

Erik A Sistermans

List of Publications by Year in descending order

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130
papers

8,689
citations

50170

46
h-index

46693

89
g-index

133
all docs

133
docs citations

133
times ranked

12229
citing authors

#	ARTICLE	IF	CITATIONS
1	Germline KRAS mutations cause Noonan syndrome. <i>Nature Genetics</i> , 2006, 38, 331-336.	9.4	670
2	Diagnostic Genome Profiling in Mental Retardation. <i>American Journal of Human Genetics</i> , 2005, 77, 606-616.	2.6	514
3	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. <i>Nature Genetics</i> , 2006, 38, 999-1001.	9.4	418
4	Guidelines for diagnostic next-generation sequencing. <i>European Journal of Human Genetics</i> , 2016, 24, 2-5.	1.4	389
5	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2006, 79, 370-377.	2.6	343
6	Patients lacking the major CNS myelin protein, proteolipid protein 1, develop length-dependent axonal degeneration in the absence of demyelination and inflammation. <i>Brain</i> , 2002, 125, 551-561.	3.7	272
7	PPIB Mutations Cause Severe Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2009, 85, 521-527.	2.6	257
8	3q29 Microdeletion Syndrome: Clinical and Molecular Characterization of a New Syndrome. <i>American Journal of Human Genetics</i> , 2005, 77, 154-160.	2.6	228
9	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. <i>American Journal of Human Genetics</i> , 2019, 105, 1091-1101.	2.6	222
10	L1 retrotransposition can occur early in human embryonic development. <i>Human Molecular Genetics</i> , 2007, 16, 1587-1592.	1.4	174
11	<i>PLS3</i> Mutations in X-Linked Osteoporosis with Fractures. <i>New England Journal of Medicine</i> , 2013, 369, 1529-1536.	13.9	171
12	Clinical spectrum of 4H leukodystrophy caused by <i>POLR3A</i> and <i>POLR3B</i> mutations. <i>Neurology</i> , 2014, 83, 1898-1905.	1.5	170
13	Disruption of the gene Euchromatin Histone Methyl Transferase1 (Eu-HMTase1) is associated with the 9q34 subtelomeric deletion syndrome. <i>Journal of Medical Genetics</i> , 2005, 42, 299-306.	1.5	162
14	CYP21 Gene Mutation Analysis in 198 Patients with 21-Hydroxylase Deficiency in The Netherlands: Six Novel Mutations and a Specific Cluster of Four Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 3852-3859.	1.8	154
15	Duplication of the proteolipid protein gene is the major cause of Pelizaeus-Merzbacher disease. <i>Neurology</i> , 1998, 50, 1749-1754.	1.5	141
16	Screening for subtelomeric rearrangements in 210 patients with unexplained mental retardation using multiplex ligation dependent probe amplification (MLPA). <i>Journal of Medical Genetics</i> , 2004, 41, 892-899.	1.5	136
17	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. <i>Human Mutation</i> , 2009, 30, 283-292.	1.1	136
18	Somatic mutations found in the healthy blood compartment of a 115-yr-old woman demonstrate oligoclonal hematopoiesis. <i>Genome Research</i> , 2014, 24, 733-742.	2.4	136

