Joris Robert Vermeesch

List of Publications by Year
in descending order

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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	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.	2.6	2,325
2	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	18.1	954
3	Chromosome instability is common in human cleavage-stage embryos. Nature Medicine, 2009, 15, 577-583.	15.2	710
4	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
5	Single molecule real-time (SMRT) sequencing comes of age: applications and utilities for medical diagnostics. Nucleic Acids Research, 2018, 46, 2159-2168.	6.5	518
6	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
7	Single-cell chromosomal imbalances detection by array CGH. Nucleic Acids Research, 2006, 34, e68-e68.	6.5	188
8	Presymptomatic Identification of Cancers in Pregnant Women During Noninvasive Prenatal Testing. JAMA Oncology, 2015, 1, 814.	3.4	180
9	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.	1.0	175
10	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.	1.1	166
11	Prenatal and pre-implantation genetic diagnosis. Nature Reviews Genetics, 2016, 17, 643-656.	7.7	155
12	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.	1.1	149
13	Improved reference genome for the domestic horse increases assembly contiguity and composition. Communications Biology, 2018, 1, 197.	2.0	148
14	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	1.4	144
15	Single-cell paired-end genome sequencing reveals structural variation per cell cycle. Nucleic Acids Research, 2013, 41, 6119-6138.	6.5	142
16	Molecular Karyotyping: Array CGH Quality Criteria for Constitutional Genetic Diagnosis. Journal of Histochemistry and Cytochemistry, 2005, 53, 413-422.	1.3	141
17	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.	2.6	132
18	Current use of noninvasive prenatal testing in Europe, Australia and the USA: A graphical presentation. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 722-730.	1.3	121

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19	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.	2.2	115
20	Concurrent Whole-Genome Haplotyping and Copy-Number Profiling of Single Cells. American Journal of Human Genetics, 2015, 96, 894-912.	2.6	110
21	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.	1.4	108
22	GBSX: a toolkit for experimental design and demultiplexing genotyping by sequencing experiments. BMC Bioinformatics, 2015, 16, 73.	1.2	102
23	Molecular genetics of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2070-2081.	0.7	96
24	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	1.1	92
25	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	0.7	91
26	Diagnosis of miscarriages by molecular karyotyping: Benefits and pitfalls. Genetics in Medicine, 2009, 11, 646-654.	1.1	90
27	Nonallelic homologous recombination between retrotransposable elements is a driver of de novo unbalanced translocations. Genome Research, 2013, 23, 411-418.	2.4	90
28	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	15.2	90
29	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.	0.4	87
30	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
31	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	1.4	85
32	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.	6.5	78
33	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	4.0	77
34	Characterization of centromere alterations in liposarcomas. Genes Chromosomes and Cancer, 2000, 29, 117-129.	1.5	73
35	Zygotes segregate entire parental genomes in distinct blastomere lineages causing cleavage-stage chimerism and mixoploidy. Genome Research, 2016, 26, 567-578.	2.4	73
36	Subtelomeric imbalances in phenotypically normal individuals. Human Mutation, 2007, 28, 958-967.	1.1	72

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37	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. Human Mutation, 2012, 33, 906-915.	1.1	69
38	Genome stability of bovine in vivo-conceived cleavage-stage embryos is higher compared to in vitro-produced embryos. Human Reproduction, 2017, 32, 2348-2357.	0.4	69
39	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.	1.1	66
40	Large-scale analysis of tandem repeat variability in the human genome. Nucleic Acids Research, 2014, 42, 5728-5741.	6.5	66
41	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	2.6	63
42	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
43	Breakage-fusion-bridge cycles leading to inv dup del occur in human cleavage stage embryos. Human Mutation, 2011, 32, 783-793.	1.1	60
44	Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	1.1	58
45	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. Genetics in Medicine, 2021, 23, 1137-1142.	1.1	58
46	Interstitial telomeric sequences at the junction site of a jumping translocation. Human Genetics, 1997, 99, 735-737.	1.8	57
47	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. Human Reproduction, 2014, 29, 1603-1609.	0.4	57
48	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.	1.4	54
49	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. American Journal of Medical Genetics, Part A, 2015, 167, 1822-1829.	0.7	48
50	Multi-centre evaluation of a comprehensive preimplantation genetic test through haplotyping-by-sequencing. Human Reproduction, 2019, 34, 1608-1619.	0.4	48
51	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. Genetics in Medicine, 2017, 19, 306-313.	1.1	47
52	Cytogenetic and morphological analysis of early products of conception following hysteroâ€embryoscopy from couples with recurrent pregnancy loss. Prenatal Diagnosis, 2012, 32, 933-942.	1.1	45
53	Preimplantation genetic diagnosis guided by single-cell genomics. Genome Medicine, 2013, 5, 71.	3.6	45
54	Polymerase specific error rates and profiles identified by single molecule sequencing. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 784-785, 39-45.	0.4	44

