

# Joris Robert Vermeesch

## List of Publications by Year in descending order

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Version: 2024-02-01

170  
papers

11,746  
citations

43973

48  
h-index

31759

101  
g-index

181  
all docs

181  
docs citations

181  
times ranked

15135  
citing authors

#	ARTICLE	IF	CITATIONS
1	A normative chart for cognitive development in a genetically selected population. <i>Neuropsychopharmacology</i> , 2022, 47, 1379-1386.	2.8	12
2	Optimizing the diagnostic workflow for acute lymphoblastic leukemia by optical genome mapping. <i>American Journal of Hematology</i> , 2022, 97, 548-561.	2.0	36
3	Single-cell genome-wide concurrent haplotyping and copy-number profiling through genotyping-by-sequencing. <i>Nucleic Acids Research</i> , 2022, 50, e63-e63.	6.5	17
4	OUP accepted manuscript. <i>Clinical Chemistry</i> , 2022, 68, 634.	1.5	0
5	A review of normative documents on preimplantation genetic testing: Recommendations for PGT-P. <i>Genetics in Medicine</i> , 2022, 24, 1165-1175.	1.1	9
6	The Hunt for the Chromosome 22q11.2 Deletion Syndrome Schizophrenia Genes. <i>Biological Psychiatry</i> , 2022, 91, 692-693.	0.7	1
7	Primary mediastinal large B-cell lymphoma is characterized by large-scale copy-neutral loss of heterozygosity. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 603-615.	1.5	2
8	Augmenting Therapeutic Effectiveness Through Novel Analytics (ATHENA) – A Public and Private Partnership Project Funded by the Flemish Government (VLAIO). <i>Studies in Health Technology and Informatics</i> , 2022, , .	0.2	0
9	Pan-Cancer Detection and Typing by Mining Patterns in Large Genome-Wide Cell-Free DNA Sequencing Datasets. <i>Clinical Chemistry</i> , 2022, 68, 1164-1176.	1.5	6
10	International Society for Prenatal Diagnosis Position Statement: cell free (cf)DNA screening for Down syndrome in multiple pregnancies. <i>Prenatal Diagnosis</i> , 2021, 41, 1222-1232.	1.1	41
11	<i>MSH2</i> knock-down shows CTG repeat stability and concomitant upstream demethylation at the <i>DMPK</i> locus in myotonic dystrophy type 1 human embryonic stem cells. <i>Human Molecular Genetics</i> , 2021, 29, 3566-3577.	1.4	4
12	Ultra-low coverage whole genome sequencing of cfDNA in multiple myeloma: A tool for laboratory routine?. <i>Cancer Treatment and Research Communications</i> , 2021, 28, 100380.	0.7	3
13	Genome-wide abnormalities in embryos: Origins and clinical consequences. <i>Prenatal Diagnosis</i> , 2021, 41, 554-563.	1.1	9
14	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. <i>Genetics in Medicine</i> , 2021, 23, 1137-1142.	1.1	58
15	A placental trisomy 2 detected by NIPT evolved in a fetal small Supernumerary Marker Chromosome (sSMC). <i>Molecular Cytogenetics</i> , 2021, 14, 18.	0.4	2
16	The landscape of copy number variations in classical Hodgkin lymphoma: a joint KU Leuven and LYSA study on cell-free DNA. <i>Blood Advances</i> , 2021, 5, 1991-2002.	2.5	15
17	Performance and Diagnostic Value of Genome-Wide Noninvasive Prenatal Testing in Multiple Gestations. <i>Obstetrics and Gynecology</i> , 2021, 137, 1102-1108.	1.2	10
18	Comprehensive genome-wide analysis of routine non-invasive test data allows cancer prediction: A single-center retrospective analysis of over 85,000 pregnancies. <i>EClinicalMedicine</i> , 2021, 35, 100856.	3.2	42

