Erik A Sistermans

List of Publications by Year
in descending order

130 papers	8,689 citations	50276 46 h-index	46799 89 g-index
133 all docs	133 docs citations	133 times ranked	12229 citing authors

#	Article	IF	Citations
1	Clinically relevant DNA viruses in pregnancy. Prenatal Diagnosis, 2023, 43, 457-466.	2.3	6
2	The cellâ€free DNA virome of 108,349 Dutch pregnant women. Prenatal Diagnosis, 2023, 43, 448-456.	2.3	6
3	Routinization of prenatal screening with theÂnon-invasive prenatal test: pregnant womenâ€™s perspectives. European Journal of Human Genetics, 2022, 30, 661-668.	2.8	18
4	Noninvasive Prenatal Test Results Indicative of Maternal Malignancies: A Nationwide Genetic and Clinical Follow-Up Study. Journal of Clinical Oncology, 2022, 40, 2426-2435.	1.6	23
5	WisecondorFF: Improved Fetal Aneuploidy Detection from Shallow WGS through Fragment Length Analysis. Diagnostics, 2022, 12, 59.	2.6	2
6	Clinical impact of additional findings detected by genome-wide non-invasive prenatal testing: Follow-up results of the TRIDENT-2 study. American Journal of Human Genetics, 2022, 109, 1140-1152.	6.2	39
7	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
8	Circular RNA Sequencing of Maternal Platelets: A Novel Tool for the Identification of Pregnancy-Specific Biomarkers. Clinical Chemistry, 2021, 67, 508-517.	3.2	6
9	Uptake of fetal aneuploidy screening after the introduction of the nonâ€invasive prenatal test: A national populationâ€based register study. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 1265-1272.	2.8	25
10	Fragmentomic cfDNA Patterns in Noninvasive Prenatal Testing and Beyond. Journal of Biomedicine and Translational Research, 2021, 7, 38-47.	0.2	3
11	Association between low fetal fraction in cellâ€free DNA testing and adverse pregnancy outcome: A systematic review. Prenatal Diagnosis, 2021, 41, 1287-1295.	2.3	16
12	Nonâ€invasive prenatal diagnosis for translocation carriersâ€”YES please or NO go?. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 2036-2043.	2.8	1
13	Low fetal fraction in cellâ€free DNA testing is associated with adverse pregnancy outcome: Analysis of a subcohort of the TRIDENTâ€2 study. Prenatal Diagnosis, 2021, 41, 1296-1304.	2.3	13
14	The clinical benefit of genome-wide cfDNA testing cannot be extrapolated from CVS data. Genetics in Medicine, 2020, 22, 657-658.	2.4	4
15	Comprehensive multiparameter genetic analysis improves circulating tumor DNA detection in head and neck cancer patients. Oral Oncology, 2020, 109, 104852.	1.5	27
16	The bivariate NRIP1/ZEB2 RNA marker permits non-invasive presymptomatic screening of pre-eclampsia. Scientific Reports, 2020, 10, 21857.	3.3	3
17	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. American Journal of Human Genetics, 2019, 105, 1091-1101.	6.2	222
18	The Importance of Reliable Quality Control Materials for Noninvasive Prenatal Testing. Clinical Chemistry, 2019, 65, 720-722.	3.2	1