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19	Mutations in PIH1D3 Cause X-Linked Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. American Journal of Human Genetics, 2017, 100, 160-168.	2.6	136
20	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	2.6	135
21	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
22	WISECONDOR: detection of fetal aberrations from shallow sequencing maternal plasma based on a within-sample comparison scheme. Nucleic Acids Research, 2014, 42, e31-e31.	6.5	124
23	Deficiency in SLC25A1, Encoding the Mitochondrial Citrate Carrier, Causes Combined D-2- and L-2-Hydroxyglutaric Aciduria. American Journal of Human Genetics, 2013, 92, 627-631.	2.6	122
24	Trial by Dutch laboratories for evaluation of noninvasive prenatal testing. Part 2 clinical impact. Prenatal Diagnosis, 2016, 36, 1083-1090.	1.1	122
25	Rapid antibody test for diagnosing fragile X syndrome: a validation of the technique. Human Genetics, 1997, 99, 308-311.	1.8	115
26	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	1.4	107
27	Genotypic and phenotypic characterization of Noonan syndrome: New data and review of the literature. American Journal of Medical Genetics, Part A, 2005, 134A, 165-170.	0.7	101
28	Mutations in the sterol 27-hydroxylase gene (CYP27A) cause hepatitis of infancy as well as cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2002, 25, 501-513.	1.7	99
29	Imprinting Effect in Premature Ovarian Failure Confined to Paternally Inherited Fragile X Premutations. American Journal of Human Genetics, 2000, 66, 413-418.	2.6	93
30	Heterogeneous Duplications in Patients with Pelizaeus-Merzbacher Disease Suggest a Mechanism of Coupled Homologous and Nonhomologous Recombination. American Journal of Human Genetics, 2005, 77, 966-987.	2.6	93
31	Three or more copies of the proteolipid protein gene PLP1 cause severe Pelizaeus-Merzbacher disease. Brain, 2005, 128, 743-751.	3.7	91
32	Clinical and molecular characteristics of 1qter microdeletion syndrome: delineating a critical region for corpus callosum agenesis/hypogenesis. Journal of Medical Genetics, 2008, 45, 346-354.	1.5	87
33	Origin and clinical relevance of chromosomal aberrations other than the common trisomies detected by genome-wide NIPS: results of the TRIDENT study. Genetics in Medicine, 2018, 20, 480-485.	1.1	85
34	Smoothing waves in array CGH tumor profiles. Bioinformatics, 2009, 25, 1099-1104.	1.8	76
35	Calculating the fetal fraction for noninvasive prenatal testing based on genome-wide nucleosome profiles. Prenatal Diagnosis, 2016, 36, 614-621.	1.1	76
36	SMAD2 Mutations Are Associated with Arterial Aneurysms and Dissections. Human Mutation, 2015, 36, 1145-1149.	1.1	74

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37	Characteristic brain magnetic resonance imaging pattern in patients with macrocephaly and <i>PTEN</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 627-633.	0.7	70
38	MECP2 Mutation in a Boy with Severe Neonatal Encephalopathy: Clinical, Neuropathological and Molecular Findings. <i>Neuropediatrics</i> , 2002, 33, 33-36.	0.3	63
39	Tissue- and cell-specific distribution of creatine kinase B: A new and highly specific monoclonal antibody for use in immunohistochemistry. <i>Cell and Tissue Research</i> , 1995, 280, 435-446.	1.5	62
40	De novo MECP2 frameshift mutation in a boy with moderate mental retardation, obesity and gynaecomastia. <i>Clinical Genetics</i> , 2002, 61, 359-362.	1.0	59
41	Mutations in noncoding regions of the proteolipid protein gene in Pelizaeus-Merzbacher disease. <i>Neurology</i> , 2000, 55, 1089-1096.	1.5	57
42	A (G-to-A) mutation in the initiation codon of the proteolipid protein gene causing a relatively mild form of Pelizaeus-Merzbacher disease in a Dutch family. <i>Human Genetics</i> , 1996, 97, 337-339.	1.8	56
43	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. <i>European Journal of Human Genetics</i> , 2015, 23, 1142-1150.	1.4	56
44	Variable phenotypes associated with 10q23 microdeletions involving the <i>PTEN</i> and <i>BMPR1A</i> genes. <i>Clinical Genetics</i> , 2008, 74, 145-154.	1.0	52
45	A detailed clinical analysis of 13 patients with AUTS2 syndrome further delineates the phenotypic spectrum and underscores the behavioural phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 523-532.	1.5	51
46	Severe hypomyelination associated with increased levels of N-acetylaspartylglutamate in CSF. <i>Neurology</i> , 2004, 62, 1503-1508.	1.5	49
47	Spastin mutations in sporadic adult-onset upper motor neuron syndromes. <i>Annals of Neurology</i> , 2005, 58, 865-869.	2.8	47
48	Low frequency of MECP2 mutations in mentally retarded males. <i>European Journal of Human Genetics</i> , 2002, 10, 487-490.	1.4	46
49	Haploinsufficiency of ANKRD11 causes mild cognitive impairment, short stature and minor dysmorphisms. <i>European Journal of Human Genetics</i> , 2012, 20, 131-133.	1.4	45
50	Diagnosis of Fanconi Anemia: Mutation Analysis by Next-Generation Sequencing. <i>Anemia</i> , 2012, 2012, 1-7.	0.5	44
51	Genotype-phenotype studies in three families with mutations in the polyglutamine-binding protein 1 gene (PQBP1). <i>Clinical Genetics</i> , 2004, 66, 318-326.	1.0	42
52	Identification of a Dutch founder mutation in MUSK causing fetal akinesia deformation sequence. <i>European Journal of Human Genetics</i> , 2015, 23, 1151-1157.	1.4	42
53	MECP2 analysis in mentally retarded patients: implications for routine DNA diagnostics. <i>European Journal of Human Genetics</i> , 2004, 12, 24-28.	1.4	41
54	Comparing methods for fetal fraction determination and quality control of NIPT samples. <i>Prenatal Diagnosis</i> , 2017, 37, 769-773.	1.1	41

