Hanka Venselaar

List of Publications by Year
in descending order

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116 papers	7,210 citations	71097 41 h-index	64791 79 g-index
121 all docs	121 docs citations	121 times ranked	13470 citing authors

#	Article	IF	Citations
1	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
2	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the AGR2 gene. Journal of Medical Genetics, 2022, 59, 993-1001.	3.2	5
3	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	3.8	5
4	Bifunctional protein PCBD2 operates as a coâ€factor for hepatocyte nuclear factor 1Î² and modulates gene transcription. FASEB Journal, 2021, 35, e21366.	0.5	1
5	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 1246-1254.	2.4	5
6	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	4.1	8
7	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
8	Antisense oligonucleotide-based treatment of retinitis pigmentosa caused by USH2A exon 13 mutations. Molecular Therapy, 2021, 29, 2441-2455.	8.2	75
9	Point Mutation Approach to Reduce Antigenicity of Interferon Beta. International Journal of Peptide Research and Therapeutics, 2020, 26, 1353-1361.	1.9	2
10	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	2.4	15
11	Novel GANAB variants associated with polycystic liver disease. Orphanet Journal of Rare Diseases, 2020, 15, 302.	2.7	11
12	Novel defect in phosphatidylinositol 4â€kinase type 2â€alpha (<scp>PI4K2A</scp>) at the membraneâ€enzyme interface is associated with metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2020, 43, 1382-1391.	3.6	7
13	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
14	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
15	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	6.2	8
16	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
17	Mutations in GDF11 and the extracellular antagonist, Follistatin, as a likely cause of Mendelian forms of orofacial clefting in humans. Human Mutation, 2019, 40, 1813-1825.	2.5	26
18	Investigating the active site of human trimethyllysine hydroxylase. Biochemical Journal, 2019, 476, 1109-1119.	3.7	4

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19	Front Cover, Volume 40, Issue 10. Human Mutation, 2019, 40, i.	2.5	0
20	Deficiency of the human cysteine protease inhibitor cystatin M/E causes hypotrichosis and dry skin. Genetics in Medicine, 2019, 21, 1559-1567.	2.4	7
21	Mutations in CYB561 Causing a Novel Orthostatic Hypotension Syndrome. Circulation Research, 2018, 122, 846-854.	4.5	22
22	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	21.4	86
23	Toward clinical and molecular understanding of pathogenic variants in the ZBTB18 gene. Molecular Genetics & Genomic Medicine, 2018, 6, 393-400.	1.2	22
24	A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. Genetics in Medicine, 2018, 20, 1061-1068.	2.4	37
25	Functional characterization of TBR1 variants in neurodevelopmental disorder. Scientific Reports, 2018, 8, 14279.	3.3	26
26	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
27	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
28	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. American Journal of Human Genetics, 2018, 102, 1143-1157.	6.2	94
29	Retinal-Based Proton Pumping in the Near Infrared. Journal of the American Chemical Society, 2017, 139, 2338-2344.	13.7	45
30	Mutation in mitochondrial complex IV subunit COX5A causes pulmonary arterial hypertension, lactic acidemia, and failure to thrive. Human Mutation, 2017, 38, 692-703.	2.5	32
31	NewProt â€“ a protein engineering portal. Protein Engineering, Design and Selection, 2017, 30, 441-447.	2.1	11
32	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndromeâ€like phenotype and hypogammaglobulinemia. American Journal of Medical Genetics, Part A, 2017, 173, 1813-1820.	1.2	8
33	A Gate Hinge Controls the Epithelial Calcium Channel TRPV5. Scientific Reports, 2017, 7, 45489.	3.3	23
34	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
35	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	6.2	36
36	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. American Journal of Human Genetics, 2017, 101, 478-484.	6.2	84