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55	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.	15.2	43
56	FISH identifies different types of duplications with 12q13-15 as the commonly involved segment in B-cell lymphoproliferative malignancies characterized by partial trisomy 12. , 1997, 20, 155-166.		42
57	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.	2.6	42
58	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.	2.6	42
59	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.	3.2	42
60	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
61	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. Human Reproduction, 2017, 32, 687-697.	0.4	40
62	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. Genome Research, 2019, 29, 1389-1401.	2.4	39
63	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331.	1.1	37
64	Optimizing the diagnostic workflow for acute lymphoblastic leukemia by optical genome mapping. American Journal of Hematology, 2022, 97, 548-561.	2.0	36
65	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.	1.4	33
66	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.	0.7	33
67	Genome-wide haplotyping embryos developing from 0PN and 1PN zygotes increases transferrable embryos in PGT-M. Human Reproduction, 2018, 33, 2302-2311.	0.4	33
68	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 2015, 37, 527-536.	0.6	32
69	Genetic profile of isolated congenital diaphragmatic hernia revealed by targeted nextâ€generation sequencing. Prenatal Diagnosis, 2018, 38, 654-663.	1.1	31
70	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	1.1	30
71	Neurodevelopmental risk copy number variants in adults with intellectual disabilities and comorbid psychiatric disorders. British Journal of Psychiatry, 2018, 212, 287-294.	1.7	30
72	De Novo Variants in LMNB1 Cause Pronounced Syndromic Microcephaly and Disruption of Nuclear Envelope Integrity. American Journal of Human Genetics, 2020, 107, 753-762.	2.6	30

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73	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. Genome Medicine, 2017, 9, 67.	3.6	29
74	A speculative outlook on embryonic aneuploidy: Can molecular pathways be involved?. Developmental Biology, 2019, 447, 3-13.	0.9	29
75	Noninvasive prenatal diagnosis by genome-wide haplotyping of cell-free plasma DNA. Genetics in Medicine, 2020, 22, 962-973.	1.1	29
76	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. Genome Medicine, 2012, 4, 73.	3.6	28
77	DNA from Nails for Genetic Analyses in Large-Scale Epidemiologic Studies. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 2703-2712.	1.1	27
78	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.	1.2	26
79	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.	1.1	26
80	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.	1.0	26
81	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.	4.1	26
82	Interstitial 6q deletion: clinical and array CGH characterisation of a new patient. European Journal of Medical Genetics, 2005, 48, 339-345.	0.7	24
83	Pseudoautosomal Region 1 Length Polymorphism in the Human Population. PLoS Genetics, 2014, 10, e1004578.	1.5	24
84	Expanding the phenotypic spectrum of PORCN variants in two males with syndromic microphthalmia. European Journal of Human Genetics, 2015, 23, 551-554.	1.4	24
85	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128.	1.1	24
86	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.	1.8	23
87	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.	1.4	23
88	Single cell segmental aneuploidy detection is compromised by S phase. Molecular Cytogenetics, 2014, 7, 46.	0.4	23
89	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
90	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	1.4	22