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55	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
56	A mitochondrial tRNA ^{Val} gene mutation (G1642A) in a patient with mitochondrial myopathy, lactic acidosis, and stroke-like episodes. <i>Neurology</i> , 1998, 50, 293-295.	1.5	40
57	A newly recognised microdeletion syndrome involving 2p15p16.1: narrowing down the critical region by adding another patient detected by genome wide tiling path array comparative genomic hybridisation analysis. <i>Journal of Medical Genetics</i> , 2007, 45, 122-124.	1.5	40
58	Clinical impact of additional findings detected by genome-wide non-invasive prenatal testing: Follow-up results of the TRIDENT-2 study. <i>American Journal of Human Genetics</i> , 2022, 109, 1140-1152.	2.6	39
59	Inhibition of TGF β 2 signaling decreases osteogenic differentiation of fibrodysplasia ossificans progressiva fibroblasts in a novel in vitro model of the disease. <i>Bone</i> , 2016, 84, 169-180.	1.4	38
60	Diagnostic yield of a targeted gene panel in primary ciliary dyskinesia patients. <i>Human Mutation</i> , 2018, 39, 653-665.	1.1	38
61	Unusual cerebrotendinous xanthomatosis with fronto-temporal dementia phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2005, 139A, 114-117.	0.7	37
62	A triplication of the Williams-Beuren syndrome region in a patient with mental retardation, a severe expressive language delay, behavioural problems and dysmorphisms. <i>Journal of Medical Genetics</i> , 2010, 47, 271-275.	1.5	35
63	Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population. <i>European Journal of Human Genetics</i> , 2000, 8, 557-560.	1.4	34
64	Fetal fraction evaluation in non-invasive prenatal screening (NIPS). <i>European Journal of Human Genetics</i> , 2019, 27, 198-202.	1.4	34
65	Rare Genetic Variant in SORL1 May Increase Penetrance of Alzheimer's Disease in a Family with Several Generations of APOE ϵ 4 Homozygosity. <i>Journal of Alzheimer's Disease</i> , 2017, 56, 63-74.	1.2	32
66	Monitoring Standards for Molecular Genetic Testing in the United Kingdom, The Netherlands, and Ireland. <i>Genetic Testing and Molecular Biomarkers</i> , 2006, 10, 147-156.	1.7	29
67	Two male adults with pathogenic AUTS2 variants, including a two-base pair deletion, further delineate the AUTS2 syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 803-807.	1.4	28
68	Splice-site contribution in alternative splicing of PLP1 and DM20: molecular studies in oligodendrocytes. <i>Human Mutation</i> , 2006, 27, 69-77.	1.1	27
69	Altered PLP1 splicing causes hypomyelination of early myelinating structures. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 648-661.	1.7	27
70	Comprehensive multiparameter genetic analysis improves circulating tumor DNA detection in head and neck cancer patients. <i>Oral Oncology</i> , 2020, 109, 104852.	0.8	27
71	Melanocortin-4 Receptor Gene Mutations in a Dutch Cohort of Obese Children. <i>Obesity</i> , 2011, 19, 604-611.	1.5	26
72	Spinal phenotype of cerebrotendinous xanthomatosis. <i>Journal of Neurology</i> , 2004, 251, 105-107.	1.8	25