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37	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
38	Aggregation of populationâ€based genetic variation over protein domain homologues and its potential use in genetic diagnostics. Human Mutation, 2017, 38, 1454-1463.	2.5	36
39	NovelIRF6Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing. Journal of Dental Research, 2017, 96, 179-185.	5.2	12
40	Germline activating TYK2 mutations in pediatric patients with two primary acute lymphoblastic leukemia occurrences. Leukemia, 2017, 31, 821-828.	7.2	35
41	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
42	Highly conserved nucleotide phosphatase essential for membrane lipid homeostasis in Streptococcus pneumoniae. Molecular Microbiology, 2016, 101, 12-26.	2.5	24
43	Identification of a novel inactivating mutation in Isocitrate Dehydrogenase 1 (IDH1-R314C) in a high grade astrocytoma. Scientific Reports, 2016, 6, 30486.	3.3	11
44	Whole exome sequencing identifies a heterozygous missense variant in the PRDM5 gene in a family with Axenfeldâ€“Rieger syndrome. Neurogenetics, 2016, 17, 17-23.	1.4	28
45	Therapeutic NOTCH3 cysteine correction in CADASIL using exon skipping:in vitroproof of concept. Brain, 2016, 139, 1123-1135.	7.6	43
46	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	2.8	28
47	Identification of Novel Candidate Genes for Early-Onset Colorectal Cancer Susceptibility. PLoS Genetics, 2016, 12, e1005880.	3.5	52
48	SMAD2Mutations Are Associated with Arterial Aneurysms and Dissections. Human Mutation, 2015, 36, 1145-1149.	2.5	74
49	SDHA mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. European Journal of Human Genetics, 2015, 23, 202-209.	2.8	71
50	Submembranous recruitment of creatine kinase B supports formation of dynamic actin-based protrusions of macrophages and relies on its C-terminal flexible loop. European Journal of Cell Biology, 2015, 94, 114-127.	3.6	13
51	A missense variant of the ATP1A2 gene is associated with a novel phenotype of progressive sensorineural hearing loss associated with migraine. European Journal of Human Genetics, 2015, 23, 639-645.	2.8	18
52	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	6.2	27
53	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
54	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61

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55	Modulation of spectral properties and pump activity of proteorhodopsins by retinal analogues. Biochemical Journal, 2015, 467, 333-343.	3.7	26
56	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	2.8	238
57	Nonsyndromic Hearing Loss Caused by USH1G Mutations. Ear and Hearing, 2015, 36, 205-211.	2.1	20
58	Identification of a novel MET mutation in high-grade glioma resulting in an auto-active intracellular protein. Acta Neuropathologica, 2015, 130, 131-144.	7.7	43
59	A role for repressive complexes and H3K9 di-methylation in PRDM5-associated brittle cornea syndrome. Human Molecular Genetics, 2015, 24, 6565-6579.	2.9	17
60	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	6.2	55
61	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
62	De novo <scp>WNT5A</scp>â€associated autosomal dominant Robinow syndrome suggests specificity of genotype and phenotype. Clinical Genetics, 2015, 87, 34-41.	2.0	56
63	Whole-exome sequencing reveals LRP5 mutations and canonical Wnt signaling associated with hepatic cystogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 5343-5348.	7.1	79
64	Gene polymorphisms in pattern recognition receptors and susceptibility to idiopathic recurrent vulvovaginal candidiasis. Frontiers in Microbiology, 2014, 5, 483.	3.5	66
65	The effect of novel mutations on the structure and enzymatic activity of unconventional myosins associated with autosomal dominant non-syndromic hearing loss. Open Biology, 2014, 4, 140107.	3.6	19
66	A dsRNA-binding protein of a complex invertebrate DNA virus suppresses the Drosophila RNAi response. Nucleic Acids Research, 2014, 42, 12237-12248.	14.5	44
67	Mutations in PCBD1 Cause Hypomagnesemia and Renal Magnesium Wasting. Journal of the American Society of Nephrology: JASN, 2014, 25, 574-586.	6.1	68
68	A germ line mutation in cathepsin B points toward a role in asparaginase pharmacokinetics. Blood, 2014, 124, 3027-3029.	1.4	12
69	Germline Mutations in the Spindle Assembly Checkpoint Genes BUB1 and BUB3 Are Risk Factors for Colorectal Cancer. Gastroenterology, 2013, 145, 544-547.	1.3	86
70	Status quo of annotation of human disease variants. BMC Bioinformatics, 2013, 14, 352.	2.6	3
71	Familial hemiplegic migraine mutations affect Na,K-ATPase domain interactions. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 2173-2179.	3.8	14
72	Maternal Uniparental Isodisomy of Chromosome 6 Reveals a TULP1 Mutation as a Novel Cause of Cone Dysfunction. Ophthalmology, 2013, 120, 1239-1246.	5.2	36

