Hilde Van Esch

List of Publications by Year
in descending order

75 papers	3,048 citations	136950 32 h-index	189892 50 g-index
76 all docs	76 docs citations	76 times ranked	7169 citing authors

#	Article	IF	Citations
1	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
2	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
3	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
4	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
5	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
6	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	21.4	105
7	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	2.5	92
8	Quaking promotes monocyte differentiation into pro-atherogenic macrophages by controlling pre-mRNA splicing and gene expression. Nature Communications, 2016, 7, 10846.	12.8	87
9	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	6.2	82
10	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
11	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
12	Towards a 21st-century roadmap for biomedical research and drug discovery: consensus report and recommendations. Drug Discovery Today, 2017, 22, 327-339.	6.4	64
13	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63
14	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
15	Screening for FMR-1 premutations in 122 older Flemish males presenting with ataxia. European Journal of Human Genetics, 2005, 13, 121-123.	2.8	58
16	Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	2.3	58
17	Gain-of-function FHF1 mutation causes early-onset epileptic encephalopathy with cerebellar atrophy. Neurology, 2016, 86, 2162-2170.	1.1	57
18	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56

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19	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55
20	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
21	Positron Emission Tomography (PET) Quantification of GABAA Receptors in the Brain of Fragile X Patients. PLoS ONE, 2015, 10, e0131486.	2.5	52
22	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
23	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
24	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
25	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
26	Use of Multimodal Imaging and Clinical Biomarkers in Presymptomatic Carriers of C9orf72 Repeat Expansion. JAMA Neurology, 2020, 77, 1008.	9.0	45
27	A novel fragile X syndrome mutation reveals a conserved role for the carboxyâ€terminus in <scp>FMRP</scp> localization and function. EMBO Molecular Medicine, 2015, 7, 423-437.	6.9	41
28	Early myoclonic encephalopathy caused by a disruption of the neuregulin-1 receptor ErbB4. European Journal of Human Genetics, 2009, 17, 378-382.	2.8	40
29	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38
30	A novel SYN1 missense mutation in non-syndromic X-linked intellectual disability affects synaptic vesicle life cycle, clustering and mobility. Human Molecular Genetics, 2017, 26, 4699-4714.	2.9	37
31	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331.	2.5	37
32	Microdeletion of the escape genes KDM5C and IQSEC2 in a girl with severe intellectual disability and autistic features. European Journal of Medical Genetics, 2015, 58, 324-327.	1.3	36
33	Partial DiGeorge syndrome in two patients with a 10p rearrangement. Clinical Genetics, 1999, 55, 269-276.	2.0	34
34	Congenital diaphragmatic hernia is part of the new 15q24 microdeletion syndrome. European Journal of Medical Genetics, 2009, 52, 153-156.	1.3	33
35	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
36	Defective DNA Polymerase Î±-Primase Leads to X-Linked Intellectual Disability Associated with Severe Growth Retardation, Microcephaly, and Hypogonadism. American Journal of Human Genetics, 2019, 104, 957-967.	6.2	32

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37	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	2.5	30
38	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
39	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
40	Microdeletions on 6p22.3 are associated with mesomelic dysplasia Savarirayan type. Journal of Medical Genetics, 2015, 52, 476-483.	3.2	27
41	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
42	Pseudoautosomal Region 1 Length Polymorphism in the Human Population. PLoS Genetics, 2014, 10, e1004578.	3.5	24
43	Presenting symptoms in adults with the 22q11 deletion syndrome. European Journal of Medical Genetics, 2014, 57, 157-162.	1.3	24
44	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
45	Developmental delay and connective tissue disorder in four patients sharing a common microdeletion at 6q13-14. Journal of Medical Genetics, 2010, 47, 717-720.	3.2	22
46	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	2.9	20
47	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
48	No evidence of locus heterogeneity in familial microcephaly with or without chorioretinopathy, lymphedema, or mental retardation syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 52.	2.7	16
49	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	2.5	16
50	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
51	Very early premature ovarian failure in two sisters compound heterozygous for the FMR1 premutation. European Journal of Medical Genetics, 2009, 52, 37-40.	1.3	15
52	3 generation pedigree with paternal transmission of the 22q11.2 deletion syndrome: Intrafamilial phenotypic variability. European Journal of Medical Genetics, 2015, 58, 244-248.	1.3	15
53	Novel STIL Compound Heterozygous Mutations Cause Severe Fetal Microcephaly and Centriolar Lengthening. Molecular Syndromology, 2017, 8, 282-293.	0.8	15
54	Focus group discussions on secondary variants and next-generation sequencing technologies. European Journal of Medical Genetics, 2015, 58, 249-257.	1.3	14

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55	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
56	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
57	ZNF462 and KLF12 are disrupted by a de novo translocation in a patient with syndromic intellectual disability and autism spectrum disorder. European Journal of Medical Genetics, 2018, 61, 376-383.	1.3	13
58	Homozygous missense mutation in STYXL1 associated with moderate intellectual disability, epilepsy and behavioural complexities. European Journal of Medical Genetics, 2015, 58, 205-210.	1.3	11
59	Optic Nerve Hypoplasia Is a Pervasive Subcortical Pathology of Visual System in Neonates. , 2017, 58, 5485.		11
60	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	3.2	11
61	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	2.0	11
62	Challenges in molecular diagnosis of X-linked Intellectual disability. British Medical Bulletin, 2020, 133, 36-48.	6.9	10
63	MAPRE2 mutations result in altered human cranial neural crest migration, underlying craniofacial malformations in CSC-KT syndrome. Scientific Reports, 2021, 11, 4976.	3.3	10
64	ERBB4 exonic deletions on chromosome 2q34 in patients with intellectual disability or epilepsy. European Journal of Human Genetics, 2021, 29, 1377-1383.	2.8	9
65	A homozygous deletion of exon 1 in WISP3 causes progressive pseudorheumatoid dysplasia in two siblings. Human Genome Variation, 2015, 2, 15049.	0.7	8
66	X-linked mental retardation, short stature, microcephaly and hypogonadism maps to Xp22.1-p21.3 in a Belgian family. European Journal of Medical Genetics, 2005, 48, 145-152.	1.3	7
67	The Role of AKT3 Copy Number Changes in Brain Abnormalities and Neurodevelopmental Disorders: Four New Cases and Literature Review. Frontiers in Genetics, 2019, 10, 58.	2.3	7
68	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	1.2	6
69	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
70	Interstitial microdeletion of 17q11.2 is associated with hypotonia, fatigue, intellectual disability, and a subtle facial phenotype in three unrelated patients. American Journal of Medical Genetics, Part A, 2018, 176, 209-213.	1.2	2
71	Comprehensive analysis of neuronal guidance cue expression regulation during monocyte-to-macrophage differentiation reveals post-transcriptional regulation of semaphorin7A by the RNA-binding protein quaking. Innate Immunity, 2021, 27, 118-132.	2.4	2
72	RNF170 mutation causes autosomal dominant sensory ataxia with variable pyramidal involvement. European Journal of Neurology, 2022, 29, 345-349.	3.3	2

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73	A boy with an unusual association of ventral midline anomalies including a trunk-like umbilicus. Clinical Dysmorphology, 2004, 13, 261-263.	0.3	2
74	Loss-of-function variants in exon 4 of TAB2 causeÂaÂrecognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletalÂinvolvement. Genetics in Medicine, 2021, , .	2.4	1
75	Human Brain Models of Intellectual Disability: Experimental Advances and Novelties. International Journal of Molecular Sciences, 2022, 23, 6476.	4.1	0