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73	A novel homozygous 5bp deletion in FKBP10 causes clinically Bruck syndrome in an Indonesian patient. <i>European Journal of Medical Genetics</i> , 2012, 55, 17-21.	0.7	25
74	Maternal Plasma DNA and RNA Sequencing for Prenatal Testing. <i>Advances in Clinical Chemistry</i> , 2016, 74, 63-102.	1.8	25
75	Uptake of fetal aneuploidy screening after the introduction of the noninvasive prenatal test: A national population-based register study. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2021, 100, 1265-1272.	1.3	25
76	A novel microdeletion in 1(p34.2p34.3), involving the <i>SLC2A1</i> (<i>GLUT1</i>) gene, and severe delayed development. <i>Developmental Medicine and Child Neurology</i> , 2007, 49, 380-384.	1.1	23
77	Noninvasive Prenatal Test Results Indicative of Maternal Malignancies: A Nationwide Genetic and Clinical Follow-Up Study. <i>Journal of Clinical Oncology</i> , 2022, 40, 2426-2435.	0.8	23
78	Position effect on PLP1 may cause a subset of Pelizaeus-Merzbacher disease symptoms. <i>Journal of Medical Genetics</i> , 2004, 41, e121-e121.	1.5	22
79	A novel microdeletion, del(2)(q22.3q23.3) in a mentally retarded patient, detected by array-based comparative genomic hybridization. <i>Clinical Genetics</i> , 2004, 65, 429-432.	1.0	22
80	Hypomyelination and Congenital Cataract. <i>Archives of Neurology</i> , 2011, 68, 1191.	4.9	22
81	Co-localization and functional coupling of creatine kinase B and gastric H ⁺ /K ⁺ -ATPase on the apical membrane and the tubulovesicular system of parietal cells. <i>Biochemical Journal</i> , 1995, 311, 445-451.	1.7	21
82	Interstitial 2.2 Mb deletion at 9q34 in a patient with mental retardation but without classical features of the 9q subtelomeric deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 618-623.	0.7	21
83	Recommended practice for laboratory reporting of noninvasive prenatal testing of trisomies 13, 18 and 21: a consensus opinion. <i>Prenatal Diagnosis</i> , 2017, 37, 699-704.	1.1	19
84	Holoprosencephaly and preaxial polydactyly associated with a 1.24 Mb duplication encompassing FBXW11 at 5q35.1. <i>Journal of Human Genetics</i> , 2006, 51, 721-726.	1.1	18
85	Routinization of prenatal screening with the non-invasive prenatal test: pregnant women's perspectives. <i>European Journal of Human Genetics</i> , 2022, 30, 661-668.	1.4	18
86	Localization of the gene (or genes) for a syndrome with X-linked mental retardation, ataxia, weakness, hearing impairment, loss of vision and a fatal course in early childhood. <i>Human Genetics</i> , 1996, 98, 513-517.	1.8	17
87	Isochromosome 21q is overrepresented among false-negative cell-free DNA prenatal screening results involving Down syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 1490-1496.	1.4	16
88	Association between low fetal fraction in cell-free DNA testing and adverse pregnancy outcome: A systematic review. <i>Prenatal Diagnosis</i> , 2021, 41, 1287-1295.	1.1	16
89	Ataxia with vitamin E deficiency: Biochemical effects of malcompliance with vitamin E therapy. <i>Neurology</i> , 2000, 55, 1584-1586.	1.5	14
90	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. <i>European Journal of Human Genetics</i> , 2008, 16, 395-400.	1.4	14

