 2.6 | 56 |
| 22 | Maternal stress in Shank3ex4-9 mice increases pup-directed care and alters brain white matter in male offspring. <i>PLoS ONE</i> , 2019, 14, e0224876. | 1.1 | 2 |
| 23 | Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. <i>Nature Medicine</i> , 2019, 25, 439-447. | 15.2 | 160 |
| 24 | 930: Clinical utility of non-invasive prenatal screening for common dominant monogenic disorders. <i>American Journal of Obstetrics and Gynecology</i> , 2019, 220, S599. | 0.7 | 1 |
| 25 | The uptake of pan-ethnic expanded carrier screening is higher when offered during preconception or early prenatal genetic counseling. <i>Prenatal Diagnosis</i> , 2019, 39, 319-323. | 1.1 | 22 |
| 26 | 900: A pilot validation study for cell-based noninvasive prenatal testing (NIPT) in 42 cases. <i>American Journal of Obstetrics and Gynecology</i> , 2019, 220, S582-S583. | 0.7 | 0 |
| 27 | Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. <i>American Journal of Human Genetics</i> , 2019, 105, 1262-1273. | 2.6 | 47 |
| 28 | Next-Generation Sequencing for Gene Panels and Clinical Exomes. , 2019, , 553-575. | | 1 |
| 29 | Exome and genome sequencing in reproductive medicine. <i>Fertility and Sterility</i> , 2018, 109, 213-220. | 0.5 | 22 |
| 30 | Missed opportunities: unidentified genetic risk factors in prenatal care. <i>Prenatal Diagnosis</i> , 2018, 38, 75-79. | 1.1 | 9 |
| 31 | Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. <i>Genome Medicine</i> , 2018, 10, 74. | 3.6 | 105 |
| 32 | Reliable detection of subchromosomal deletions and duplications using cell-based noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2018, 38, 1069-1078. | 1.1 | 42 |
| 33 | Causative Mutations and Mechanism of Androgenetic Hydatidiform Moles. <i>American Journal of Human Genetics</i> , 2018, 103, 740-751. | 2.6 | 69 |
| 34 | Prenatal Genetic Testing and Screening. , 2018, , 65-79. | | 0 |
| 35 | Chromosomal microarray analysis on uncultured chorionic villus sampling can be complicated by confined placental mosaicism for aneuploidy and microdeletions. <i>Prenatal Diagnosis</i> , 2018, 38, 858-865. | 1.1 | 17 |
| 36 | Are First Trimester Nuchal Septations Independent Risk Factors for Chromosomal Anomalies?. <i>Journal of Ultrasound in Medicine</i> , 2017, 36, 155-161. | 0.8 | 14 |

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|----|---|-----|-----------|
| 37 | Independent variant analysis of <i>TEAD1</i> and <i>OCEL1</i> in 38 Aicardi syndrome patients. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 117-121. | 0.6 | 6 |
| 38 | Maternally expressed NLRP2 links the subcortical maternal complex (SCMC) to fertility, embryogenesis and epigenetic reprogramming. <i>Scientific Reports</i> , 2017, 7, 44667. | 1.6 | 62 |
| 39 | Positive predictive value estimates for cell-free noninvasive prenatal screening from data of a large referral genetic diagnostic laboratory. <i>American Journal of Obstetrics and Gynecology</i> , 2017, 217, 691.e1-691.e6. | 0.7 | 141 |
| 40 | Genetic screening and testing in pregnancies conceived by in vitro fertilization (IVF) with preimplantation genetic screening (PGS). <i>Fertility and Sterility</i> , 2017, 108, e292. | 0.5 | 3 |
| 41 | A non-mosaic PORCN mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 12, 57-61. | 0.4 | 13 |
| 42 | Evidence for Feasibility of Fetal Trophoblastic Cell-Based Noninvasive Prenatal Testing. <i>Obstetrical and Gynecological Survey</i> , 2017, 72, 4-5. | 0.2 | 0 |
| 43 | Diagnostic Testing Uptake After High-Risk cffDNA Screening [6B]. <i>Obstetrics and Gynecology</i> , 2017, 129, 21S-21S. | 1.2 | 0 |
| 44 | Diagnostic Testing Uptake After High-Risk cffDNA Results for Sex Chromosome Aneuploidy [11B]. <i>Obstetrics and Gynecology</i> , 2017, 129, 22S-22S. | 1.2 | 0 |
