Isabelle Schrauwen

List of Publications by Year in Descending Order

103 papers	1,693 citations	23 h-index	37 g-index
111 ext. papers	2,146 ext. citations	4.5 avg, IF	4.08 L-index

#	Paper	IF	Citations
103	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
102	Novel variants in the RDH5 Gene in a Chinese Han family with fundus albipunctatus.. *BMC Ophthalmology*, **2022**, 22, 69	2.3	
101	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	2.3	1
100	Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family.. *Genes*, **2022**, 13,	4.2	1
99	SCN1A Variants as the Underlying Cause of Genetic Epilepsy with Febrile Seizures Plus in Two Multi-Generational Colombian Families. *Genes*, **2022**, 13, 754	4.2	
98	A Monoallelic Variant in Is Associated with Non-Syndromic Autosomal Dominant Hearing Impairment in a South African Family. *Genes*, **2021**, 12,	4.2	1
97	Genomic analysis of childhood hearing loss in the Yoruba population of Nigeria. *European Journal of Human Genetics*, **2021**,	5.3	2
96	A novel 4.25 kb heterozygous deletion in PAX6 in a Chinese Han family with congenital aniridia combined with cataract and nystagmus. *BMC Ophthalmology*, **2021**, 21, 353	2.3	2
95	Practical approach to the genetic diagnosis of unsolved dystrophinopathies: a stepwise strategy in the genomic era. *Journal of Medical Genetics*, **2021**, 58, 743-751	5.8	5
94	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	6.3	6
93	A novel variant in gene is associated with autosomal dominant non-syndromic hearing impairment (DFNA71) in a Cameroonian family. *Experimental Biology and Medicine*, **2021**, 246, 1524-1532	3.7	3
92	Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. *Journal of Human Genetics*, **2021**, 66, 1009-1018	4.3	2
91	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. *Molecular Genetics & Genomic Medicine*, **2021**, e1703	2.3	0
90	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. *European Journal of Human Genetics*, **2021**,	5.3	1
89	Delineating the genotypic and phenotypic spectrum of -related neurodevelopmental disorders. *Journal of Medical Genetics*, **2021**,	5.8	1
88	Identification of autosomal recessive nonsyndromic hearing impairment genes through the study of consanguineous and non-consanguineous families: past, present, and future. *Human Genetics*, **2021**, 1	6.3	1
87	Further confirmation of the association of SLC12A2 with non-syndromic autosomal-dominant hearing impairment. *Journal of Human Genetics*, **2021**, 66, 1169-1175	4.3	2

86	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. *Genetics in Medicine*, **2021**, 23, 2122-2137	8.1	3
85	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. *Human Genetics*, **2021**, 1	6.3	2
84	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	11	1
83	Bi-Allelic Novel Variants in Identified in a Cameroonian Multiplex Family with Non-Syndromic Hearing Impairment. *Genes*, **2020**, 11,	4.2	3
82	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	7.3	10
81	Genes Implicated in Rare Congenital Inner Ear and Cochleovestibular Nerve Malformations. *Ear and Hearing*, **2020**, 41, 983-989	3.4	3
80	Autosomal Dominantly Inherited GREB1L Variants in Individuals with Profound Sensorineural Hearing Impairment. *Genes*, **2020**, 11,	4.2	8
79	ESHRD: deconvolution of brain homogenate RNA expression data to identify cell-type-specific alterations in Alzheimer's disease. *Aging*, **2020**, 12, 4124-4162	5.6	2
78	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	4.4	2
77	Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. *Journal of Human Genetics*, **2020**, 65, 187-192	4.3	1
76	Utilizing RNA and outlier analysis to identify an intronic splice-altering variant in AP4S1 in a sibling pair with progressive spastic paraplegia. *Human Mutation*, **2020**, 41, 412-419	4.7	1
75	Splicing Characteristics of Dystrophin Pseudoexons and Identification of a Novel Pathogenic Intronic Variant in the Gene. *Genes*, **2020**, 11,	4.2	4
74	Congenital myasthenic syndrome caused by a frameshift insertion mutation in. *Neurology: Genetics*, **2020**, 6, e468	3.8	3
73	Long-read whole-genome sequencing for the genetic diagnosis of dystrophinopathies. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2041-2046	5.3	6
72	Heterozygosity mapping for human dominant trait variants. *Human Mutation*, **2019**, 40, 996-1004	4.7	3
71	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. *European Journal of Human Genetics*, **2019**, 27, 1456-1465	5.3	16
70	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	5.3	6
69	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	3.2	7