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73	Structural model of a putrescine-cadaverine permease from Trypanosoma cruzi predicts residues vital for transport and ligand binding. Biochemical Journal, 2013, 452, 423-432.	3.7	10
74	Homozygosity mapping identifies genetic defects in four consanguineous families with retinal dystrophy from Pakistan. Clinical Genetics, 2013, 84, 290-293.	2.0	7
75	Genotype and clinical care correlations in craniosynostosis: Findings from a cohort of 630 Australian and New Zealand patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 259-270.	1.6	44
76	A Patient with Complex I Deficiency Caused by a Novel ACAD9 Mutation Not Responding to Riboflavin Treatment. JIMD Reports, 2013, 12, 37-45.	1.5	29
77	Bacterial CS2 Hydrolases from Acidithiobacillus thiooxidans Strains Are Homologous to the Archaeal Catenane CS2 Hydrolase. Journal of Bacteriology, 2013, 195, 4046-4056.	2.2	27
78	SNP Linkage Analysis and Whole Exome Sequencing Identify a Novel POU4F3 Mutation in Autosomal Dominant Late-Onset Nonsyndromic Hearing Loss (DFNA15). PLoS ONE, 2013, 8, e79063.	2.5	28
79	NPHP4 Variants Are Associated With Pleiotropic Heart Malformations. Circulation Research, 2012, 110, 1564-1574.	4.5	46
80	Membrane Topology and Intracellular Processing of Cyclin M2 (CNNM2). Journal of Biological Chemistry, 2012, 287, 13644-13655.	3.4	86
81	A novel COCH mutation associated with autosomal dominant nonsyndromic hearing loss disrupts the structural stability of the vWFA2 domain. Journal of Molecular Medicine, 2012, 90, 1321-1331.	3.9	20
82	A catalytic defect in mitochondrial respiratory chain complex I due to a mutation in NDUFS2 in a patient with Leigh syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 168-175.	3.8	26
83	Dominant missense mutations in ABCC9 cause CantÃº syndrome. Nature Genetics, 2012, 44, 793-796.	21.4	184
84	Phosphorylation target site specificity for AGC kinases DMPK E and lats2. Journal of Cellular Biochemistry, 2012, 113, 2126-2135.	2.6	1
85	A Germline Mutation in Cathepsin B in a Child with ALL Points towards a Key Role for This Enzyme in L-Asparaginase Pharmacokinetics.. Blood, 2012, 120, 2458-2458.	1.4	0
86	Identification of recurrent and novel mutations in TULP1 in Pakistani families with early-onset retinitis pigmentosa. Molecular Vision, 2012, 18, 1226-37.	1.1	17
87	The structureâ€“function relationship of the Aspergillus fumigatus cyp51A L98H conversion by site-directed mutagenesis: The mechanism of L98H azole resistance. Fungal Genetics and Biology, 2011, 48, 1062-1070.	2.1	92
88	Mass Spectrometry Analysis of Hepcidin Peptides in Experimental Mouse Models. PLoS ONE, 2011, 6, e16762.	2.5	25
89	Mutations in SMAD3 cause a syndromic form of aortic aneurysms and dissections with early-onset osteoarthritis. Nature Genetics, 2011, 43, 121-126.	21.4	583
90	Mutation in subdomain G' of mitochondrial elongation factor G1 is associated with combined OXPHOS deficiency in fibroblasts but not in muscle. European Journal of Human Genetics, 2011, 19, 275-279.	2.8	42