| 45 | Comparative Diagnostic Testing Uptake and Confirmation Rates After High-Risk cffDNA for Trisomy 21, 18, and 13 [32N]. <i>Obstetrics and Gynecology</i> , 2017, 129, S151-S151. | 1.2 | 0 |
| 46 | Chronic Maternal Low-Protein Diet in Mice Affects Anxiety, Night-Time Energy Expenditure and Sleep Patterns, but Not Circadian Rhythm in Male Offspring. <i>PLoS ONE</i> , 2017, 12, e0170127. | 1.1 | 23 |
| 47 | Recent advances in prenatal genetic screening and testing. <i>F1000Research</i> , 2016, 5, 2591. | 0.8 | 51 |
| 48 | 648: Missed opportunities: genetic counseling indications in apparent low risk patients referred for first trimester screening. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 214, S345. | 0.7 | 0 |
| 49 | Evidence for feasibility of fetal trophoblastic cell-based noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2016, 36, 1009-1019. | 1.1 | 78 |
| 50 | Comparison of three whole genome amplification methods for detection of genomic aberrations in single cells. <i>Prenatal Diagnosis</i> , 2016, 36, 823-830. | 1.1 | 22 |
| 51 | Genome-wide copy number analysis on DNA from fetal cells isolated from the blood of pregnant women. <i>Prenatal Diagnosis</i> , 2016, 36, 1127-1134. | 1.1 | 68 |
| 52 | 73: Prenatal maternal stress and Shank3ex4-9 mutation alter hippocampal stratum radiatum white matter (WM) in male mice offspring. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 214, S54. | 0.7 | 0 |
| 53 | 575: Pre and perinatal fluoxetine (FLX) exposure of Shank3ex4-9 mutant and WT mice affects neurobehavior in offspring and indicates gene-environment (GXE) interaction. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 214, S309. | 0.7 | 0 |
| 54 | Reproductive genetic counseling challenges associated with diagnostic exome sequencing in a large academic private reproductive genetic counseling practice. <i>Prenatal Diagnosis</i> , 2015, 35, 1022-1029. | 1.1 | 47 |

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|----|--|-----|-----------|
| 55 | Pregnant patients' risk perception of prenatal test results with uncertain fetal clinical significance: ultrasound versus advanced genetic testing. <i>Prenatal Diagnosis</i> , 2015, 35, 1213-1217. | 1.1 | 11 |
| 56 | A genome-wide screen for copy number alterations in an adolescent pilot cohort with m ^ã ¼llerian anomalies. <i>Fertility and Sterility</i> , 2015, 103, 487-493. | 0.5 | 8 |
| 57 | No evidence for mutations in NLRP7, NLRP2 or KHDC3L in women with unexplained recurrent pregnancy loss or infertility. <i>Human Reproduction</i> , 2015, 30, 232-238. | 0.4 | 12 |
| 58 | Noninvasive Prenatal Testing and Fetal Sonographic Screening. <i>Journal of Ultrasound in Medicine</i> , 2015, 34, 363-369. | 0.8 | 4 |
| 59 | Genome-Wide Sequencing for Prenatal Detection of Fetal Single-Gene Disorders. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2015, 5, a023077. | 2.9 | 47 |
| 60 | Counseling Challenges with Variants of Uncertain Significance and Incidental Findings in Prenatal Genetic Screening and Diagnosis. <i>Journal of Clinical Medicine</i> , 2014, 3, 1018-1032. | 1.0 | 59 |
| 61 | A Rare Human Syndrome Provides Genetic Evidence that WNT Signaling Is Required for Reprogramming of Fibroblasts to Induced Pluripotent Stem Cells. <i>Cell Reports</i> , 2014, 9, 1770-1780. | 2.9 | 29 |
| 62 | Prenatal diagnosis of CLOVES syndrome confirmed by detection of a mosaic <i>PIK3CA</i> mutation in cultured amniocytes. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2633-2637. | 0.7 | 26 |
| 63 | Current controversies in prenatal diagnosis 2: should incidental findings arising from prenatal testing always be reported to patients?. <i>Prenatal Diagnosis</i> , 2014, 34, 12-17. | 1.1 | 10 |