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91	Introducing WISECONDOR for noninvasive prenatal diagnostics. <i>Expert Review of Molecular Diagnostics</i> , 2014, 14, 513-515.	1.5	14
92	Mosaic maternal 10qter deletions are associated with FRA10B expansions and may cause false-positive noninvasive prenatal screening results. <i>Genetics in Medicine</i> , 2018, 20, 1472-1476.	1.1	14
93	Partial iris hypoplasia in a patient with an interstitial subtelomeric 6p deletion not including the forkhead transcription factor gene FOXC1. <i>European Journal of Human Genetics</i> , 2005, 13, 1169-1171.	1.4	13
94	Low fetal fraction in cell-free DNA testing is associated with adverse pregnancy outcome: Analysis of a subcohort of the TRIDENT-2 study. <i>Prenatal Diagnosis</i> , 2021, 41, 1296-1304.	1.1	13
95	First steps in exploring prospective exome sequencing of consanguineous couples. <i>European Journal of Medical Genetics</i> , 2014, 57, 613-616.	0.7	11
96	Cerebrotendinous xanthomatosis: report of two Brazilian brothers. <i>Arquivos De Neuro-Psiquiatria</i> , 2004, 62, 1085-1089.	0.3	10
97	Feasibility of preconception screening for thalassaemia in Indonesia: exploring the opinion of Javanese mothers. <i>Ethnicity and Health</i> , 2011, 16, 483-499.	1.5	9
98	p.Ser252Trp and p.Pro253Arg mutations in FGFR2 gene causing Apert syndrome: the first clinical and molecular report of Indonesian patients. <i>Singapore Medical Journal</i> , 2013, 54, e72-e75.	0.3	9
99	A unique second donor splice site in the intron 5 sequence of the HLA-A*11 alleles results in a class I transcript encoding a molecule with an elongated cytoplasmic domain. <i>Tissue Antigens</i> , 2000, 55, 422-428.	1.0	8
100	A novel 2.3% Mb microduplication of 12q24.21q24.23 detected by genome-wide tiling-path resolution array comparative genomic hybridization in a girl with syndromic mental retardation. <i>Clinical Dysmorphology</i> , 2006, 15, 133-137.	0.1	8
101	Noncoding RNA-regulated gain-of-function of STOX2 in Finnish pre-eclamptic families. <i>Scientific Reports</i> , 2016, 6, 32129.	1.6	8
102	Tissue- and cell-specific distribution of creatine kinase B: A new and highly specific monoclonal antibody for use in immunohistochemistry. <i>Cell and Tissue Research</i> , 1995, 280, 435-446.	1.5	8
103	Production of native creatine kinase B in insect cells using a baculovirus expression vector. <i>Molecular and Cellular Biochemistry</i> , 1995, 143, 59-65.	1.4	7
104	X-linked adrenomyeloneuropathy due to a novel missense mutation in the ABCD1 start codon presenting as demyelinating neuropathy. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 353-355.	1.4	7
105	Supporting the generalist genes hypothesis for intellectual ability/disability: the case of <sc>SNAP25</sc>. <i>Genes, Brain and Behavior</i> , 2012, 11, 767-771.	1.1	6
106	Susceptibility allele-specific loss of miR-1324-mediated silencing of the INO80B chromatin-assembly complex gene in pre-eclampsia. <i>Human Molecular Genetics</i> , 2015, 24, 118-127.	1.4	6
107	A novel <i>CCM2</i> variant in a family with non-progressive cognitive complaints and cerebral microbleeds. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2017, 174, 220-226.	1.1	6
108	Circular RNA Sequencing of Maternal Platelets: A Novel Tool for the Identification of Pregnancy-Specific Biomarkers. <i>Clinical Chemistry</i> , 2021, 67, 508-517.	1.5	6