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19	22q11.2 Low Copy Repeats Expanded in the Human Lineage. <i>Frontiers in Genetics</i> , 2021, 12, 706641.	1.1	11
20	Noninvasive prenatal testing suggesting a maternal malignancy: What do we tell the prospective parents in Belgium?. <i>Prenatal Diagnosis</i> , 2021, 41, 1264-1272.	1.1	20
21	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. <i>Molecular Psychiatry</i> , 2021, 26, 4496-4510.	4.1	87
22	Haplotyping-based preimplantation genetic testing reveals parent-of-origin specific mechanisms of aneuploidy formation. <i>Npj Genomic Medicine</i> , 2021, 6, 81.	1.7	21
23	A benchmark of structural variation detection by long reads through a realistic simulated model. <i>Genome Biology</i> , 2021, 22, 342.	3.8	21
24	Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. <i>PLoS Computational Biology</i> , 2021, 17, e1009684.	1.5	6
25	Detection of incipient tumours by screening of circulating plasma DNA: hype or hope?. <i>Acta Clinica Belgica</i> , 2020, 75, 9-18.	0.5	9
26	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. <i>Human Mutation</i> , 2020, 41, 449-464.	1.1	17
27	Pathogenic variants in <i>CDC45</i> on the remaining allele in patients with a chromosome 22q11.2 deletion result in a novel autosomal recessive condition. <i>Genetics in Medicine</i> , 2020, 22, 326-335.	1.1	17
28	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. <i>American Journal of Human Genetics</i> , 2020, 106, 26-40.	2.6	42
29	Extreme enrichment of VNTR-associated polymorphicity in human subtelomeres: genes with most VNTRs are predominantly expressed in the brain. <i>Translational Psychiatry</i> , 2020, 10, 369.	2.4	15
30	Genotype-phenotype correlations of <i>UBA2</i> mutations in patients with ectrodactyly. <i>European Journal of Medical Genetics</i> , 2020, 63, 104009.	0.7	4
31	Breast Cancer Detection and Treatment Monitoring Using a Noninvasive Prenatal Testing Platform: Utility in Pregnant and Nonpregnant Populations. <i>Clinical Chemistry</i> , 2020, 66, 1414-1423.	1.5	9
32	De Novo Variants in <i>LMNB1</i> Cause Pronounced Syndromic Microcephaly and Disruption of Nuclear Envelope Integrity. <i>American Journal of Human Genetics</i> , 2020, 107, 753-762.	2.6	30
33	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. <i>Nature Medicine</i> , 2020, 26, 1912-1918.	15.2	90
34	Current use of noninvasive prenatal testing in Europe, Australia and the USA: A graphical presentation. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2020, 99, 722-730.	1.3	121
35	Identity-by-state-based haplotyping expands the application of comprehensive preimplantation genetic testing. <i>Human Reproduction</i> , 2020, 35, 718-726.	0.4	6
36	Noninvasive prenatal diagnosis by genome-wide haplotyping of cell-free plasma DNA. <i>Genetics in Medicine</i> , 2020, 22, 962-973.	1.1	29