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91	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	6.2	88
92	Microcephaly with Simplified Gyration, Epilepsy, and Infantile Diabetes Linked to Inappropriate Apoptosis of Neural Progenitors. American Journal of Human Genetics, 2011, 89, 265-276.	6.2	77
93	Genotypeâ€“Phenotype Correlation in DFNB8/10 Families with TMPRSS3 Mutations. JARO - Journal of the Association for Research in Otolaryngology, 2011, 12, 753-766.	1.8	69
94	Variation in Genes of Î²-glucan Recognition Pathway and Susceptibility to Opportunistic Infections in HIV-Positive Patients. Immunological Investigations, 2011, 40, 735-750.	2.0	7
95	The future of HOPE: what can and cannot be predicted about the molecular effects of a disease causing point mutation in a protein?. EMBnet Journal, 2011, 17, 25.	0.6	3
96	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. American Journal of Human Genetics, 2010, 87, 146-153.	6.2	50
97	The alpha-kinase family: an exceptional branch on the protein kinase tree. Cellular and Molecular Life Sciences, 2010, 67, 875-890.	5.4	104
98	Homology modelling and spectroscopy, a never-ending love story. European Biophysics Journal, 2010, 39, 551-563.	2.2	51
99	Protein structure analysis of mutations causing inheritable diseases. An e-Science approach with life scientist friendly interfaces. BMC Bioinformatics, 2010, 11, 548.	2.6	819
100	The moonlighting function of pyruvate carboxylase resides in the non-catalytic end of the TIM barrel. Biochimica Et Biophysica Acta - Molecular Cell Research, 2010, 1803, 1038-1042.	4.1	14
101	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. Human Mutation, 2010, 31, 656-666.	2.5	126
102	Secondary and tertiary structure modeling reveals effects of novel mutations in polycystic liver disease genes PRKCSH and SEC63. Clinical Genetics, 2010, 78, 47-56.	2.0	42
103	Functional Analysis of the Kv1.1 N255D Mutation Associated with Autosomal Dominant Hypomagnesemia. Journal of Biological Chemistry, 2010, 285, 171-178.	3.4	50
104	Acyl-CoA Dehydrogenase 9 Is Required for the Biogenesis of Oxidative Phosphorylation Complex I. Cell Metabolism, 2010, 12, 283-294.	16.2	172
105	A Novel Homozygous Nonsense Mutation inCABP4Causes Congenital Coneâ€“Rod Synaptic Disorder. , 2009, 50, 2344.		76
106	Mutations in NDUFAF3 (C3ORF60), Encoding an NDUFAF4 (C6ORF66)-Interacting Complex I Assembly Protein, Cause Fatal Neonatal Mitochondrial Disease. American Journal of Human Genetics, 2009, 84, 718-727.	6.2	155
107	Human Dectin-1 Deficiency and Mucocutaneous Fungal Infections. New England Journal of Medicine, 2009, 361, 1760-1767.	27.0	671
108	Role of the C-terminal linear region of EGF-like growth factors in ErbB specificity. Growth Factors, 2009, 27, 163-172.	1.7	4

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109	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606.	3.2	194
110	Clinical and molecular characterizations of novelPOU3F4mutations reveal that DFN3 is due to null function of POU3F4 protein. Physiological Genomics, 2009, 39, 195-201.	2.3	37
111	Gene structure and mutant alleles of PCDH15: nonsyndromic deafness DFNB23 and type 1 Usher syndrome. Human Genetics, 2008, 124, 215-223.	3.8	81
112	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. Nature Genetics, 2008, 40, 1335-1340.	21.4	65
113	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	6.2	127
114	A novel (Leu183Pro-)mutation in the HFE-gene co-inherited with the Cys282Tyr mutation in two unrelated Dutch hemochromatosis patients. Blood Cells, Molecules, and Diseases, 2008, 40, 334-338.	1.4	10
115	Role of the Î±-Kinase Domain in Transient Receptor Potential Melastatin 6 Channel and Regulation by Intracellular ATP. Journal of Biological Chemistry, 2008, 283, 19999-20007.	3.4	48
116	Negative Constraints Underlie the ErbB Specificity of Epidermal Growth Factor-like Ligands. Journal of Biological Chemistry, 2006, 281, 40033-40040.	3.4	5