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19	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
20	Fetal fraction evaluation in non-invasive prenatal screening (NIPS). European Journal of Human Genetics, 2019, 27, 198-202.	2.8	34
21	Mosaic maternal 10qter deletions are associated with FRA10B expansions and may cause false-positive noninvasive prenatal screening results. Genetics in Medicine, 2018, 20, 1472-1476.	2.4	14
22	Diagnostic yield of a targeted gene panel in primary ciliary dyskinesia patients. Human Mutation, 2018, 39, 653-665.	2.5	38
23	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.	2.4	85
24	Isochromosome 21q is overrepresented among false-negative cell-free DNA prenatal screening results involving Down syndrome. European Journal of Human Genetics, 2018, 26, 1490-1496.	2.8	16
25	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.	6.2	136
26	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	2.5	3
27	Recommended practice for laboratory reporting of nonâ€invasive prenatal testing of trisomies 13, 18 and 21: a consensus opinion. Prenatal Diagnosis, 2017, 37, 699-704.	2.3	19
28	Rare Genetic Variant in SORL1 May Increase Penetrance of Alzheimerâ€™s Disease in a Family with Several Generations of APOE-É›4 Homozygosity. Journal of Alzheimer's Disease, 2017, 56, 63-74.	2.6	32
29	Comparing methods for fetal fraction determination and quality control of NIPT samples. Prenatal Diagnosis, 2017, 37, 769-773.	2.3	41
30	A novel CCM2 variant in a family with nonâ€progressive cognitive complaints and cerebral microbleeds. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 220-226.	1.7	6
31	WISExome: a within-sample comparison approach to detect copy number variations in whole exome sequencing data. European Journal of Human Genetics, 2017, 25, 1354-1363.	2.8	5
32	Response to letter to the editor <scp>PDâ€17â€0390</scp>, a comment on â€œComparing methods for fetal fraction determination and quality control of NIPT samplesâ€• Prenatal Diagnosis, 2017, 37, 1266-1267.	2.3	1
33	Calculating the fetal fraction for noninvasive prenatal testing based on genomeâ€wide nucleosome profiles. Prenatal Diagnosis, 2016, 36, 614-621.	2.3	76
34	A detailed clinical analysis of 13 patients with AUTS2 syndrome further delineates the phenotypic spectrum and underscores the behavioural phenotype. Journal of Medical Genetics, 2016, 53, 523-532.	3.2	51
35	Maternal Plasma DNA and RNA Sequencing for Prenatal Testing. Advances in Clinical Chemistry, 2016, 74, 63-102.	3.7	25
36	Trial by Dutch laboratories for evaluation of nonâ€invasive prenatal testing. Part Iâ€”clinical impact. Prenatal Diagnosis, 2016, 36, 1083-1090.	2.3	122

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37	Noncoding RNA-regulated gain-of-function of STOX2 in Finnish pre-eclamptic families. Scientific Reports, 2016, 6, 32129.	3.3	8
38	Cover Image, Volume 36, Issue 7. Prenatal Diagnosis, 2016, 36, i.	2.3	0
39	Inhibition of TGFÎ² signaling decreases osteogenic differentiation of fibrodysplasia ossificans progressiva fibroblasts in a novel in vitro model of the disease. Bone, 2016, 84, 169-180.	2.9	38
40	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	2.8	389
41	Subtelomeric chromosomal anomalies in infantile epileptic encephalopathies. Journal of Pediatric Neurology, 2015, 08, 391-396.	0.2	0
42	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
43	SMAD2Mutations Are Associated with Arterial Aneurysms and Dissections. Human Mutation, 2015, 36, 1145-1149.	2.5	74
44	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. European Journal of Human Genetics, 2015, 23, 1142-1150.	2.8	56
45	Susceptibility allele-specific loss of miR-1324-mediated silencing of the INO80B chromatin-assembly complex gene in pre-eclampsia. Human Molecular Genetics, 2015, 24, 118-127.	2.9	6
46	Identification of a Dutch founder mutation in MUSK causing fetal akinesia deformation sequence. European Journal of Human Genetics, 2015, 23, 1151-1157.	2.8	42
47	Maternal Malignancies Detected With Noninvasive Prenatal Testing. JAMA - Journal of the American Medical Association, 2015, 314, 2192.	7.4	4
48	Two male adults with pathogenic AUTS2 variants, including a two-base pair deletion, further delineate the AUTS2 syndrome. European Journal of Human Genetics, 2015, 23, 803-807.	2.8	28
49	WISECONDOR: detection of fetal aberrations from shallow sequencing maternal plasma based on a within-sample comparison scheme. Nucleic Acids Research, 2014, 42, e31-e31.	14.5	124
50	Introducing WISECONDOR for noninvasive prenatal diagnostics. Expert Review of Molecular Diagnostics, 2014, 14, 513-515.	3.1	14
51	Characteristic brain magnetic resonance imaging pattern in patients with macrocephaly and PTEN mutations. American Journal of Medical Genetics, Part A, 2014, 164, 627-633.	1.2	70
52	Somatic mutations found in the healthy blood compartment of a 115-yr-old woman demonstrate oligoclonal hematopoiesis. Genome Research, 2014, 24, 733-742.	5.5	136
53	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170
54	First steps in exploring prospective exome sequencing of consanguineous couples. European Journal of Medical Genetics, 2014, 57, 613-616.	1.3	11