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109	Clinically relevant DNA viruses in pregnancy. <i>Prenatal Diagnosis</i> , 2023, 43, 457-466.	1.1	6
110	The cell-free DNA virome of 108,349 Dutch pregnant women. <i>Prenatal Diagnosis</i> , 2023, 43, 448-456.	1.1	6
111	Presenile Cataract: Consider Cholestanol. <i>JAMA Ophthalmology</i> , 2006, 124, 1490.	2.6	5
112	A Novel GJC2 Mutation Associated with Hypomyelination and MÃ¼llerian Agenesis Syndrome: Coincidence or a New Entity?. <i>Neuropediatrics</i> , 2012, 43, 159-161.	0.3	5
113	WISExome: a within-sample comparison approach to detect copy number variations in whole exome sequencing data. <i>European Journal of Human Genetics</i> , 2017, 25, 1354-1363.	1.4	5
114	LEOPARD syndrome with partly normal skin and sex chromosome mosaicism. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2612-2615.	0.7	4
115	Maternal Malignancies Detected With Noninvasive Prenatal Testing. <i>JAMA - Journal of the American Medical Association</i> , 2015, 314, 2192.	3.8	4
116	The clinical benefit of genome-wide cfDNA testing cannot be extrapolated from CVS data. <i>Genetics in Medicine</i> , 2020, 22, 657-658.	1.1	4
117	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. <i>Human Mutation</i> , 2017, 38, 594-599.	1.1	3
118	Fragmentomic cfDNA Patterns in Noninvasive Prenatal Testing and Beyond. <i>Journal of Biomedicine and Translational Research</i> , 2021, 7, 38-47.	0.2	3
119	The bivariate NRIP1/ZEB2 RNA marker permits non-invasive presymptomatic screening of pre-eclampsia. <i>Scientific Reports</i> , 2020, 10, 21857.	1.6	3
120	Complete sequence analysis of the A*1103 allele. <i>Tissue Antigens</i> , 2000, 55, 68-70.	1.0	2
121	Use of TaqI Digestion May Lead to Incorrect Molecular Diagnosis of Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. <i>Molecular Genetics and Metabolism</i> , 2000, 70, 322-324.	0.5	2
122	A severe connatal form of Pelizaeus Merzbacher disease in a Czech boy caused by a novel mutation (725C>A, Ala242Glu) at the <sc>Tjimpym> codon</sc> in the PLP gene. <i>International Journal of Molecular Medicine</i> , 2002, 9, 125.	1.8	2
123	WisecondorFF: Improved Fetal Aneuploidy Detection from Shallow WGS through Fragment Length Analysis. <i>Diagnostics</i> , 2022, 12, 59.	1.3	2
124	Response to letter to the editor <sc>PD>0390</sc>, a comment on âœComparing methods for fetal fraction determination and quality control of NIPT samples>. <i>Prenatal Diagnosis</i> , 2017, 37, 1266-1267.	1.1	1
125	The Importance of Reliable Quality Control Materials for Noninvasive Prenatal Testing. <i>Clinical Chemistry</i> , 2019, 65, 720-722.	1.5	1
126	Non-invasive prenatal diagnosis for translocation carriersâ€”YES please or NO go?. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2021, 100, 2036-2043.	1.3	1

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127	Novel mutation in the SPAST gene in a patient with spastic paraparesis. Journal of Neurology, 2008, 255, 303-304.	1.8	0
128	N-Acetylaspartylglutamate in CNS Hypomyelination. Neuropediatrics, 2011, 42, 74-77.	0.3	0
129	Subtelomeric chromosomal anomalies in infantile epileptic encephalopathies. Journal of Pediatric Neurology, 2015, 08, 391-396.	0.0	0
130	Cover Image, Volume 36, Issue 7. Prenatal Diagnosis, 2016, 36, i.	1.1	0