| 64 | Do recent US Supreme Court rulings on patenting of genes and genetic diagnostics affect the practice of genetic screening and diagnosis in prenatal and reproductive care?. <i>Prenatal Diagnosis</i> , 2014, 34, 921-926. | 1.1 | 7 |
| 65 | NLRP7 affects trophoblast lineage differentiation, binds to overexpressed YY1 and alters CpG methylation. <i>Human Molecular Genetics</i> , 2014, 23, 706-716. | 1.4 | 54 |
| 66 | Noninvasive Prenatal Screening by Next-Generation Sequencing. <i>Annual Review of Genomics and Human Genetics</i> , 2014, 15, 327-347. | 2.5 | 31 |
| 67 | No evidence for mutations in <i>NLRP7</i> and <i>KHDC3L</i> in women with androgenetic hydatidiform moles. <i>Prenatal Diagnosis</i> , 2013, 33, 1242-1247. | 1.1 | 13 |
| 68 | The PDPK1 gene variants do not seem to be associated with premature ovarian failure. <i>Fertility and Sterility</i> , 2013, 100, S345-S346. | 0.5 | 1 |
| 69 | Genome-Wide Array-Based Copy Number Profiling in Human Placentas From Unexplained Stillbirths. <i>Obstetrical and Gynecological Survey</i> , 2012, 67, 84-86. | 0.2 | 0 |
| 70 | Multiplex ligationâ€dependent probe amplification (MLPA) and prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2012, 32, 315-320. | 1.1 | 26 |
| 71 | Ophthalmologic findings in Aicardi syndrome. <i>Journal of AAPOS</i> , 2012, 16, 238-241. | 0.2 | 31 |
| 72 | Deletion of Porcn in Mice Leads to Multiple Developmental Defects and Models Human Focal Dermal Hypoplasia (Goltz Syndrome). <i>PLoS ONE</i> , 2012, 7, e32331. | 1.1 | 49 |

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|----|--|-----|-----------|
| 73 | Detection of 1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. <i>Prenatal Diagnosis</i> , 2012, 32, 10-20. | 1.1 | 29 |
| 74 | New technologies for the assessment of chromosomes in prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2012, 32, 307-308. | 1.1 | 4 |
| 75 | Prenatal chromosomal microarray analysis in a diagnostic laboratory; experience with >1000 cases and review of the literature. <i>Prenatal Diagnosis</i> , 2012, 32, 351-361. | 1.1 | 103 |
| 76 | 26: Prenatal array comparative genomic hybridization: when is it indicated and what sample is best? Our experience in over 1000 prenatal cases. <i>American Journal of Obstetrics and Gynecology</i> , 2012, 206, S17. | 0.7 | 0 |
| 77 | A Metagenomic Approach to Characterization of the Vaginal Microbiome Signature in Pregnancy. <i>PLoS ONE</i> , 2012, 7, e36466. | 1.1 | 572 |
| 78 | Copy-number changes in prenatal diagnosis. <i>Expert Review of Molecular Diagnostics</i> , 2011, 11, 579-592. | 1.5 | 23 |
| 79 | 716: Evaluation of fetal demise by array comparative genomic hybridization of formalin-fixed paraffin-embedded tissue: a pilot study. <i>American Journal of Obstetrics and Gynecology</i> , 2011, 204, S282. | 0.7 | 1 |
| 80 | Genome-wide array-based copy number profiling in human placentas from unexplained stillbirths. <i>Prenatal Diagnosis</i> , 2011, 31, 932-944. | 1.1 | 23 |
| 81 | Chronic Maternal Protein Deprivation in Mice Is Associated with Overexpression of the Cohesin-Mediator Complex in Liver of Their Offspring. <i>Journal of Nutrition</i> , 2011, 141, 2106-2112. | 1.3 | 8 |
| 82 | Prospective trial on obstructive sleep apnea in pregnancy and fetal heart rate monitoring. <i>American Journal of Obstetrics and Gynecology</i> , 2010, 202, 552.e1-552.e7. | 0.7 | 90 |
| 83 | Terminal osseous dysplasia with pigmentary defects (TODPD): Follow-up of the first reported family, characterization of the radiological phenotype, and refinement of the linkage region. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1825-1831. | 0.7 | 9 |
