

Hilde Peeters

List of Publications by Year in descending order

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Version: 2024-02-01

70
papers

3,962
citations

218381

26
h-index

138251

58
g-index

71
all docs

71
docs citations

71
times ranked

7988
citing authors

#	ARTICLE	IF	CITATIONS
1	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
3	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
4	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	9.4	229
5	Validation of a telephone zygosity questionnaire in twins of known zygosity. Behavior Genetics, 1998, 28, 159-163.	1.4	185
6	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
7	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
8	Human laterality disorders. European Journal of Medical Genetics, 2006, 49, 349-362.	0.7	134
9	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
10	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
11	Insights into the genetic architecture of the human face. Nature Genetics, 2021, 53, 45-53.	9.4	94
12	Genetic testing and counseling in the case of an autism diagnosis: A caregivers perspective. European Journal of Medical Genetics, 2016, 59, 452-458.	0.7	64
13	Predicting fetoplacental chromosomal mosaicism during noninvasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	1.1	58
14	Shared heritability of human face and brain shape. Nature Genetics, 2021, 53, 830-839.	9.4	57
15	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
16	Shooting a moving target. Researching autism genes: An interview study with professionals. European Journal of Medical Genetics, 2016, 59, 32-38.	0.7	53
17	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
18	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

#	ARTICLE	IF	CITATIONS
19	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
20	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. <i>Genetics in Medicine</i> , 2015, 17, 460-466.	1.1	45
21	Narrowing the critical deletion region for autism spectrum disorders on 16p11.2. , 2011, 156, 243-245.		44
22	<i>CREBBP</i> mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2681-2693.	0.7	43
23	Multiple pilomatricomas with somatic <i>CTNNB1</i> mutations in children with constitutive mismatch repair deficiency. <i>Genes Chromosomes and Cancer</i> , 2013, 52, 656-664.	1.5	40
24	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	2.6	40
25	Interest in expanded carrier screening among individuals and couples in the general population: systematic review of the literature. <i>Human Reproduction Update</i> , 2020, 26, 335-355.	5.2	36
26	The East Flanders Prospective Twin Survey (EFPTS): An Actual Perception. <i>Twin Research and Human Genetics</i> , 2013, 16, 58-63.	0.3	34
27	Homozygous loss-of-function mutation in ALMS1 causes the lethal disorder mitogenic cardiomyopathy in two siblings. <i>European Journal of Medical Genetics</i> , 2014, 57, 532-535.	0.7	31
28	Platelet studies in autism spectrum disorder patients and first-degree relatives. <i>Molecular Autism</i> , 2015, 6, 57.	2.6	28
29	The ethics of complexity. Genetics and autism, a literature review. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 305-316.	1.1	23
30	The East Flanders Prospective Twin Survey (EFPTS): 55 Years Later. <i>Twin Research and Human Genetics</i> , 2019, 22, 454-459.	0.3	23
31	Executive functioning and localâ€“global visual processing: candidate endophenotypes for autism spectrum disorder?. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2017, 58, 258-269.	3.1	21
32	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	3.7	21
33	Six NSCL/P Loci Show Associations With Normal-Range Craniofacial Variation. <i>Frontiers in Genetics</i> , 2018, 9, 502.	1.1	20
34	Compound heterozygous loss-of-function mutations in KIF20A are associated with a novel lethal congenital cardiomyopathy in two siblings. <i>PLoS Genetics</i> , 2018, 14, e1007138.	1.5	18
35	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. <i>American Journal of Human Genetics</i> , 2020, 107, 963-976.	2.6	18
36	Large-scale open-source three-dimensional growth curves for clinical facial assessment and objective description of facial dysmorphism. <i>Scientific Reports</i> , 2021, 11, 12175.	1.6	17