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37	<i>LEF1</i> haploinsufficiency causes ectodermal dysplasia. <i>Clinical Genetics</i> , 2020, 97, 595-600.	1.0	11
38	Ultra-low depth sequencing of plasma cell DNA for the detection of copy number aberrations in multiple myeloma. <i>Genes Chromosomes and Cancer</i> , 2020, 59, 465-471.	1.5	3
39	The clinical relevance of intragenic NRXN1 deletions. <i>Journal of Medical Genetics</i> , 2020, 57, 347-355.	1.5	11
40	Prenatally detected copy number variants in a national cohort: A postnatal follow-up study. <i>Prenatal Diagnosis</i> , 2020, 40, 1272-1283.	1.1	5
41	PREIMPLANTATION GENETIC TESTING: Single-cell technologies at the forefront of PGT and embryo research. <i>Reproduction</i> , 2020, 160, A19-A31.	1.1	10
42	Opportunities of Genome Imaging for Genetic Diagnosis in Acute Lymphoblastic Leukemia. <i>Blood</i> , 2020, 136, 10-11.	0.6	0
43	Pregnant women with confirmed neoplasms should not have noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2019, 39, 1162-1165.	1.1	17
44	Fetal sex determination in twin pregnancies using non-invasive prenatal testing. <i>Npj Genomic Medicine</i> , 2019, 4, 15.	1.7	10
45	Multi-centre evaluation of a comprehensive preimplantation genetic test through haplotyping-by-sequencing. <i>Human Reproduction</i> , 2019, 34, 1608-1619.	0.4	48
46	Noninvasive Prenatal Testing and Detection of Occult Maternal Malignancies. <i>Clinical Chemistry</i> , 2019, 65, 1484-1486.	1.5	19
47	In vitro fertilization does not increase the incidence of de novo copy number alterations in fetal and placental lineages. <i>Nature Medicine</i> , 2019, 25, 1699-1705.	15.2	43
48	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. <i>Genome Research</i> , 2019, 29, 1389-1401.	2.4	39
49	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. <i>Genetics in Medicine</i> , 2019, 21, 2774-2780.	1.1	16
50	The sudden death of the combined first trimester aneuploidy screening, a single centre experience in Belgium. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, e294-e297.	1.4	4
51	Preeclampsia is Associated with Sex-Specific Transcriptional and Proteomic Changes in Fetal Erythroid Cells. <i>International Journal of Molecular Sciences</i> , 2019, 20, 2038.	1.8	16
52	Current Controversies in Prenatal Diagnosis 3: Gene editing should replace embryo selection following PGD. <i>Prenatal Diagnosis</i> , 2019, 39, 344-350.	1.1	8
53	Diagnostic implications of genetic copy number variation in epilepsy plus. <i>Epilepsia</i> , 2019, 60, 689-706.	2.6	61
54	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. <i>Journal of Neurodevelopmental Disorders</i> , 2019, 11, 3.	1.5	6

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55	Non-Syndromic Cleft Lip with or without Cleft Palate: Genome-Wide Association Study in Europeans Identifies a Suggestive Risk Locus at 16p12.1 and Supports SH3PXD2A as a Clefing Susceptibility Gene. <i>Genes</i> , 2019, 10, 1023.	1.0	26
56	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. <i>Human Molecular Genetics</i> , 2019, 28, 3724-3733.	1.4	7
57	A speculative outlook on embryonic aneuploidy: Can molecular pathways be involved?. <i>Developmental Biology</i> , 2019, 447, 3-13.	0.9	29
58	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. <i>Molecular Psychiatry</i> , 2019, 24, 1748-1768.	4.1	26
59	NIPTmer: rapid k-mer-based software package for detection of fetal aneuploidies. <i>Scientific Reports</i> , 2018, 8, 5616.	1.6	12
60	Single molecule real-time (SMRT) sequencing comes of age: applications and utilities for medical diagnostics. <i>Nucleic Acids Research</i> , 2018, 46, 2159-2168.	6.5	518
61	Predicting fetoplacental chromosomal mosaicism during noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2018, 38, 258-266.	1.1	58
62	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. <i>Human Molecular Genetics</i> , 2018, 27, 1150-1163.	1.4	22
63	Congenital diaphragmatic hernia as a part of Nance-Horan syndrome?. <i>European Journal of Human Genetics</i> , 2018, 26, 359-366.	1.4	7
64	Conventional and Single-Molecule Targeted Sequencing Method for Specific Variant Detection in IKBKG while Bypassing the IKBKG1 Pseudogene. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 195-202.	1.2	26
65	Noise-robust assessment of SNP array based CNV calls through local noise estimation of log R ratios. <i>Statistical Applications in Genetics and Molecular Biology</i> , 2018, 17, .	0.2	0
66	Novel CASK mutations in cases with syndromic microcephaly. <i>Human Mutation</i> , 2018, 39, 993-1001.	1.1	16
67	Neurodevelopmental risk copy number variants in adults with intellectual disabilities and comorbid psychiatric disorders. <i>British Journal of Psychiatry</i> , 2018, 212, 287-294.	1.7	30
68	Clinical characteristics of patients with low functional IL-6 production upon TLR/IL-1R stimulation. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 768-770.	1.5	0
69	Improved reference genome for the domestic horse increases assembly contiguity and composition. <i>Communications Biology</i> , 2018, 1, 197.	2.0	148
70	Mapping the landscape of tandem repeat variability by targeted long read single molecule sequencing in familial X-linked intellectual disability. <i>BMC Medical Genomics</i> , 2018, 11, 123.	0.7	5
71	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2172-2181.	0.7	33
72	Genome-wide haplotyping embryos developing from OPN and 1PN zygotes increases transferrable embryos in PGT-M. <i>Human Reproduction</i> , 2018, 33, 2302-2311.	0.4	33