| 84 | PORCN Mutations and Variants Identified in Patients with Focal Dermal Hypoplasia Through Diagnostic Gene Sequencing. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 709-713. | 0.3 | 13 |
| 85 | Pheochromocytoma and Von Hippel-Lindau in Pregnancy. <i>American Journal of Perinatology</i> , 2010, 27, 257-263. | 0.6 | 10 |
| 86 | Management of Ornithine Transcarbamylase Deficiency in Pregnancy. <i>American Journal of Perinatology</i> , 2010, 27, 775-784. | 0.6 | 44 |
| 87 | Applications of Array Comparative Genomic Hybridization in Obstetrics. <i>Obstetrics and Gynecology Clinics of North America</i> , 2010, 37, 71-85. | 0.7 | 25 |
| 88 | Clinical application of microarray-based comparative genomic hybridization in prenatal diagnosis. <i>Expert Review of Obstetrics and Gynecology</i> , 2009, 4, 81-92. | 0.4 | 1 |
| 89 | 708: Oligo-based array CGH on a single cell - the way toward noninvasive prenatal diagnosis of genomic imbalance. <i>American Journal of Obstetrics and Gynecology</i> , 2009, 201, S256-S257. | 0.7 | 0 |
| 90 | 823: Management of ornithine transcarbamylase deficiency in pregnancy. <i>American Journal of Obstetrics and Gynecology</i> , 2009, 201, S294-S295. | 0.7 | 1 |

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| 91 | A genome-wide screen for copy number alterations in Aicardi syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2113-2121. | 0.7 | 14 |
| 92 | Clinical use of array comparative genomic hybridization (aCGH) for prenatal diagnosis in 300 cases. Prenatal Diagnosis, 2009, 29, 29-39. | 1.1 | 180 |
| 93 | Non-random X chromosome inactivation in Aicardi syndrome. Human Genetics, 2009, 125, 211-216. | 1.8 | 20 |
| 94 | Goltz syndrome: report of two severe cases. BMJ Case Reports, 2009, 2009, bcr0920080909-bcr0920080909. | 0.2 | 1 |
| 95 | Neuroimaging aspects of Aicardi syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2871-2878. | 0.7 | 74 |
| 96 | 441: Prospective trial on obstructive sleep apnea (OSA) in pregnancy and fetal heart rate monitoring. American Journal of Obstetrics and Gynecology, 2008, 199, S132. | 0.7 | 0 |
| 97 | 602: Genome-wide array-based copy number profiling in placentas from unexplained stillbirth. American Journal of Obstetrics and Gynecology, 2008, 199, S174. | 0.7 | 0 |
| 98 | 607: The search for reliable prognostic factors in fetal gastroschisis. American Journal of Obstetrics and Gynecology, 2008, 199, S175. | 0.7 | 0 |
| 99 | 685: Prenatal detection of genomic imbalances in six days from uncultured amniocytes by targeted oligonucleotide array CGH. American Journal of Obstetrics and Gynecology, 2008, 199, S195. | 0.7 | 0 |
| 100 | A recurrent intragenic genomic duplication, other novel mutations in NLRP7 and imprinting defects in recurrent biparental hydatidiform moles. Molecular Human Reproduction, 2008, 14, 33-40. | 1.3 | 114 |
| 101 | Phenotype and Management of Aicardi Syndrome: New Findings from a Survey of 69 Children. Journal of Child Neurology, 2007, 22, 176-184. | 0.7 | 41 |
| 102 | Mutations in X-linked PORCN, a putative regulator of Wnt signaling, cause focal dermal hypoplasia. Nature Genetics, 2007, 39, 836-838. | 9.4 | 250 |
| 103 | 601: Rapid prenatal diagnosis of cytogenetic abnormalities by array CGH analysis. American Journal of Obstetrics and Gynecology, 2007, 197, S173. | 0.7 | 1 |
| 104 | Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. Genetics in Medicine, 2006, 8, 719-727. | 1.1 | 154 |
| 105 | Comparative genomic hybridization and prenatal diagnosis. Current Opinion in Obstetrics and Gynecology, 2006, 18, 185-191. | 0.9 | 48 |
| 106 | Expression profiling of clonal lymphocyte cell cultures from Rett syndrome patients. BMC Medical Genetics, 2006, 7, 61. | 2.1 | 34 |