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55	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.	6.2	122
56	PLS3 Mutations in X-Linked Osteoporosis with Fractures. New England Journal of Medicine, 2013, 369, 1529-1536.	27.0	171
57	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.	6.2	135
58	p.Ser252Trp and p.Pro253Arg mutations in FGFR2 gene causing Apert syndrome: the first clinical and molecular report of Indonesian patients. Singapore Medical Journal, 2013, 54, e72-e75.	0.6	9
59	Haploinsufficiency of ANKRD11 causes mild cognitive impairment, short stature and minor dysmorphisms. European Journal of Human Genetics, 2012, 20, 131-133.	2.8	45
60	A Novel GJC2 Mutation Associated with Hypomyelination and MÃ¼llerian Agenesis Syndrome: Coincidence or a New Entity?. Neuropediatrics, 2012, 43, 159-161.	0.6	5
61	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
62	Diagnosis of Fanconi Anemia: Mutation Analysis by Next-Generation Sequencing. Anemia, 2012, 2012, 1-7.	1.7	44
63	Supporting the generalist genes hypothesis for intellectual ability/disability: the case of <scp>SNAP25</scp>. Genes, Brain and Behavior, 2012, 11, 767-771.	2.2	6
64	A novel homozygous 5bp deletion in FKBP10 causes clinically Bruck syndrome in an Indonesian patient. European Journal of Medical Genetics, 2012, 55, 17-21.	1.3	25
65	X-linked adrenomyeloneuropathy due to a novel missense mutation in the ABCD1 start codon presenting as demyelinating neuropathy. Journal of the Peripheral Nervous System, 2011, 16, 353-355.	3.1	7
66	Melanocortinâ€4 Receptor Gene Mutations in a Dutch Cohort of Obese Children. Obesity, 2011, 19, 604-611.	3.0	26
67	N-Acetylaspartylglutamate in CNS Hypomyelination. Neuropediatrics, 2011, 42, 74-77.	0.6	0
68	Feasibility of preconception screening for thalassaemia in Indonesia: exploring the opinion of Javanese mothers. Ethnicity and Health, 2011, 16, 483-499.	2.5	9
69	Hypomyelination and Congenital Cataract. Archives of Neurology, 2011, 68, 1191.	4.5	22
70	A triplication of the Williams-Beuren syndrome region in a patient with mental retardation, a severe expressive language delay, behavioural problems and dysmorphisms. Journal of Medical Genetics, 2010, 47, 271-275.	3.2	35
71	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. Human Mutation, 2009, 30, 283-292.	2.5	136
72	PPIB Mutations Cause Severe Osteogenesis Imperfecta. American Journal of Human Genetics, 2009, 85, 521-527.	6.2	257