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91	Tetrasomy 12pter-12p13.31 in a girl with partial Pallisterâ€“Killian syndrome phenotype. European Journal of Medical Genetics, 2005, 48, 319-327.	0.7	21
92	Trisomy of chromosome 16p13.3 due to an unbalanced insertional translocation into chromosome 22p13. European Journal of Medical Genetics, 2005, 48, 355-359.	0.7	21
93	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.	0.7	21
94	Haplotyping-based preimplantation genetic testing reveals parent-of-origin specific mechanisms of aneuploidy formation. Npj Genomic Medicine, 2021, 6, 81.	1.7	21
95	A benchmark of structural variation detection by long reads through a realistic simulated model. Genome Biology, 2021, 22, 342.	3.8	21
96	New array approaches to explore single cells genomes. Frontiers in Genetics, 2012, 3, 44.	1.1	20
97	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.	0.7	20
98	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.	1.4	20
99	Nonâ€invasive prenatal testing suggesting a maternal malignancy: What do we tell the prospective parents in Belgium?. Prenatal Diagnosis, 2021, 41, 1264-1272.	1.1	20
100	Noninvasive Prenatal Testing and Detection of Occult Maternal Malignancies. Clinical Chemistry, 2019, 65, 1484-1486.	1.5	19
101	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.	1.2	18
102	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.	0.7	17
103	Maternal vitamin B12 deficiency and abnormal cell-free DNA results in pregnancy. Prenatal Diagnosis, 2016, 36, 790-793.	1.1	17
104	Pregnant women with confirmed neoplasms should not have noninvasive prenatal testing. Prenatal Diagnosis, 2019, 39, 1162-1165.	1.1	17
105	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
106	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.	1.1	17
107	Single-cell genome-wide concurrent haplotyping and copy-number profiling through genotyping-by-sequencing. Nucleic Acids Research, 2022, 50, e63-e63.	6.5	17
108	NGS-Logistics: federated analysis of NGS sequence variants across multiple locations. Genome Medicine, 2014, 6, 71.	3.6	16

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109	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	1.1	16
110	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. Genetics in Medicine, 2019, 21, 2774-2780.	1.1	16
111	Preeclampsia is Associated with Sex-Specific Transcriptional and Proteomic Changes in Fetal Erythroid Cells. International Journal of Molecular Sciences, 2019, 20, 2038.	1.8	16
112	Extreme enrichment of VNTR-associated polymorphicity in human subtelomeres: genes with most VNTRs are predominantly expressed in the brain. Translational Psychiatry, 2020, 10, 369.	2.4	15
113	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.	2.5	15
114	Reply to Hochstenbach et al. European Journal of Human Genetics, 2006, 14, 1063-1064.	1.4	14
115	Array-Based Approaches in Prenatal Diagnosis. Methods in Molecular Biology, 2012, 838, 151-171.	0.4	13
116	An incidental finding of maternal multiple myeloma by non invasive prenatal testing. Prenatal Diagnosis, 2017, 37, 1257-1260.	1.1	13
117	NIPTmer: rapid k-mer-based software package for detection of fetal aneuploidies. Scientific Reports, 2018, 8, 5616.	1.6	12
118	A normative chart for cognitive development in a genetically selected population. Neuropsychopharmacology, 2022, 47, 1379-1386.	2.8	12
119	How can zygotes segregate entire parental genomes into distinct blastomeres? The zygote metaphase revisited. BioEssays, 2017, 39, 1600226.	1.2	11
120	LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	1.0	11
121	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	1.5	11
122	22q11.2 Low Copy Repeats Expanded in the Human Lineage. Frontiers in Genetics, 2021, 12, 706641.	1.1	11
123	Aneuploidy and Copy Number Variation in Early Human Development. Seminars in Reproductive Medicine, 2012, 30, 302-308.	0.5	10
124	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.	1.4	10
125	Fetal sex determination in twin pregnancies using non-invasive prenatal testing. Npj Genomic Medicine, 2019, 4, 15.	1.7	10
126	Performance and Diagnostic Value of Genome-Wide Noninvasive Prenatal Testing in Multiple Gestations. Obstetrics and Gynecology, 2021, 137, 1102-1108.	1.2	10