| 107 | Biparental hydatidiform moles: a maternal effect mutation affecting imprinting in the offspring. Human Reproduction Update, 2006, 12, 233-242. | 5.2 | 64 |
| 108 | Newborn Screening and Prenatal Diagnosis for Rett Syndrome: Implications for Therapy. Journal of Child Neurology, 2005, 20, 779-783. | 0.7 | 14 |

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|-----|---|-----|-----------|
| 109 | Facial and physical features of Aicardi syndrome: Infants to teenagers. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 254-258. | 0.7 | 52 |
| 110 | Tooth Enamel Defects in Mice with a Deletion at the <i>Arhgap6/AmelX</i> Locus. <i>Calcified Tissue International</i> , 2005, 77, 23-29. | 1.5 | 17 |
| 111 | Recurrent Biparental Hydatidiform Mole: Additional Evidence for a 1.1-Mb Locus in 19q13.4 and Candidate Gene Analysis. <i>Journal of the Society for Gynecologic Investigation</i> , 2005, 12, 376-383. | 1.9 | 24 |
| 112 | Mutations in exon 1 of <i>MECP2</i> are a rare cause of Rett syndrome. <i>Journal of Medical Genetics</i> , 2005, 42, e15-e15. | 1.5 | 50 |
| 113 | A NotI/EcoRV promoter library for studies of genetic and epigenetic alterations in mouse models of human malignancies. <i>Genomics</i> , 2004, 84, 647-660. | 1.3 | 25 |
| 114 | Presence of filamin in the astrocytic inclusions of Aicardi syndrome. <i>Pediatric Neurology</i> , 2004, 30, 7-15. | 1.0 | 32 |
| 115 | The Product of the Imprinted Gene <i>IPL</i> Marks Human Villous Cytotrophoblast and is Lost in Complete Hydatidiform Mole. <i>Placenta</i> , 2003, 24, 835-842. | 0.7 | 68 |
| 116 | Recurrent pregnancy loss due to familial and non-familial habitual molar pregnancy. <i>International Journal of Gynecology and Obstetrics</i> , 2003, 83, 179-186. | 1.0 | 22 |
| 117 | GENETICEFFECTS OF METHYLATION DIETS. <i>Annual Review of Nutrition</i> , 2002, 22, 255-282. | 4.3 | 191 |
| 118 | Microphthalmia with linear skin defects (MLS), Aicardi, and Goltz syndromes: are they related X-linked dominant male-lethal disorders?. <i>Cytogenetic and Genome Research</i> , 2002, 99, 289-296. | 0.6 | 36 |
| 119 | Loss of holocytochrome c-type synthetase causes the male lethality of X-linked dominant micro-phthmia with linear skin defects (MLS) syndrome. <i>Human Molecular Genetics</i> , 2002, 11, 3237-3248. | 1.4 | 58 |
| 120 | Genetic basis of rett syndrome. <i>Mental Retardation and Developmental Disabilities Research Reviews</i> , 2002, 8, 82-86. | 3.5 | 44 |
| 121 | Mutations in the gene encoding methyl-CpG-binding protein 2 cause Rett syndrome. <i>Brain and Development</i> , 2001, 23, S147-S151. | 0.6 | 49 |
| 122 | Analysis of <i>Mid1</i> , <i>Hccs</i> , <i>Arhgap6</i> , and <i>Msl3l1</i> in X-linked polydactyly (<i>Xpl</i>) and Patchy-fur (<i>Paf</i>) mutant mice. <i>Mammalian Genome</i> , 2001, 12, 796-798. | 1.0 | 6 |
| 123 | Skewed X Inactivation in X-Linked Disorders. <i>Seminars in Reproductive Medicine</i> , 2001, 19, 183-192. | 0.5 | 99 |
| 124 | The human homologue (<i>PEG3</i>) of the mouse paternally expressed gene 3 (<i>Peg3</i>) is maternally imprinted but not mutated in women with familial recurrent hydatidiform molar pregnancies. <i>Journal of the Society for Gynecologic Investigation</i> , 2001, 8, 305-313. | 1.9 | 7 |
| 125 | Terminal osseous dysplasia and pigmentary defects: Clinical characterization of a novel male lethal X-linked syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000, 94, 102-112. | 2.4 | 25 |
| 126 | Influence of mutation type and X chromosome inactivation on Rett syndrome phenotypes. <i>Annals of Neurology</i> , 2000, 47, 670-679. | 2.8 | 314 |

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|-----|---|-----|-----------|