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73	Smoothing waves in array CGH tumor profiles. Bioinformatics, 2009, 25, 1099-1104.	4.1	76
74	Novel mutation in the SPAST gene in a patient with spastic paraparesis. Journal of Neurology, 2008, 255, 303-304.	3.6	0
75	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. European Journal of Human Genetics, 2008, 16, 395-400.	2.8	14
76	Variable phenotypes associated with 10q23 microdeletions involving the PTEN and BMPR1A genes. Clinical Genetics, 2008, 74, 145-154.	2.0	52
77	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.	3.2	87
78	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. Journal of Medical Genetics, 2007, 45, 122-124.	3.2	40
79	L1 retrotransposition can occur early in human embryonic development. Human Molecular Genetics, 2007, 16, 1587-1592.	2.9	174
80	LEOPARD syndrome with partly normal skin and sex chromosome mosaicism. American Journal of Medical Genetics, Part A, 2007, 143A, 2612-2615.	1.2	4
81	A novel microdeletion in 1(p34.2p34.3), involving the SLC2A1 (GLUT1) gene, and severe delayed development. Developmental Medicine and Child Neurology, 2007, 49, 380-384.	2.1	23
82	Monitoring Standards for Molecular Genetic Testing in the United Kingdom, The Netherlands, and Ireland. Genetic Testing and Molecular Biomarkers, 2006, 10, 147-156.	1.7	29
83	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. American Journal of Human Genetics, 2006, 79, 370-377.	6.2	343
84	A novel 2.3â€‰Mb microduplication of 12q24.21q24.23 detected by genome-wide tiling-path resolution array comparative genomic hybridization in a girl with syndromic mental retardation. Clinical Dysmorphology, 2006, 15, 133-137.	0.3	8
85	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	21.4	670
86	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
87	Holoprosencephaly and preaxial polydactyly associated with a 1.24ÂMb duplication encompassing FBXW11 at 5q35.1. Journal of Human Genetics, 2006, 51, 721-726.	2.3	18
88	Splice-site contribution in alternative splicing ofPLP1 andDM20: molecular studies in oligodendrocytes. Human Mutation, 2006, 27, 69-77.	2.5	27
89	Interstitial 2.2 Mb deletion at 9q34 in a patient with mental retardation but without classical features of the 9q subtelomeric deletion syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 618-623.	1.2	21
90	Presenile Cataract: Consider Cholestanol. JAMA Ophthalmology, 2006, 124, 1490.	2.4	5

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91	Partial iris hypoplasia in a patient with an interstitial subtelomeric 6p deletion not including the forkhead transcription factor gene FOXC1. European Journal of Human Genetics, 2005, 13, 1169-1171.	2.8	13
92	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.	1.2	101
93	Unusual cerebrotendinous xanthomatosis with fronto-temporal dementia phenotype. American Journal of Medical Genetics, Part A, 2005, 139A, 114-117.	1.2	37
94	Spastin mutations in sporadic adult-onset upper motor neuron syndromes. Annals of Neurology, 2005, 58, 865-869.	5.3	47
95	Disruption of the gene Euchromatin Histone Methyl Transferase1 (Eu-HMTase1) is associated with the 9q34 subtelomeric deletion syndrome. Journal of Medical Genetics, 2005, 42, 299-306.	3.2	162
96	Three or more copies of the proteolipid protein gene PLP1 cause severe Pelizaeus-Merzbacher disease. Brain, 2005, 128, 743-751.	7.6	91
97	3q29 Microdeletion Syndrome: Clinical and Molecular Characterization of a New Syndrome. American Journal of Human Genetics, 2005, 77, 154-160.	6.2	228
98	Diagnostic Genome Profiling in Mental Retardation. American Journal of Human Genetics, 2005, 77, 606-616.	6.2	514
99	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.	6.2	93
100	Cerebrotendinous xanthomatosis: report of two Brazilian brothers. Arquivos De Neuro-Psiquiatria, 2004, 62, 1085-1089.	0.8	10
101	Position effect on PLP1 may cause a subset of Pelizaeus-Merzbacher disease symptoms. Journal of Medical Genetics, 2004, 41, e121-e121.	3.2	22
102	Severe hypomyelination associated with increased levels of N -acetylaspartylglutamate in CSF. Neurology, 2004, 62, 1503-1508.	1.1	49
103	Screening for subtelomeric rearrangements in 210 patients with unexplained mental retardation using multiplex ligation dependent probe amplification (MLPA). Journal of Medical Genetics, 2004, 41, 892-899.	3.2	136
104	Genotype-phenotype studies in three families with mutations in the polyglutamine-binding protein 1 gene (PQBP1). Clinical Genetics, 2004, 66, 318-326.	2.0	42
105	A novel microdeletion, del(2)(q22.3q23.3) in a mentally retarded patient, detected by array-based comparative genomic hybridization. Clinical Genetics, 2004, 65, 429-432.	2.0	22
106	MECP2 analysis in mentally retarded patients: implications for routine DNA diagnostics. European Journal of Human Genetics, 2004, 12, 24-28.	2.8	41
107	Spinal phenotype of cerebrotendinous xanthomatosis. Journal of Neurology, 2004, 251, 105-107.	3.6	25
108	CYP21 Gene Mutation Analysis in 198 Patients with 21-Hydroxylase Deficiency in The Netherlands: Six Novel Mutations and a Specific Cluster of Four Mutations. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3852-3859.	3.6	154