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73	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. <i>Prenatal Diagnosis</i> , 2018, 38, 1120-1128.	1.1	24
74	Molecular genetics of 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2070-2081.	0.7	96
75	Efficient CRISPR/Cas9-mediated editing of trinucleotide repeat expansion in myotonic dystrophy patient-derived iPSC and myogenic cells. <i>Nucleic Acids Research</i> , 2018, 46, 8275-8298.	6.5	78
76	Genetic profile of isolated congenital diaphragmatic hernia revealed by targeted next-generation sequencing. <i>Prenatal Diagnosis</i> , 2018, 38, 654-663.	1.1	31
77	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. <i>Frontiers in Genetics</i> , 2018, 9, 150.	1.1	26
78	Response to a comment on "Predicting fetoplacental chromosomal mosaicism during non-invasive prenatal testing". <i>Prenatal Diagnosis</i> , 2018, 38, 722-723.	1.1	1
79	The genetic structure of the Belgian population. <i>Human Genomics</i> , 2018, 12, 6.	1.4	7
80	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1897-1909.	0.7	7
81	Unraveling the Landscape of Copy Number Aberrations in Hodgkin Lymphoma: A Joint KU Leuven and Lysa Study on Circulating Cell Free DNA. <i>Blood</i> , 2018, 132, 2836-2836.	0.6	0
82	Mutational Processes Shaping the Genome in Early Human Embryos. <i>Cell</i> , 2017, 168, 751-753.	13.5	9
83	How can zygotes segregate entire parental genomes into distinct blastomeres? The zygote metaphase revisited. <i>BioEssays</i> , 2017, 39, 1600226.	1.2	11
84	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. <i>European Journal of Human Genetics</i> , 2017, 25, 694-701.	1.4	33
85	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. <i>Human Reproduction</i> , 2017, 32, 687-697.	0.4	40
86	Low-pass Sequencing of Plasma Cell DNA and of ccfDNA for the Detection of Copy Number Aberrations and Early Response Monitoring in Multiple Myeloma. <i>Clinical Lymphoma, Myeloma and Leukemia</i> , 2017, 17, e95.	0.2	0
87	An incidental finding of maternal multiple myeloma by non invasive prenatal testing. <i>Prenatal Diagnosis</i> , 2017, 37, 1257-1260.	1.1	13
88	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the <i>GPR98</i> Locus on 5q14.3. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	22
89	Genome stability of bovine in vivo-conceived cleavage-stage embryos is higher compared to in vitro-produced embryos. <i>Human Reproduction</i> , 2017, 32, 2348-2357.	0.4	69
90	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. <i>American Journal of Psychiatry</i> , 2017, 174, 1054-1063.	4.0	77