68	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. *Human Genetics*, **2019**, 138, 593-600	6.3	11
67	Insufficient evidence for a role of SERPINF1 in otosclerosis. *Molecular Genetics and Genomics*, **2019**, 294, 1001-1006	3.1	6
66	A Start Codon Variant in Underlies Symphalangism and Ossicular Chain Malformations Affecting Both the Incus and the Stapes. *Case Reports in Genetics*, **2019**, 2019, 2836263	0.7	
65	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	2.3	5
64	Family history of Alzheimer's disease alters cognition and is modified by medical and genetic factors. *ELife*, **2019**, 8,	8.9	12
63	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	6.3	13
62	Compound heterozygous mutations in SNAP29 is associated with Pelizaeus-Merzbacher-like disorder (PMLD). *Human Genetics*, **2019**, 138, 1409-1417	6.3	7
61	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	4.3	17
60	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. *Human Mutation*, **2019**, 40, 53-72	4.7	26
59	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	8.1	10
58	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Lange-Nielsen Syndrome and Romano-Ward Syndrome. *Human Mutation*, **2019**, 40, 162-176	4.7	18
57	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	2.9	7
56	A novel FBXO28 frameshift mutation in a child with developmental delay, dysmorphic features, and intractable epilepsy: A second gene that may contribute to the 1q41-q42 deletion phenotype. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1549-1558	2.5	6
55	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. *BMC Medical Genetics*, **2018**, 19, 122	2.1	12
54	Confirmation of the Role of DHX38 in the Etiology of Early-Onset Retinitis Pigmentosa **2018**, 59, 4552-4557		9
53	Novel missense and 3RUTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. *Journal of Human Genetics*, **2018**, 63, 1099-1107	4.3	1
52	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. *Human Genetics*, **2018**, 137, 735-752	6.3	24
51	De novo variants in GREB1L are associated with non-syndromic inner ear malformations and deafness. *Human Genetics*, **2018**, 137, 459-470	6.3	14

50	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. *Human Genetics*, **2018**, 137, 471-478	6.3	9
49	A de novo splice site mutation in CASK causes FG syndrome-4 and congenital nystagmus. *American Journal of Medical Genetics, Part A*, **2017**, 173, 611-617	2.5	6
48	Ca-binding protein 2 inhibits Ca-channel inactivation in mouse inner hair cells. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E1717-E1726	11.5	26
47	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	3.5	12
46	Exploring genome-wide DNA methylation patterns in Aicardi syndrome. *Epigenomics*, **2017**, 9, 1373-1386	4.4	7
45	Cellular and Animal Models of Neurologic Disease **2017**, 114-122		
44	Compound heterozygous mutations in in a deaf child with absent cochlear nerves. *Neurology: Genetics*, **2017**, 3, e153	3.8	6
43	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	2.5	18
42	Case Report: Novel mutations in are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. *F1000Research* , **2017**, 6, 553	3.6	20
41	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	2.8	12
40	A comprehensive catalogue of the coding and non-coding transcripts of the human inner ear. *Hearing Research*, **2016**, 333, 266-274	3.9	37
39	Advances in Molecular Genetics and the Molecular Biology of Deafness. *BioMed Research International*, **2016**, 2016, 5629093	3	1
38	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. *Human Mutation*, **2016**, 37, 812-9	4.7	59
37	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	11	19
36	Delineating the GRIN1 phenotypic spectrum: A distinct genetic NMDA receptor encephalopathy. *Neurology*, **2016**, 86, 2171-8	6.5	108
35	Genome-wide association analysis demonstrates the highly polygenic character of age-related hearing impairment. *European Journal of Human Genetics*, **2015**, 23, 110-5	5.3	66
34	A De Novo Mutation in TEAD1 Causes Non-X-Linked Aicardi Syndrome **2015**, 56, 3896-904		18
33	A Frame-Shift Mutation in CAV1 Is Associated with a Severe Neonatal Progeroid and Lipodystrophy Syndrome. *PLoS ONE*, **2015**, 10, e0131797	3.7	27