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127	PREIMPLANTATION GENETIC TESTING: Single-cell technologies at the forefront of PGT and embryo research. Reproduction, 2020, 160, A19-A31.	1.1	10
128	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.	1.1	9
129	Mutational Processes Shaping the Genome in Early Human Embryos. Cell, 2017, 168, 751-753.	13.5	9
130	Detection of incipient tumours by screening of circulating plasma DNA: hype or hope?. Acta Clinica Belgica, 2020, 75, 9-18.	0.5	9
131	Breast Cancer Detection and Treatment Monitoring Using a Noninvasive Prenatal Testing Platform: Utility in Pregnant and Nonpregnant Populations. Clinical Chemistry, 2020, 66, 1414-1423.	1.5	9
132	Genomeâ€wide abnormalities in embryos: Origins and clinical consequences. Prenatal Diagnosis, 2021, 41, 554-563.	1.1	9
133	A review of normative documents on preimplantation genetic testing: Recommendations for PGT-P. Genetics in Medicine, 2022, 24, 1165-1175.	1.1	9
134	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.4	8
135	Current Controversies in Prenatal Diagnosis 3: Gene editing should replace embryo selection following PGD. Prenatal Diagnosis, 2019, 39, 344-350.	1.1	8
136	Congenital diaphragmatic hernia as a part of Nanceâ€“Horan syndrome?. European Journal of Human Genetics, 2018, 26, 359-366.	1.4	7
137	The genetic structure of the Belgian population. Human Genomics, 2018, 12, 6.	1.4	7
138	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.	0.7	7
139	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	1.4	7
140	Copy Number Variation Analysis by Array Analysis of Single Cells Following Whole Genome Amplification. Methods in Molecular Biology, 2015, 1347, 197-219.	0.4	6
141	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	1.5	6
142	Identity-by-state-based haplotyping expands the application of comprehensive preimplantation genetic testing. Human Reproduction, 2020, 35, 718-726.	0.4	6
143	Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. PLoS Computational Biology, 2021, 17, e1009684.	1.5	6
144	Pan-Cancer Detection and Typing by Mining Patterns in Large Genome-Wide Cell-Free DNA Sequencing Datasets. Clinical Chemistry, 2022, 68, 1164-1176.	1.5	6

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145	25ÂMb deletion of 13q13.3â†'q21.31 in a patient without retinoblastoma. European Journal of Medical Genetics, 2005, 48, 363-366.	0.7	5
146	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.	0.7	5
147	Prenatally detected copy number variants in a national cohort: A postnatal followâ€up study. Prenatal Diagnosis, 2020, 40, 1272-1283.	1.1	5
148	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.	1.4	4
149	Genotype-phenotype correlations of UBA2 mutations in patients with ectrodactyly. European Journal of Medical Genetics, 2020, 63, 104009.	0.7	4
150	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.	1.4	4
151	Lack of Evidence That Male Fetal Microchimerism is Present in Endometriosis. Reproductive Sciences, 2015, 22, 1115-1121.	1.1	3
152	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.	1.5	3
153	Ultra-low coverage whole genome sequencing of ccfDNA in multiple myeloma: A tool for laboratory routine?. Cancer Treatment and Research Communications, 2021, 28, 100380.	0.7	3
154	Fusion of NUP214 to ABL1 on Amplified Extrachromosomal Elements in T-ALL.. Blood, 2004, 104, 141-141.	0.6	3
155	From chromosomes to molecular karyotyping. European Journal of Medical Genetics, 2005, 48, 211-213.	0.7	2
156	A placental trisomy 2 detected by NIPT evolved in a fetal small Supernumerary Marker Chromosome (sSMC). Molecular Cytogenetics, 2021, 14, 18.	0.4	2
157	Primary mediastinal large Bâ€cell lymphoma is characterized by largeâ€scale copyâ€neutral loss of heterozygosity. Genes Chromosomes and Cancer, 2022, 61, 603-615.	1.5	2
158	Chromosome Instability Is Common in Human Cleavage-Stage Embryos. Obstetrical and Gynecological Survey, 2012, 67, 787-788.	0.2	1
159	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
160	Accuracy and Clinical Value of Maternal Incidental Findings During Noninvasive Prenatal Testing for Fetal Aneuploidies. Obstetrical and Gynecological Survey, 2017, 72, 469-470.	0.2	1
161	Array CGH. Springer Protocols, 2017, , 567-586.	0.1	1
162	Response to a comment on â€œPredicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testingâ€• Prenatal Diagnosis, 2018, 38, 722-723.	1.1	1

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163	The Hunt for the Chromosome 22q11.2 Deletion Syndrome Schizophrenia Genes. Biological Psychiatry, 2022, 91, 692-693.	0.7	1
164	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.	0.2	0
165	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, .	0.2	0
166	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.	1.5	0
167	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.	0.6	0
168	Opportunities of Genome Imaging for Genetic Diagnosis in Acute Lymphoblastic Leukemia. Blood, 2020, 136, 10-11.	0.6	0
169	OUP accepted manuscript. Clinical Chemistry, 2022, 68, 634.	1.5	0
170	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, , .	0.2	0