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91	Accuracy and Clinical Value of Maternal Incidental Findings During Noninvasive Prenatal Testing for Fetal Aneuploidies. <i>Obstetrical and Gynecological Survey</i> , 2017, 72, 469-470.	0.2	1
92	Array CGH. <i>Springer Protocols</i> , 2017, , 567-586.	0.1	1
93	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. <i>Genetics in Medicine</i> , 2017, 19, 306-313.	1.1	47
94	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. <i>Human Mutation</i> , 2017, 38, 324-331.	1.1	37
95	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. <i>Genome Medicine</i> , 2017, 9, 67.	3.6	29
96	Maternal vitamin B12 deficiency and abnormal cell-free DNA results in pregnancy. <i>Prenatal Diagnosis</i> , 2016, 36, 790-793.	1.1	17
97	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. <i>Human Mutation</i> , 2016, 37, 661-668.	1.1	30
98	Zygotes segregate entire parental genomes in distinct blastomere lineages causing cleavage-stage chimerism and mixoploidy. <i>Genome Research</i> , 2016, 26, 567-578.	2.4	73
99	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. <i>European Journal of Medical Genetics</i> , 2016, 59, 436-443.	0.7	20
100	Current controversies in prenatal diagnosis 3: industry drives innovation in research and clinical application of genetic prenatal diagnosis and screening. <i>Prenatal Diagnosis</i> , 2016, 36, 1172-1177.	1.1	9
101	Prenatal and pre-implantation genetic diagnosis. <i>Nature Reviews Genetics</i> , 2016, 17, 643-656.	7.7	155
102	Variant discovery and breakpoint region prediction for studying the human 22q11.2 deletion using BAC clone and whole genome sequencing analysis. <i>Human Molecular Genetics</i> , 2016, 25, 3754-3767.	1.4	20
103	A catalog of hemizygous variation in 127 22q11 deletion patients. <i>Human Genome Variation</i> , 2016, 3, 15065.	0.4	8
104	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. <i>Human Mutation</i> , 2016, 37, 804-811.	1.1	92
105	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. <i>European Journal of Human Genetics</i> , 2016, 24, 51-58.	1.4	10
106	Polymerase specific error rates and profiles identified by single molecule sequencing. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2016, 784-785, 39-45.	0.4	44
107	Pulmonary transcriptome analysis in the rabbit model of surgically-induced diaphragmatic hernia treated with fetal tracheal occlusion. <i>DMM Disease Models and Mechanisms</i> , 2016, 9, 221-8.	1.2	18
108	22q11.2 deletion syndrome. <i>Nature Reviews Disease Primers</i> , 2015, 1, 15071.	18.1	954

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109	Genotypic and phenotypic variation in six patients with solitary median maxillary central incisor syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2451-2458.	0.7	21
110	Concurrent Whole-Genome Haplotyping and Copy-Number Profiling of Single Cells. <i>American Journal of Human Genetics</i> , 2015, 96, 894-912.	2.6	110
111	Presymptomatic Identification of Cancers in Pregnant Women During Noninvasive Prenatal Testing. <i>JAMA Oncology</i> , 2015, 1, 814.	3.4	180
112	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	1.4	85
113	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. <i>American Journal of Human Genetics</i> , 2015, 97, 790-800.	2.6	63
114	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. <i>Brain and Development</i> , 2015, 37, 527-536.	0.6	32
115	Non-invasive detection of genomic imbalances in Hodgkin/Reed-Sternberg cells in early and advanced stage Hodgkin's lymphoma by sequencing of circulating cell-free DNA: a technical proof-of-principle study. <i>Lancet Haematology</i> , 2015, 2, e55-e65.	2.2	115
116	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. <i>European Journal of Human Genetics</i> , 2015, 23, 1286-1293.	1.4	108
117	GBSX: a toolkit for experimental design and demultiplexing genotyping by sequencing experiments. <i>BMC Bioinformatics</i> , 2015, 16, 73.	1.2	102
118	Expanding the phenotypic spectrum of PORCN variants in two males with syndromic microphthalmia. <i>European Journal of Human Genetics</i> , 2015, 23, 551-554.	1.4	24
119	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1822-1829.	0.7	48
120	Copy Number Variation Analysis by Array Analysis of Single Cells Following Whole Genome Amplification. <i>Methods in Molecular Biology</i> , 2015, 1347, 197-219.	0.4	6
121	Lack of Evidence That Male Fetal Microchimerism is Present in Endometriosis. <i>Reproductive Sciences</i> , 2015, 22, 1115-1121.	1.1	3
122	NGS-Logistics: federated analysis of NGS sequence variants across multiple locations. <i>Genome Medicine</i> , 2014, 6, 71.	3.6	16
123	Pseudoautosomal Region 1 Length Polymorphism in the Human Population. <i>PLoS Genetics</i> , 2014, 10, e1004578.	1.5	24
124	Prenatal management of the fetus with isolated congenital diaphragmatic hernia in the era of the TOTAL trial. <i>Seminars in Fetal and Neonatal Medicine</i> , 2014, 19, 338-348.	1.1	149
125	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. <i>Human Reproduction</i> , 2014, 29, 1603-1609.	0.4	57
126	DNA from Nails for Genetic Analyses in Large-Scale Epidemiologic Studies. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2014, 23, 2703-2712.	1.1	27



