Isabelle Schrauwen

List of Publications by Year
in descending order

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105 papers	2,494 citations	236925 25 h-index	265206 42 g-index
111 all docs	111 docs citations	111 times ranked	4046 citing authors

#	Article	IF	Citations
1	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.1	157
2	Variants in triggering receptor expressed on myeloid cells 2 are associated with both behavioral variant frontotemporal lobar degeneration and Alzheimer's disease. Neurobiology of Aging, 2013, 34, 2077.e11-2077.e18.	3.1	124
3	A Mutation in CABP2 , Expressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment. American Journal of Human Genetics, 2012, 91, 636-645.	6.2	96
4	Genome-wide association analysis demonstrates the highly polygenic character of age-related hearing impairment. European Journal of Human Genetics, 2015, 23, 110-115.	2.8	84
5	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. Human Mutation, 2016, 37, 812-819.	2.5	76
6	The coding polymorphism T263I in TGF-Î²1 is associated with otosclerosis in two independent populations. Human Molecular Genetics, 2007, 16, 2021-2030.	2.9	75
7	The etiology of otosclerosis: A combination of genes and environment. Laryngoscope, 2010, 120, 1195-1202.	2.0	75
8	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. Human Mutation, 2011, 32, 825-834.	2.5	73
9	A Genome-wide Analysis Identifies Genetic Variants in the RELN Gene Associated with Otosclerosis. American Journal of Human Genetics, 2009, 84, 328-338.	6.2	66
10	A sensitive and specific diagnostic test for hearing loss using a microdroplet PCRâ€based approach and next generation sequencing. American Journal of Medical Genetics, Part A, 2013, 161, 145-152.	1.2	61
11	Association of Bone Morphogenetic Proteins With Otosclerosis. Journal of Bone and Mineral Research, 2008, 23, 507-516.	2.8	58
12	A new locus for otosclerosis, OTSC8, maps to the pericentromeric region of chromosome 9. Human Genetics, 2008, 123, 267-272.	3.8	54
13	A seventh locus for otosclerosis, OTSC7, maps to chromosome 6q13â€“16.1. European Journal of Human Genetics, 2007, 15, 362-368.	2.8	53
14	A comprehensive catalogue of the coding and non-coding transcripts of the human inner ear. Hearing Research, 2016, 333, 266-274.	2.0	51
15	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	2.5	48
16	A Frame-Shift Mutation in CAV1 Is Associated with a Severe Neonatal Progeroid and Lipodystrophy Syndrome. PLoS ONE, 2015, 10, e0131797.	2.5	46
17	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Langeâ€Nielsen Syndrome and Romanoâ€Ward Syndrome. Human Mutation, 2019, 40, 162-176.	2.5	44
18	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44

