Joris Robert Vermeesch

List of Publications by Year
in descending order

170 papers	11,746 citations	44066 48 h-index	31843 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. Neuropsychopharmacology, 2022, 47, 1379-1386.	5.4	12
2	Optimizing the diagnostic workflow for acute lymphoblastic leukemia by optical genome mapping. American Journal of Hematology, 2022, 97, 548-561.	4.1	36
3	Single-cell genome-wide concurrent haplotyping and copy-number profiling through genotyping-by-sequencing. Nucleic Acids Research, 2022, 50, e63-e63.	14.5	17
4	OUP accepted manuscript. Clinical Chemistry, 2022, 68, 634.	3.2	0
5	A review of normative documents on preimplantation genetic testing: Recommendations for PGT-P. Genetics in Medicine, 2022, 24, 1165-1175.	2.4	9
6	The Hunt for the Chromosome 22q11.2 Deletion Syndrome Schizophrenia Genes. Biological Psychiatry, 2022, 91, 692-693.	1.3	1
7	Primary mediastinal large Bâ€cell lymphoma is characterized by largeâ€scale copyâ€neutral loss of heterozygosity. Genes Chromosomes and Cancer, 2022, 61, 603-615.	2.8	2
8	Augmenting THerapeutic Effectiveness Through Novel Analytics (ATHENA) â€“ A Public and Private Partnership Project Funded by the Flemish Government (VLAIO). Studies in Health Technology and Informatics, 2022, , .	0.3	0
9	Pan-Cancer Detection and Typing by Mining Patterns in Large Genome-Wide Cell-Free DNA Sequencing Datasets. Clinical Chemistry, 2022, 68, 1164-1176.	3.2	6
10	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.	2.3	41
11	MSH2knock-down shows CTG repeat stability and concomitant upstream demethylation at theDMPKlocus in myotonic dystrophy type 1 human embryonic stem cells. Human Molecular Genetics, 2021, 29, 3566-3577.	2.9	4
12	Ultra-low coverage whole genome sequencing of ccfDNA in multiple myeloma: A tool for laboratory routine?. Cancer Treatment and Research Communications, 2021, 28, 100380.	1.7	3
13	Genomeâ€wide abnormalities in embryos: Origins and clinical consequences. Prenatal Diagnosis, 2021, 41, 554-563.	2.3	9
14	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. Genetics in Medicine, 2021, 23, 1137-1142.	2.4	58
15	A placental trisomy 2 detected by NIPT evolved in a fetal small Supernumerary Marker Chromosome (sSMC). Molecular Cytogenetics, 2021, 14, 18.	0.9	2
16	The landscape of copy number variations in classical Hodgkin lymphoma: a joint KU Leuven and LYSA study on cell-free DNA. Blood Advances, 2021, 5, 1991-2002.	5.2	15
17	Performance and Diagnostic Value of Genome-Wide Noninvasive Prenatal Testing in Multiple Gestations. Obstetrics and Gynecology, 2021, 137, 1102-1108.	2.4	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. EClinicalMedicine, 2021, 35, 100856.	7.1	42

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19	22q11.2 Low Copy Repeats Expanded in the Human Lineage. Frontiers in Genetics, 2021, 12, 706641.	2.3	11
20	Nonâ€invasive prenatal testing suggesting a maternal malignancy: What do we tell the prospective parents in Belgium?. Prenatal Diagnosis, 2021, 41, 1264-1272.	2.3	20
21	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87
22	Haplotyping-based preimplantation genetic testing reveals parent-of-origin specific mechanisms of aneuploidy formation. Npj Genomic Medicine, 2021, 6, 81.	3.8	21
23	A benchmark of structural variation detection by long reads through a realistic simulated model. Genome Biology, 2021, 22, 342.	8.8	21
24	Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. PLoS Computational Biology, 2021, 17, e1009684.	3.2	6
25	Detection of incipient tumours by screening of circulating plasma DNA: hype or hope?. Acta Clinica Belgica, 2020, 75, 9-18.	1.2	9
26	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
27	Pathogenic variants in CDC45 on the remaining allele in patients with a chromosome 22q11.2 deletion result in a novel autosomal recessive condition. Genetics in Medicine, 2020, 22, 326-335.	2.4	17
28	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	6.2	42
29	Extreme enrichment of VNTR-associated polymorphicity in human subtelomeres: genes with most VNTRs are predominantly expressed in the brain. Translational Psychiatry, 2020, 10, 369.	4.8	15
30	Genotype-phenotype correlations of UBA2 mutations in patients with ectrodactyly. European Journal of Medical Genetics, 2020, 63, 104009.	1.3	4
31	Breast Cancer Detection and Treatment Monitoring Using a Noninvasive Prenatal Testing Platform: Utility in Pregnant and Nonpregnant Populations. Clinical Chemistry, 2020, 66, 1414-1423.	3.2	9
32	De Novo Variants in LMNB1 Cause Pronounced Syndromic Microcephaly and Disruption of Nuclear Envelope Integrity. American Journal of Human Genetics, 2020, 107, 753-762.	6.2	30
33	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	30.7	90
34	Current use of noninvasive prenatal testing in Europe, Australia and the USA: A graphical presentation. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 722-730.	2.8	121
35	Identity-by-state-based haplotyping expands the application of comprehensive preimplantation genetic testing. Human Reproduction, 2020, 35, 718-726.	0.9	6
36	Noninvasive prenatal diagnosis by genome-wide haplotyping of cell-free plasma DNA. Genetics in Medicine, 2020, 22, 962-973.	2.4	29