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37	Sesn1 is a novel gene for left-right asymmetry and mediating nodal signaling. <i>Human Molecular Genetics</i> , 2006, 15, 3369-3377.	1.4	16
38	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. <i>Genetics in Medicine</i> , 2019, 21, 2774-2780.	1.1	16
39	Time trends in the natural dizygotic twinning rate. <i>Human Reproduction</i> , 2011, 26, 2247-2252.	0.4	15
40	NRXN1 \pm is associated with increased excitability in ASD iPSC-derived neurons. <i>BMC Neuroscience</i> , 2021, 22, 56.	0.8	14
41	The Influence of Microdeletions and Microduplications of 16p11.2 on Global Transcription Profiles. <i>Journal of Child Neurology</i> , 2015, 30, 1947-1953.	0.7	13
42	ZNF462 and KLF12 are disrupted by a de novo translocation in a patient with syndromic intellectual disability and autism spectrum disorder. <i>European Journal of Medical Genetics</i> , 2018, 61, 376-383.	0.7	13
43	3D facial phenotyping by biometric sibling matching used in contemporary genomic methodologies. <i>PLoS Genetics</i> , 2021, 17, e1009528.	1.5	13
44	Spatially Dense 3D Facial Heritability and Modules of Co-heritability in a Father-Offspring Design. <i>Frontiers in Genetics</i> , 2018, 9, 554.	1.1	12
45	Recurrent involvement of chromosomal region 6q21 in heterotaxy. <i>American Journal of Medical Genetics Part A</i> , 2001, 103, 44-47.	2.4	11
46	Focal preauricular dermal dysplasia: distinctive congenital lesions with a bilateral and symmetric distribution. <i>European Journal of Medical Genetics</i> , 2006, 49, 135-139.	0.7	11
47	Homozygous missense mutation in STYXL1 associated with moderate intellectual disability, epilepsy and behavioural complexities. <i>European Journal of Medical Genetics</i> , 2015, 58, 205-210.	0.7	11
48	The clinical relevance of intragenic NRXN1 deletions. <i>Journal of Medical Genetics</i> , 2020, 57, 347-355.	1.5	11
49	The Intersection of the Genetic Architectures of Orofacial Clefts and Normal Facial Variation. <i>Frontiers in Genetics</i> , 2021, 12, 626403.	1.1	10
50	Testing the face shape hypothesis in twins discordant for nonsyndromic orofacial clefting. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2886-2892.	0.7	7
51	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1897-1909.	0.7	7
52	Expanded carrier screening in Flanders (Belgium): an online survey on the perspectives of nonpregnant reproductive-aged women. <i>Personalized Medicine</i> , 2021, 18, 361-373.	0.8	6
53	Variants in TTC25 affect autistic trait in patients with autism spectrum disorder and general population. <i>European Journal of Human Genetics</i> , 2017, 25, 982-987.	1.4	5
54	Knowledge, attitudes and preferences regarding reproductive genetic carrier screening among reproductive-aged men and women in Flanders (Belgium). <i>European Journal of Human Genetics</i> , 2022, , .	1.4	5

#	ARTICLE	IF	CITATIONS
55	Reasons affecting the uptake of reproductive genetic carrier screening among nonpregnant reproductive-aged women in Flanders (Belgium). <i>Journal of Genetic Counseling</i> , 2022, 31, 1043-1053.	0.9	5
56	The ethics of patenting autism genes. <i>Nature Reviews Genetics</i> , 2018, 19, 247-248.	7.7	3
57	Multi-Scale Part-Based Syndrome Classification of 3D Facial Images. <i>IEEE Access</i> , 2022, 10, 23450-23462.	2.6	3
58	Orthognathic surgery in patients with systemic diseases. <i>Oral and Maxillofacial Surgery</i> , 2023, 27, 235-243.	0.6	3
59	Usefulness of fragile X checklist and CGG distribution in specialized institutions in Kinshasa, DR Congo. <i>Journal of Community Genetics</i> , 2019, 10, 153-159.	0.5	2
60	Frequency and Management of Craniofacial Syndromes. <i>Journal of Craniofacial Surgery</i> , 2020, 31, 1091-1097.	0.3	2
61	Two female siblings with congenital heart disease, postaxial polydactyly, ectopic neurohypophyseal gland, hair anomalies and characteristic facial features: a new syndrome?. <i>Clinical Dysmorphology</i> , 2006, 15, 71-74.	0.1	1
62	Accuracy and Clinical Value of Maternal Incidental Findings During Noninvasive Prenatal Testing for Fetal Aneuploidies. <i>Obstetrical and Gynecological Survey</i> , 2017, 72, 469-470.	0.2	1
63	Olfactory function in patients with nonsyndromic orofacial clefts and their unaffected relatives. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2375-2381.	0.7	1
64	Maxillofacial and oral surgery in patients with thrombophilia: safe territory for the oral surgeon? A single-center retrospective study. <i>Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology</i> , 2021, 132, 514-522.	0.2	1
65	8p21.3 deletions are rare causes of non-syndromic autism spectrum disorder. <i>Neurogenetics</i> , 2021, 22, 207-213.	0.7	1
66	3D analysis of facial morphology in Dutch children with cancer. <i>Computer Methods and Programs in Biomedicine</i> , 2021, 205, 106093.	2.6	1
67	Aarskog syndrome: from prenatal features towards postnatal diagnosis. <i>Ultrasound</i> , 2011, 19, 107-109.	0.3	0
68	Noise-robust assessment of SNP array based CNV calls through local noise estimation of log R ratios. <i>Statistical Applications in Genetics and Molecular Biology</i> , 2018, 17, .	0.2	0
69	A de-novo 15q24.2 deletion involving SIN3A is associated with emotional, behavioural, motor problems and hypersensitivity in a girl with above average intelligence and typical facial features.. <i>Clinical Dysmorphology</i> , 2020, 29, 210-213.	0.1	0
70	Abstract P6-08-03: Germline mutational landscape in 5422 individuals at risk for hereditary breast and ovarian cancer who underwent multi-gene panel testing. , 2020, , .		0