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19	Ca ²⁺ -binding protein 2 inhibits Ca ²⁺ -channel inactivation in mouse inner hair cells. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E1717-E1726.	7.1	42
20	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	3.8	42
21	Autosomal Recessive Stickler Syndrome in Two Families Is Caused by Mutations in theCOL9A1Gene. , 2011, 52, 4774.		40
22	Broadening the phenotype of <scp>LRP2</scp> mutations: a new mutation in <scp>LRP2</scp> causes a predominantly ocular phenotype suggestive of Stickler syndrome. Clinical Genetics, 2014, 86, 282-286.	2.0	37
23	A Syndromic Intellectual Disability Disorder Caused by Variants in TELO2 , a Gene Encoding a Component of the TTT Complex. American Journal of Human Genetics, 2016, 98, 909-918.	6.2	35
24	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. Journal of Human Genetics, 2019, 64, 153-160.	2.3	32
25	Detection of Rare Nonsynonymous Variants in TGFB1 in Otosclerosis Patients. Annals of Human Genetics, 2009, 73, 171-175.	0.8	31
26	Family history of Alzheimerâ€™s disease alters cognition and is modified by medical and genetic factors. ELife, 2019, 8, .	6.0	30
27	Genetic variants in the RELN gene are associated with otosclerosis in multiple European populations. Human Genetics, 2010, 127, 155-162.	3.8	28
28	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. Journal of Bone and Mineral Research, 2019, 34, 375-386.	2.8	27
29	Identification of Novel Genetic Risk Loci in Maltese Dogs with Necrotizing Meningoencephalitis and Evidence of a Shared Genetic Risk across Toy Dog Breeds. PLoS ONE, 2014, 9, e112755.	2.5	27
30	No Evidence for Association Between the Renin-Angiotensin-Aldosterone System and Otosclerosis in a Large Belgian-Dutch Population. Otology and Neurotology, 2009, 30, 1079-1083.	1.3	26
31	Case Report: Compound heterozygous nonsense mutations in TRMT10A are associated with microcephaly, delayed development, and periventricular white matter hyperintensities. F1000Research, 2015, 4, 912.	1.6	26
32	Two unrelated children with overlapping 6q25.3 deletions, motor speech disorders, and language delays. American Journal of Medical Genetics, Part A, 2017, 173, 2659-2669.	1.2	25
33	Association of COL1A1 and TGFB1 Polymorphisms with Otosclerosis in a Tunisian Population. Annals of Human Genetics, 2011, 75, 598-604.	0.8	24
34	Characterization of X Chromosome Inactivation Using Integrated Analysis of Whole-Exome and mRNA Sequencing. PLoS ONE, 2014, 9, e113036.	2.5	24
35	De novo variants in GREB1L are associated with non-syndromic inner ear malformations and deafness. Human Genetics, 2018, 137, 459-470.	3.8	24
36	Case Report: Novel mutations in TBC1D24 are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. F1000Research, 2017, 6, 553.	1.6	24

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37	Autosomal Dominantly Inherited GREB1L Variants in Individuals with Profound Sensorineural Hearing Impairment. Genes, 2020, 11, 687.	2.4	23
38	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029.	3.8	23
39	A De Novo Mutation inTEAD1Causes Nonâ€“X-Linked Aicardi Syndrome. , 2015, 56, 3896.		22
40	Longâ€read wholeâ€genome sequencing for the genetic diagnosis of dystrophinopathies. Annals of Clinical and Translational Neurology, 2020, 7, 2041-2046.	3.7	22
41	A new locus for otosclerosis, OTSC10, maps to chromosome 1q41-44. Clinical Genetics, 2011, 79, 495-497.	2.0	20
42	COL1A1 association and otosclerosis: A metaâ€analysis. American Journal of Medical Genetics, Part A, 2012, 158A, 1066-1070.	1.2	20
43	Practical approach to the genetic diagnosis of unsolved dystrophinopathies: a stepwise strategy in the genomic era. Journal of Medical Genetics, 2021, 58, 743-751.	3.2	20
44	Transcriptional profiling of multiple system atrophy cerebellar tissue highlights differences between the parkinsonian and cerebellar sub-types of the disease. Acta Neuropathologica Communications, 2020, 8, 76.	5.2	20
45	Whole transcriptome profiling of the human hippocampus suggests an involvement of the KIBRA rs17070145 polymorphism in differential activation of the MAPK signaling pathway. Hippocampus, 2017, 27, 784-793.	1.9	19
46	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. European Journal of Human Genetics, 2019, 27, 1456-1465.	2.8	19
47	Genetic variants in RELN are associated with otosclerosis in a nonâ€European population from Tunisia. Annals of Human Genetics, 2010, 74, 399-405.	0.8	18
48	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	3.8	18
49	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC Medical Genetics, 2018, 19, 122.	2.1	18
50	Exome Sequencing of Two Siblings with Sporadic Autism Spectrum Disorder and Severe Speech Sound Disorder Suggests Pleiotropic and Complex Effects. Behavior Genetics, 2019, 49, 399-414.	2.1	18
51	Genetic Association Analysis in a Clinically and Histologically Confirmed Otosclerosis Population Confirms Association With the TGFB1 Gene but Suggests an Association of the RELN Gene With a Clinically Indistinguishable Otosclerosis-Like Phenotype. Otology and Neurotology, 2014, 35, 1058-1064.	1.3	17
52	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. Genetics in Medicine, 2019, 21, 1199-1208.	2.4	17
53	A de novo missense mutation in ZMYND11 is associated with global developmental delay, seizures, and hypotonia. Journal of Physical Education and Sports Management, 2016, 2, a000851.	1.2	16
54	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16