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37	LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	2.0	11
38	Ultraâ€low depth sequencing of plasma cell <scp>DNA</scp> for the detection of copy number aberrations in multiple myeloma. Genes Chromosomes and Cancer, 2020, 59, 465-471.	2.8	3
39	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	3.2	11
40	Prenatally detected copy number variants in a national cohort: A postnatal followâ€up study. Prenatal Diagnosis, 2020, 40, 1272-1283.	2.3	5
41	PREIMPLANTATION GENETIC TESTING: Single-cell technologies at the forefront of PGT and embryo research. Reproduction, 2020, 160, A19-A31.	2.6	10
42	Opportunities of Genome Imaging for Genetic Diagnosis in Acute Lymphoblastic Leukemia. Blood, 2020, 136, 10-11.	1.4	0
43	Pregnant women with confirmed neoplasms should not have noninvasive prenatal testing. Prenatal Diagnosis, 2019, 39, 1162-1165.	2.3	17
44	Fetal sex determination in twin pregnancies using non-invasive prenatal testing. Npj Genomic Medicine, 2019, 4, 15.	3.8	10
45	Multi-centre evaluation of a comprehensive preimplantation genetic test through haplotyping-by-sequencing. Human Reproduction, 2019, 34, 1608-1619.	0.9	48
46	Noninvasive Prenatal Testing and Detection of Occult Maternal Malignancies. Clinical Chemistry, 2019, 65, 1484-1486.	3.2	19
47	In vitro fertilization does not increase the incidence of de novo copy number alterations in fetal and placental lineages. Nature Medicine, 2019, 25, 1699-1705.	30.7	43
48	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. Genome Research, 2019, 29, 1389-1401.	5.5	39
49	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. Genetics in Medicine, 2019, 21, 2774-2780.	2.4	16
50	The sudden death of the combined first trimester aneuploidy screening, a single centre experience in Belgium. Clinical Chemistry and Laboratory Medicine, 2019, 57, e294-e297.	2.3	4
51	Preeclampsia is Associated with Sex-Specific Transcriptional and Proteomic Changes in Fetal Erythroid Cells. International Journal of Molecular Sciences, 2019, 20, 2038.	4.1	16
52	Current Controversies in Prenatal Diagnosis 3: Gene editing should replace embryo selection following PGD. Prenatal Diagnosis, 2019, 39, 344-350.	2.3	8
53	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
54	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	3.1	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 Clefting Susceptibility Gene. Genes, 2019, 10, 1023.	2.4	26
56	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	2.9	7
57	A speculative outlook on embryonic aneuploidy: Can molecular pathways be involved?. Developmental Biology, 2019, 447, 3-13.	2.0	29
58	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26
59	NIPTmer: rapid k-mer-based software package for detection of fetal aneuploidies. Scientific Reports, 2018, 8, 5616.	3.3	12
60	Single molecule real-time (SMRT) sequencing comes of age: applications and utilities for medical diagnostics. Nucleic Acids Research, 2018, 46, 2159-2168.	14.5	518
61	Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	2.3	58
62	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	2.9	22
63	Congenital diaphragmatic hernia as a part of Nanceâ€“Horan syndrome?. European Journal of Human Genetics, 2018, 26, 359-366.	2.8	7
64	Conventional and Single-Molecule Targeted Sequencing Method for Specific Variant Detection in IKBKG while Bypassing the IKBKGP1 Pseudogene. Journal of Molecular Diagnostics, 2018, 20, 195-202.	2.8	26
65	Noise-robust assessment of SNP array based CNV calls through local noise estimation of log R ratios. Statistical Applications in Genetics and Molecular Biology, 2018, 17, .	0.6	0
66	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	2.5	16
67	Neurodevelopmental risk copy number variants in adults with intellectual disabilities and comorbid psychiatric disorders. British Journal of Psychiatry, 2018, 212, 287-294.	2.8	30
68	Clinical characteristics of patients with low functional IL-6 production upon TLR/IL-1R stimulation. Journal of Allergy and Clinical Immunology, 2018, 141, 768-770.	2.9	0
69	Improved reference genome for the domestic horse increases assembly contiguity and composition. Communications Biology, 2018, 1, 197.	4.4	148
70	Mapping the landscape of tandem repeat variability by targeted long read single molecule sequencing in familial X-linked intellectual disability. BMC Medical Genomics, 2018, 11, 123.	1.5	5
71	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. American Journal of Medical Genetics, Part A, 2018, 176, 2172-2181.	1.2	33
72	Genome-wide haplotyping embryos developing from 0PN and 1PN zygotes increases transferrable embryos in PGT-M. Human Reproduction, 2018, 33, 2302-2311.	0.9	33