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109	A severe connatal form of Pelizaeus Merzbacher disease in a Czech boy caused by a novel mutation (725C>A, Ala242Glu) at the <jimpymsd codon> in the PLP gene. International Journal of Molecular Medicine, 2002, 9, 125.	4.0	2
110	Patients lacking the major CNS myelin protein, proteolipid protein 1, develop length-dependent axonal degeneration in the absence of demyelination and inflammation. Brain, 2002, 125, 551-561.	7.6	272
111	MECP2 Mutation in a Boy with Severe Neonatal Encephalopathy: Clinical, Neuropathological and Molecular Findings. Neuropediatrics, 2002, 33, 33-36.	0.6	63
112	Deâ€ƒnovo MECP2 frameshift mutation in a boy with moderate mental retardation, obesity and gynaecomastia. Clinical Genetics, 2002, 61, 359-362.	2.0	59
113	Low frequency of MECP2 mutations in mentally retarded males. European Journal of Human Genetics, 2002, 10, 487-490.	2.8	46
114	Mutations in the sterol 27-hydoxylase gene (CYP27A) cause hepatitis of infancy as well as cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2002, 25, 501-513.	3.6	99
115	Complete sequence analysis of the A*1103 allele. Tissue Antigens, 2000, 55, 68-70.	1.0	2
116	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. Tissue Antigens, 2000, 55, 422-428.	1.0	8
117	Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population. European Journal of Human Genetics, 2000, 8, 557-560.	2.8	34
118	Ataxia with vitamin E deficiency: Biochemical effects of malcompliance with vitamin E therapy. Neurology, 2000, 55, 1584-1586.	1.1	14
119	Mutations in noncoding regions of the proteolipid protein gene in Pelizaeusâ€“Merzbacher disease. Neurology, 2000, 55, 1089-1096.	1.1	57
120	Use of TaqI Digestion May Lead to Incorrect Molecular Diagnosis of Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. Molecular Genetics and Metabolism, 2000, 70, 322-324.	1.1	2
121	Imprinting Effect in Premature Ovarian Failure Confined to Paternally Inherited Fragile X Premutations. American Journal of Human Genetics, 2000, 66, 413-418.	6.2	93
122	A mitochondrial tRNA ^{Val} gene mutation (G1642A) in a patient with mitochondrial myopathy, lactic acidosis, and stroke-like episodes. Neurology, 1998, 50, 293-295.	1.1	40
123	Duplication of the proteolipid protein gene is the major cause of Pelizaeusâ€Merzbacher disease. Neurology, 1998, 50, 1749-1754.	1.1	141
124	Rapid antibody test for diagnosing fragile X syndrome: a validation of the technique. Human Genetics, 1997, 99, 308-311.	3.8	115
125	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. Human Genetics, 1996, 97, 337-339.	3.8	56
126	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. Human Genetics, 1996, 98, 513-517.	3.8	17

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127	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. Biochemical Journal, 1995, 311, 445-451.	3.7	21
128	Production of native creatine kinase B in insect cells using a baculovirus expression vector. Molecular and Cellular Biochemistry, 1995, 143, 59-65.	3.1	7
129	Tissue- and cell-specific distribution of creatine kinase B: A new and highly specific monoclonal antibody for use in immunohistochemistry. Cell and Tissue Research, 1995, 280, 435-446.	2.9	62
130	Tissue- and cell-specific distribution of creatine kinase B: A new and highly specific monoclonal antibody for use in immunohistochemistry. Cell and Tissue Research, 1995, 280, 435-446.	2.9	8