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55	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	3.8	16
56	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
57	Involvement of T-cell receptor-Î² alterations in the development of otosclerosis linked to OTSC2. Genes and Immunity, 2010, 11, 246-253.	4.1	14
58	Compound heterozygous mutations in SNAP29 is associated with Pelizaeus-Merzbacher-like disorder (PMLD). Human Genetics, 2019, 138, 1409-1417.	3.8	14
59	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
60	A de novo splice site mutation in CASK causes FG syndromeâ€4 and congenital nystagmus. American Journal of Medical Genetics, Part A, 2017, 173, 611-617.	1.2	11
61	Insufficient evidence for a role of SERPINF1 in otosclerosis. Molecular Genetics and Genomics, 2019, 294, 1001-1006.	2.1	11
62	Rare Variants in BMP2 and BMP4 Found in Otosclerosis Patients Reduce Smad Signaling. Otology and Neurotology, 2014, 35, 395-400.	1.3	10
63	Neonatal epileptic encephalopathy caused by de novo GNAO1 mutation misdiagnosed as atypical Rett syndrome: Cautions in interpretation of genomic test results. Seminars in Pediatric Neurology, 2018, 26, 28-32.	2.0	10
64	Hearing impairment locus heterogeneity and identification of PLS1 as a new autosomal dominant gene in Hungarian Roma. European Journal of Human Genetics, 2019, 27, 869-878.	2.8	10
65	A 1â€‰bp deletion in the dual reading frame deafness gene LRTOMT causes a frameshift from the first into the second reading frame. American Journal of Medical Genetics, Part A, 2011, 155, 2021-2023.	1.2	9
66	A novel FBXO28 frameshift mutation in a child with developmental delay, dysmorphic features, and intractable epilepsy: A second gene that may contribute to the 1q41â€q42 deletion phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1549-1558.	1.2	9
67	Splicing Characteristics of Dystrophin Pseudoexons and Identification of a Novel Pathogenic Intronic Variant in the DMD Gene. Genes, 2020, 11, 1180.	2.4	9
68	Bi-Allelic Novel Variants in CLIC5 Identified in a Cameroonian Multiplex Family with Non-Syndromic Hearing Impairment. Genes, 2020, 11, 1249.	2.4	9
69	Exploring genome-wide DNA methylation patterns in Aicardi syndrome. Epigenomics, 2017, 9, 1373-1386.	2.1	8
70	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	1.9	8
71	A novel variant in DMXL2 gene is associated with autosomal dominant non-syndromic hearing impairment (DFNA71) in a Cameroonian family. Experimental Biology and Medicine, 2021, 246, 1524-1532.	2.4	8
72	Further confirmation of the association of SLC12A2 with non-syndromic autosomal-dominant hearing impairment. Journal of Human Genetics, 2021, 66, 1169-1175.	2.3	8