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73	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128.	2.3	24
74	Molecular genetics of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2070-2081.	1.2	96
75	Efficient CRISPR/Cas9-mediated editing of trinucleotide repeat expansion in myotonic dystrophy patient-derived iPS and myogenic cells. Nucleic Acids Research, 2018, 46, 8275-8298.	14.5	78
76	Genetic profile of isolated congenital diaphragmatic hernia revealed by targeted nextâ€generation sequencing. Prenatal Diagnosis, 2018, 38, 654-663.	2.3	31
77	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. Frontiers in Genetics, 2018, 9, 150.	2.3	26
78	Response to a comment on â€œPredicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testingâ€. Prenatal Diagnosis, 2018, 38, 722-723.	2.3	1
79	The genetic structure of the Belgian population. Human Genomics, 2018, 12, 6.	2.9	7
80	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909.	1.2	7
81	Unraveling the Landscape of Copy Number Aberrations in Hodgkin Lymphoma: A Joint KU Leuven and Lysa Study on Circulating Cell Free DNA. Blood, 2018, 132, 2836-2836.	1.4	0
82	Mutational Processes Shaping the Genome in Early Human Embryos. Cell, 2017, 168, 751-753.	28.9	9
83	How can zygotes segregate entire parental genomes into distinct blastomeres? The zygote metaphase revisited. BioEssays, 2017, 39, 1600226.	2.5	11
84	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	2.8	33
85	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. Human Reproduction, 2017, 32, 687-697.	0.9	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. Clinical Lymphoma, Myeloma and Leukemia, 2017, 17, e95.	0.4	0
87	An incidental finding of maternal multiple myeloma by non invasive prenatal testing. Prenatal Diagnosis, 2017, 37, 1257-1260.	2.3	13
88	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the GPR98 Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
89	Genome stability of bovine in vivo-conceived cleavage-stage embryos is higher compared to in vitro-produced embryos. Human Reproduction, 2017, 32, 2348-2357.	0.9	69
90	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	7.2	77

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91	Accuracy and Clinical Value of Maternal Incidental Findings During Noninvasive Prenatal Testing for Fetal Aneuploidies. Obstetrical and Gynecological Survey, 2017, 72, 469-470.	0.4	1
92	Array CGH. Springer Protocols, 2017, , 567-586.	0.3	1
93	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. Genetics in Medicine, 2017, 19, 306-313.	2.4	47
94	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331.	2.5	37
95	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. Genome Medicine, 2017, 9, 67.	8.2	29
96	Maternal vitamin B12 deficiency and abnormal cell-free DNA results in pregnancy. Prenatal Diagnosis, 2016, 36, 790-793.	2.3	17
97	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	2.5	30
98	Zygotes segregate entire parental genomes in distinct blastomere lineages causing cleavage-stage chimerism and mixoploidy. Genome Research, 2016, 26, 567-578.	5.5	73
99	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. European Journal of Medical Genetics, 2016, 59, 436-443.	1.3	20
100	Current controversies in prenatal diagnosis 3: industry drives innovation in research and clinical application of genetic prenatal diagnosis and screening. Prenatal Diagnosis, 2016, 36, 1172-1177.	2.3	9
101	Prenatal and pre-implantation genetic diagnosis. Nature Reviews Genetics, 2016, 17, 643-656.	16.3	155
102	Variant discovery and breakpoint region prediction for studying the human 22q11.2 deletion using BAC clone and whole genome sequencing analysis. Human Molecular Genetics, 2016, 25, 3754-3767.	2.9	20
103	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.7	8
104	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	2.5	92
105	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. European Journal of Human Genetics, 2016, 24, 51-58.	2.8	10
106	Polymerase specific error rates and profiles identified by single molecule sequencing. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 784-785, 39-45.	1.0	44
107	Pulmonary transcriptome analysis in the rabbit model of surgically-induced diaphragmatic hernia treated with fetal tracheal occlusion. DMM Disease Models and Mechanisms, 2016, 9, 221-8.	2.4	18
108	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954