| 127 | Terminal Osseous Dysplasia with Pigmentary Defects Maps to Human Chromosome Xq27.3-Xqter. American Journal of Human Genetics, 2000, 66, 1461-1464. | 2.6 | 15 |
| 128 | Methyl-CpG-binding protein 2 mutations in Rett syndrome. Current Opinion in Genetics and Development, 2000, 10, 275-279. | 1.5 | 101 |
| 129 | Influence of mutation type and X chromosome inactivation on Rett syndrome phenotypes. Annals of Neurology, 2000, 47, 670-679. | 2.8 | 12 |
| 130 | Discordance between Fetal RhD Typing Using Molecular Methods and Neonatal Typing with Serology. Gynecologic and Obstetric Investigation, 1999, 48, 229-231. | 0.7 | 3 |
| 131 | Genetic Mapping of a Maternal Locus Responsible for Familial Hydatidiform Moles. Human Molecular Genetics, 1999, 8, 667-667. | 1.4 | 130 |
| 132 | Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. Nature Genetics, 1999, 23, 185-188. | 9.4 | 4,459 |
| 133 | Characterization of a Novel Chromo Domain Gene in Xp22.3 with Homology to Drosophila msl-3. Genomics, 1999, 59, 77-84. | 1.3 | 28 |
| 134 | Genomic structure of a human holocytochrome-c-type synthetase gene in Xp22.3 and mutation analysis in patients with Rett syndrome. American Journal of Medical Genetics Part A, 1998, 78, 179-181. | 2.4 | 10 |
| 135 | Characterization and Physical Mapping in Human and Mouse of a Novel RING Finger Gene in Xp22. Genomics, 1998, 51, 251-261. | 1.3 | 26 |
| 136 | Detection of Intrauterine Viral Infection Using the Polymerase Chain Reaction. Molecular Genetics and Metabolism, 1998, 63, 85-95. | 0.5 | 81 |
| 137 | Applied molecular genetic techniques for prenatal diagnosis. Current Opinion in Obstetrics and Gynecology, 1998, 10, 97-103. | 0.9 | 6 |
| 138 | Prenatal Diagnosis of the RhD Fetal Blood Type on Amniotic Fluid by Polymerase Chain Reaction. Obstetrics and Gynecology, 1996, 87, 419-422. | 1.2 | 29 |
| 139 | Fetal RhD typing by polymerase chain reaction in pregnancies complicated by rhesus alloimmunization. Obstetrics and Gynecology, 1996, 88, 1061-1067. | 1.2 | 42 |
| 140 | Single-cell analysis of the RhD blood type for use in preimplantation diagnosis in the prevention of severe hemolytic disease of the newborn. American Journal of Obstetrics and Gynecology, 1995, 172, 533-540. | 0.7 | 37 |
| 141 | Cerebral Vasospasm in Eclampsia: Transcranial Doppler Ultrasound Findings. Journal of Maternal-Fetal and Neonatal Medicine, 1994, 3, 9-13. | 0.7 | 14 |
| 142 | Preimplantation single cell analyses of dystrophin gene deletions using whole genome amplification. Nature Genetics, 1994, 6, 19-23. | 9.4 | 106 |
| 143 | Molecular analysis of human platelet antigen system 1 antigen on single cells can be applied to preimplantation genetic diagnosis for prevention of alloimmune thrombocytopenia. American Journal of Obstetrics and Gynecology, 1994, 170, 807-812. | 0.7 | 13 |
| 144 | Prenatal diagnosis and clinical findings in a case of hexasomy 12p. American Journal of Medical Genetics Part A, 1993, 47, 1171-1174. | 2.4 | 8 |

| # | ARTICLE | IF | CITATIONS |
|-----|--|-----|-----------|
| 145 | The Effect of Gestational Age and Fetal Indomethacin Levels on the Incidence of Constriction of the Fetal Ductus Arteriosus. <i>Obstetrics and Gynecology</i> , 1993, 82, 500-503. | 1.2 | 5 |
| 146 | Prostaglandin Synthetase Inhibitors in Pregnancy. <i>Obstetrical and Gynecological Survey</i> , 1993, 48, 493-502. | 0.2 | 65 |
| 147 | Circulating trophoblast numbers as a potential marker for pregnancy complications. <i>Prenatal Diagnosis</i> , 0, , . | 1.1 | 3 |
| 148 | Lack of consensus among healthcare professionals at a large academic medical center on the use of exome sequencing for prenatal diagnosis. <i>Journal of Genetic Counseling</i> , 0, , . | 0.9 | 0 |