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73	Clinical and genetic analysis of two Tunisian otosclerosis families. American Journal of Medical Genetics, Part A, 2007, 143A, 1653-1660.	1.2	7
74	Phenotype of the first otosclerosis family linked to OTSC10. Laryngoscope, 2011, 121, 838-845.	2.0	7
75	Utilizing RNA and outlier analysis to identify an intronic spliceâ€altering variant in AP4S1 in a sibling pair with progressive spastic paraplegia. Human Mutation, 2020, 41, 412-419.	2.5	7
76	Genomic analysis of childhood hearing loss in the Yoruba population of Nigeria. European Journal of Human Genetics, 2022, 30, 42-52.	2.8	7
77	Compound heterozygous mutations in MASP1 in a deaf child with absent cochlear nerves. Neurology: Genetics, 2017, 3, e153.	1.9	6
78	A de novo SIX1 variant in a patient with a rare nonsyndromic cochleovestibular nerve abnormality, cochlear hypoplasia, and bilateral sensorineural hearing loss. Molecular Genetics & Genomic Medicine, 2019, 7, e995.	1.2	6
79	Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2020, 65, 187-192.	2.3	6
80	Genes Implicated in Rare Congenital Inner Ear and Cochleovestibular Nerve Malformations. Ear and Hearing, 2020, 41, 983-989.	2.1	6
81	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. European Journal of Human Genetics, 2021, , .	2.8	6
82	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. Human Genetics, 2022, 141, 951-963.	3.8	6
83	Genome wide analysis in a family with sensorineural hearing loss, autism and mental retardation. Gene, 2012, 510, 102-106.	2.2	5
84	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	3.2	5
85	Maternal mosaicism underlies the inheritance of a rare germline AKT3 variant which is responsible for megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome in two Roma half-siblings. Experimental and Molecular Pathology, 2020, 115, 104471.	2.1	5
86	A Monoallelic Variant in REST Is Associated with Non-Syndromic Autosomal Dominant Hearing Impairment in a South African Family. Genes, 2021, 12, 1765.	2.4	5
87	Heterozygosity mapping for human dominant trait variants. Human Mutation, 2019, 40, 996-1004.	2.5	4
88	Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. Journal of Human Genetics, 2021, 66, 1009-1018.	2.3	4
89	ESHRD: deconvolution of brain homogenate RNA expression data to identify cell-type-specific alterations in Alzheimerâ€™s disease. Aging, 2020, 12, 4124-4162.	3.1	4
90	A novel 4.25â€‰kb heterozygous deletion in PAX6 in a Chinese Han family with congenital aniridia combined with cataract and nystagmus. BMC Ophthalmology, 2021, 21, 353.	1.4	4

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91	Methods to Improve Molecular Diagnosis in Genomic Cold Cases in Pediatric Neurology. Genes, 2022, 13, 333.	2.4	4
92	Genome-wide analysis reveals a novel autosomal-recessive hearing loss locus DFNB80 on chromosome 2p16.1-p21. Journal of Human Genetics, 2013, 58, 98-101.	2.3	3
93	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	2.3	3
94	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. Molecular Genetics & Genomic Medicine, 2021, , e1703.	1.2	3
95	Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family. Genes, 2022, 13, 662.	2.4	3
96	Identification of autosomal recessive nonsyndromic hearing impairment genes through the study of consanguineous and non-consanguineous families: past, present, and future. Human Genetics, 2022, 141, 413-430.	3.8	2
97	SCN1A Variants as the Underlying Cause of Genetic Epilepsy with Febrile Seizures Plus in Two Multi-Generational Colombian Families. Genes, 2022, 13, 754.	2.4	2
98	Advances in Molecular Genetics and the Molecular Biology of Deafness. BioMed Research International, 2016, 2016, 1-2.	1.9	1
99	Autosomal recessive nonsyndromic hearing impairment in two Finnish families due to the population enriched CABP2 c.637+1G>T variant. Molecular Genetics & Genomic Medicine, 2022, , e1866.	1.2	1
100	A monoallelic variant in EYA1 is associated with <scp>Branchioâ€Otic</scp> syndrome in a Malian family. Molecular Genetics & Genomic Medicine, 0, , .	1.2	1
101	Commentary on â€œOtosclerosis: Thirty-Year Follow-Up After Surgeryâ€. Annals of Otology, Rhinology and Laryngology, 2011, 120, 615-616.	1.1	0
102	Cellular and Animal Models of Neurologic Disease. , 2017, , 114-122.		0
103	A Start Codon Variant in NOG Underlies Symphalangism and Ossicular Chain Malformations Affecting Both the Incus and the Stapes. Case Reports in Genetics, 2019, 2019, 1-5.	0.2	0
104	Exome Sequencing Identifies a Novel FBN1 Variant in a Pakistani Family with Marfan Syndrome That Includes Left Ventricle Diastolic Dysfunction. Genes, 2021, 12, 1915.	2.4	0
105	Novel variants in the RDH5 Gene in a Chinese Han family with fundus albipunctatus. BMC Ophthalmology, 2022, 22, 69.	1.4	0