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109	Genotypic and phenotypic variation in six patients with solitary median maxillary central incisor syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2451-2458.	1.2	21
110	Concurrent Whole-Genome Haplotyping and Copy-Number Profiling of Single Cells. American Journal of Human Genetics, 2015, 96, 894-912.	6.2	110
111	Presymptomatic Identification of Cancers in Pregnant Women During Noninvasive Prenatal Testing. JAMA Oncology, 2015, 1, 814.	7.1	180
112	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
113	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63
114	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 2015, 37, 527-536.	1.1	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. Lancet Haematology,the, 2015, 2, e55-e65.	4.6	115
116	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. European Journal of Human Genetics, 2015, 23, 1286-1293.	2.8	108
117	GBSX: a toolkit for experimental design and demultiplexing genotyping by sequencing experiments. BMC Bioinformatics, 2015, 16, 73.	2.6	102
118	Expanding the phenotypic spectrum of PORCN variants in two males with syndromic microphthalmia. European Journal of Human Genetics, 2015, 23, 551-554.	2.8	24
119	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. American Journal of Medical Genetics, Part A, 2015, 167, 1822-1829.	1.2	48
120	Copy Number Variation Analysis by Array Analysis of Single Cells Following Whole Genome Amplification. Methods in Molecular Biology, 2015, 1347, 197-219.	0.9	6
121	Lack of Evidence That Male Fetal Microchimerism is Present in Endometriosis. Reproductive Sciences, 2015, 22, 1115-1121.	2.5	3
122	NGS-Logistics: federated analysis of NGS sequence variants across multiple locations. Genome Medicine, 2014, 6, 71.	8.2	16
123	Pseudoautosomal Region 1 Length Polymorphism in the Human Population. PLoS Genetics, 2014, 10, e1004578.	3.5	24
124	Prenatal management of the fetus with isolated congenital diaphragmatic hernia in the era of the TOTAL trial. Seminars in Fetal and Neonatal Medicine, 2014, 19, 338-348.	2.3	149
125	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. Human Reproduction, 2014, 29, 1603-1609.	0.9	57
126	DNA from Nails for Genetic Analyses in Large-Scale Epidemiologic Studies. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 2703-2712.	2.5	27