32	Case Report: Compound heterozygous nonsense mutations in TRMT10A are associated with microcephaly, delayed development, and periventricular white matter hyperintensities. *F1000Research*, **2015**, 4, 912	3.6	17
31	Broadening the phenotype of LRP2 mutations: a new mutation in LRP2 causes a predominantly ocular phenotype suggestive of Stickler syndrome. *Clinical Genetics*, **2014**, 86, 282-6	4	26
30	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, 1058-64	2.6	13
29	Characterization of X chromosome inactivation using integrated analysis of whole-exome and mRNA sequencing. *PLoS ONE*, **2014**, 9, e113036	3.7	12
28	Rare variants in BMP2 and BMP4 found in otosclerosis patients reduce Smad signaling. *Otology and Neurotology*, **2014**, 35, 395-400	2.6	6
27	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	3.7	17
26	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	4.3	3
25	A sensitive and specific diagnostic test for hearing loss using a microdroplet PCR-based approach and next generation sequencing. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 145-52	2.5	56
24	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.e11-8	5.6	111
23	A mutation in CABP2, expressed in cochlear hair cells, causes autosomal-recessive hearing impairment. *American Journal of Human Genetics*, **2012**, 91, 636-45	11	77
22	Genome wide analysis in a family with sensorineural hearing loss, autism and mental retardation. *Gene*, **2012**, 510, 102-6	3.8	5
21	COL1A1 association and otosclerosis: a meta-analysis. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1066-70	2.5	15
20	Commentary on "Otosclerosis: thirty-year follow-up after surgery". *Annals of Otology, Rhinology and Laryngology*, **2011**, 120, 615-6	2.1	
19	A new locus for otosclerosis, OTSC10, maps to chromosome 1q41-44. *Clinical Genetics*, **2011**, 79, 495-7	4	17
18	Association of COL1A1 and TGFB1 polymorphisms with otosclerosis in a Tunisian population. *Annals of Human Genetics*, **2011**, 75, 598-604	2.2	18
17	Phenotype of the first otosclerosis family linked to OTSC10. *Laryngoscope*, **2011**, 121, 838-45	3.6	6
16	A 1 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**, 155A, 2021-3	2.5	8
15	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. *Human Mutation*, **2011**, 32, 825-34	4.7	62

14	Autosomal recessive Stickler syndrome in two families is caused by mutations in the COL9A1 gene **2011**, 52, 4774-9		34
13	Genetic variants in RELN are associated with otosclerosis in a non-European population from Tunisia. *Annals of Human Genetics*, **2010**, 74, 399-405	2.2	15
12	Involvement of T-cell receptor-beta alterations in the development of otosclerosis linked to OTSC2. *Genes and Immunity*, **2010**, 11, 246-53	4.4	11
11	Genetic variants in the RELN gene are associated with otosclerosis in multiple European populations. *Human Genetics*, **2010**, 127, 155-62	6.3	23
10	The etiology of otosclerosis: a combination of genes and environment. *Laryngoscope*, **2010**, 120, 1195-2026		58
9	Detection of rare nonsynonymous variants in TGFB1 in otosclerosis patients. *Annals of Human Genetics*, **2009**, 73, 171-5	2.2	25
8	A genome-wide analysis identifies genetic variants in the RELN gene associated with otosclerosis. *American Journal of Human Genetics*, **2009**, 84, 328-38	11	52
7	No evidence for association between the renin-angiotensin-aldosterone system and otosclerosis in a large Belgian-Dutch population. *Otology and Neurotology*, **2009**, 30, 1079-83	2.6	23
6	Association of bone morphogenetic proteins with otosclerosis. *Journal of Bone and Mineral Research*, **2008**, 23, 507-16	6.3	51
5	A new locus for otosclerosis, OTSC8, maps to the pericentromeric region of chromosome 9. *Human Genetics*, **2008**, 123, 267-72	6.3	46
4	Clinical and genetic analysis of two Tunisian otosclerosis families. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1653-60	2.5	6
3	A seventh locus for otosclerosis, OTSC7, maps to chromosome 6q13-16.1. *European Journal of Human Genetics*, **2007**, 15, 362-8	5.3	48
2	The coding polymorphism T263I in TGF-beta1 is associated with otosclerosis in two independent populations. *Human Molecular Genetics*, **2007**, 16, 2021-30	5.6	67
1	Transcriptional profiling of Multiple System Atrophy cerebellar tissue highlights differences between the parkinsonian and cerebellar sub-types of the disease		1