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127	Single cell segmental aneuploidy detection is compromised by S phase. <i>Molecular Cytogenetics</i> , 2014, 7, 46.	0.4	23
128	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. <i>European Journal of Medical Genetics</i> , 2014, 57, 151-156.	0.7	91
129	A prospective study of the clinical utility of prenatal chromosomal microarray analysis in fetuses with ultrasound abnormalities and an exploration of a framework for reporting unclassified variants and risk factors. <i>Genetics in Medicine</i> , 2014, 16, 469-476.	1.1	66
130	Exome sequencing identifies ZFPM2 as a cause of familial isolated congenital diaphragmatic hernia and possibly cardiovascular malformations. <i>European Journal of Medical Genetics</i> , 2014, 57, 247-252.	0.7	17
131	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 173-182.	2.6	219
132	Large-scale analysis of tandem repeat variability in the human genome. <i>Nucleic Acids Research</i> , 2014, 42, 5728-5741.	6.5	66
133	Single-cell paired-end genome sequencing reveals structural variation per cell cycle. <i>Nucleic Acids Research</i> , 2013, 41, 6119-6138.	6.5	142
134	Preimplantation genetic diagnosis guided by single-cell genomics. <i>Genome Medicine</i> , 2013, 5, 71.	3.6	45
135	Nonallelic homologous recombination between retrotransposable elements is a driver of de novo unbalanced translocations. <i>Genome Research</i> , 2013, 23, 411-418.	2.4	90
136	The Future of Prenatal Cytogenetics: From Copy Number Variations to Non-invasive Prenatal Testing. <i>Current Genetic Medicine Reports</i> , 2013, 1, 91-98.	1.9	1
137	Aneuploidy and Copy Number Variation in Early Human Development. <i>Seminars in Reproductive Medicine</i> , 2012, 30, 302-308.	0.5	10
138	Chromosome Instability Is Common in Human Cleavage-Stage Embryos. <i>Obstetrical and Gynecological Survey</i> , 2012, 67, 787-788.	0.2	1
139	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. <i>Genome Medicine</i> , 2012, 4, 73.	3.6	28
140	New array approaches to explore single cells genomes. <i>Frontiers in Genetics</i> , 2012, 3, 44.	1.1	20
141	Cytogenetic and morphological analysis of early products of conception following hysteroembryoscopy from couples with recurrent pregnancy loss. <i>Prenatal Diagnosis</i> , 2012, 32, 933-942.	1.1	45
142	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. <i>Human Mutation</i> , 2012, 33, 906-915.	1.1	69
143	Array-Based Approaches in Prenatal Diagnosis. <i>Methods in Molecular Biology</i> , 2012, 838, 151-171.	0.4	13
144	Breakage-fusion-bridge cycles leading to inv dup del occur in human cleavage stage embryos. <i>Human Mutation</i> , 2011, 32, 783-793.	1.1	60

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145	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2010, 86, 749-764.	2.6	2,325
146	Diagnosis of miscarriages by molecular karyotyping: Benefits and pitfalls. <i>Genetics in Medicine</i> , 2009, 11, 646-654.	1.1	90
147	What next for preimplantation genetic screening? High mitotic chromosome instability rate provides the biological basis for the low success rate. <i>Human Reproduction</i> , 2009, 24, 2679-2682.	0.4	87
148	Chromosome instability is common in human cleavage-stage embryos. <i>Nature Medicine</i> , 2009, 15, 577-583.	15.2	710
149	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
150	Autosomal-Dominant Microtia Linked to Five Tandem Copies of a Copy-Number-Variable Region at Chromosome 4p16. <i>American Journal of Human Genetics</i> , 2008, 82, 181-187.	2.6	42
151	Submicroscopic chromosomal imbalances detected by array-CGH are a frequent cause of congenital heart defects in selected patients. <i>European Heart Journal</i> , 2007, 28, 2778-2784.	1.0	175
152	Subtelomeric imbalances in phenotypically normal individuals. <i>Human Mutation</i> , 2007, 28, 958-967.	1.1	72
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