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127	Single cell segmental aneuploidy detection is compromised by S phase. Molecular Cytogenetics, 2014, 7, 46.	0.9	23
128	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	1.3	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. Genetics in Medicine, 2014, 16, 469-476.	2.4	66
130	Exome sequencing identifies ZFPM2 as a cause of familial isolated congenital diaphragmatic hernia and possibly cardiovascular malformations. European Journal of Medical Genetics, 2014, 57, 247-252.	1.3	17
131	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
132	Large-scale analysis of tandem repeat variability in the human genome. Nucleic Acids Research, 2014, 42, 5728-5741.	14.5	66
133	Single-cell paired-end genome sequencing reveals structural variation per cell cycle. Nucleic Acids Research, 2013, 41, 6119-6138.	14.5	142
134	Preimplantation genetic diagnosis guided by single-cell genomics. Genome Medicine, 2013, 5, 71.	8.2	45
135	Nonallelic homologous recombination between retrotransposable elements is a driver of de novo unbalanced translocations. Genome Research, 2013, 23, 411-418.	5.5	90
136	The Future of Prenatal Cytogenetics: From Copy Number Variations to Non-invasive Prenatal Testing. Current Genetic Medicine Reports, 2013, 1, 91-98.	1.9	1
137	Aneuploidy and Copy Number Variation in Early Human Development. Seminars in Reproductive Medicine, 2012, 30, 302-308.	1.1	10
138	Chromosome Instability Is Common in Human Cleavage-Stage Embryos. Obstetrical and Gynecological Survey, 2012, 67, 787-788.	0.4	1
139	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. Genome Medicine, 2012, 4, 73.	8.2	28
140	New array approaches to explore single cells genomes. Frontiers in Genetics, 2012, 3, 44.	2.3	20
141	Cytogenetic and morphological analysis of early products of conception following hysteroâ€embryoscopy from couples with recurrent pregnancy loss. Prenatal Diagnosis, 2012, 32, 933-942.	2.3	45
142	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. Human Mutation, 2012, 33, 906-915.	2.5	69
143	Array-Based Approaches in Prenatal Diagnosis. Methods in Molecular Biology, 2012, 838, 151-171.	0.9	13
144	Breakage-fusion-bridge cycles leading to inv dup del occur in human cleavage stage embryos. Human Mutation, 2011, 32, 783-793.	2.5	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. American Journal of Human Genetics, 2010, 86, 749-764.	6.2	2,325
146	Diagnosis of miscarriages by molecular karyotyping: Benefits and pitfalls. Genetics in Medicine, 2009, 11, 646-654.	2.4	90
147	What next for preimplantation genetic screening? High mitotic chromosome instability rate provides the biological basis for the low success rate. Human Reproduction, 2009, 24, 2679-2682.	0.9	87
148	Chromosome instability is common in human cleavage-stage embryos. Nature Medicine, 2009, 15, 577-583.	30.7	710
149	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
150	Autosomal-Dominant Microtia Linked to Five Tandem Copies of a Copy-Number-Variable Region at Chromosome 4p16. American Journal of Human Genetics, 2008, 82, 181-187.	6.2	42
151	Submicroscopic chromosomal imbalances detected by array-CGH are a frequent cause of congenital heart defects in selected patients. European Heart Journal, 2007, 28, 2778-2784.	2.2	175
152	Subtelomeric imbalances in phenotypically normal individuals. Human Mutation, 2007, 28, 958-967.	2.5	72
153	Detection of genomic copy number changes in patients with idiopathic mental retardation by high-resolution X-array-CGH: important role for increased gene dosage ofXLMRgenes. Human Mutation, 2007, 28, 1034-1042.	2.5	166
154	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	2.8	144
155	Reply to Hochstenbach et al. European Journal of Human Genetics, 2006, 14, 1063-1064.	2.8	14
156	Single-cell chromosomal imbalances detection by array CGH. Nucleic Acids Research, 2006, 34, e68-e68.	14.5	188
157	Molecular Karyotyping: Array CGH Quality Criteria for Constitutional Genetic Diagnosis. Journal of Histochemistry and Cytochemistry, 2005, 53, 413-422.	2.5	141
158	Exon Array CGH: Detection of Copy-Number Changes at the Resolution of Individual Exons in the Human Genome. American Journal of Human Genetics, 2005, 76, 750-762.	6.2	132
159	Interstitial 6q deletion: clinical and array CGH characterisation of a new patient. European Journal of Medical Genetics, 2005, 48, 339-345.	1.3	24
160	Tetrasomy 12pter-12p13.31 in a girl with partial Pallisterâ€“Killian syndrome phenotype. European Journal of Medical Genetics, 2005, 48, 319-327.	1.3	21
161	From chromosomes to molecular karyotyping. European Journal of Medical Genetics, 2005, 48, 211-213.	1.3	2
162	25ÂMb deletion of 13q13.3â†'q21.31 in a patient without retinoblastoma. European Journal of Medical Genetics, 2005, 48, 363-366.	1.3	5

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163	Trisomy of chromosome 16p13.3 due to an unbalanced insertional translocation into chromosome 22p13. European Journal of Medical Genetics, 2005, 48, 355-359.	1.3	21
164	Fusion of NUP214 to ABL1 on Amplified Extrachromosomal Elements in T-ALL.. Blood, 2004, 104, 141-141.	1.4	3
165	No evidence for a parental inversion polymorphism predisposing to rearrangements at 22q11.2 in the DiGeorge/Velocardiofacial syndrome. European Journal of Human Genetics, 2003, 11, 109-111.	2.8	23
166	Characterization of centromere alterations in liposarcomas. Genes Chromosomes and Cancer, 2000, 29, 117-129.	2.8	73
167	Noonan-like phenotype in monozygotic twins with a duplication-deficiency of the long arm of chromosome 18 resulting from a maternal paracentric inversion. Human Genetics, 1998, 103, 497-505.	3.8	23
168	The IL-9 receptor gene, located in the Xq/Yq pseudoautosomal region, has an autosomal origin, escapes X inactivation and is expressed from the Y. Human Molecular Genetics, 1997, 6, 1-8.	2.9	54
169	Interstitial telomeric sequences at the junction site of a jumping translocation. Human Genetics, 1997, 99, 735-737.	3.8	57
170	FISH identifies different types of duplications with 12q13-15 as the commonly involved segment in B-cell lymphoproliferative malignancies characterized by partial trisomy 12. Genes Chromosomes and Cancer, 1997, 20, 155-166.	2.8	42