Ann Swillen

List of Publications by Citations

66 papers	3,351 citations	24 h-index	57 g-index
67 ext. papers	4,174 ext. citations	7.5 avg, IF	4.74 L-index

#	Paper	IF	Citations
66	22q11.2 deletion syndrome. *Nature Reviews Disease Primers*, **2015**, 1, 15071	51.1	492
65	Psychiatric disorders from childhood to adulthood in 22q11.2 deletion syndrome: results from the International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome. *American Journal of Psychiatry*, **2014**, 171, 627-39	11.9	472
64	Practical guidelines for managing patients with 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2011**, 159, 332-9.e1	3.6	381
63	VEGF: a modifier of the del22q11 (DiGeorge) syndrome?. *Nature Medicine*, **2003**, 9, 173-82	50.5	256
62	PTEN mutation in a family with Cowden syndrome and autism. *American Journal of Medical Genetics Part A*, **2001**, 105, 521-4		171
61	Practical guidelines for managing adults with 22q11.2 deletion syndrome. *Genetics in Medicine*, **2015**, 17, 599-609	8.1	154
60	Cognitive decline preceding the onset of psychosis in patients with 22q11.2 deletion syndrome. *JAMA Psychiatry*, **2015**, 72, 377-85	14.5	139
59	Involvement of hyperprolinemia in cognitive and psychiatric features of the 22q11 deletion syndrome. *Human Molecular Genetics*, **2007**, 16, 83-91	5.6	129
58	Neuropsychological, learning and psychosocial profile of primary school aged children with the velo-cardio-facial syndrome (22q11 deletion): evidence for a nonverbal learning disability?. *Child Neuropsychology*, **1999**, 5, 230-41	2.7	118
57	Developmental trajectories in 22q11.2 deletion. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2015**, 169, 172-81	3.1	102
56	O4.8. VULNERABLE PERIODS FOR COGNITIVE DEVELOPMENT IN INDIVIDUALS AT HIGH GENOMIC RISK OF SCHIZOPHRENIA. *Schizophrenia Bulletin*, **2018**, 44, S86-S86	1.3	78
55	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. *American Journal of Psychiatry*, **2017**, 174, 1054-1063	11.9	58
54	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 96, 753-64	11	54
53	Mathematical learning disabilities in children with 22q11.2 deletion syndrome: a review. *Developmental Disabilities Research Reviews*, **2009**, 15, 4-10		53
52	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. *Molecular Psychiatry*, **2021**, 26, 4496-4510	15.1	39
51	Basic number processing and difficulties in single-digit arithmetic: evidence from Velo-Cardio-Facial Syndrome. *Cortex*, **2009**, 45, 177-88	3.8	37
50	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. *Nature Medicine*, **2020**, 26, 1912-1918	50.5	35

49	Deletion in chromosome region 22q11 in a child with CHARGE association. *Clinical Genetics*, **1998**, 53, 408-10	4	34
48	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
47	Familial deletions of chromosome 22q11: The Leuven experience **1998**, 80, 531-532		27
46	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. *Neurology*, **2018**, 90, e2059-e2067	6.5	25
45	Early motor development in young children with 22q.11 deletion syndrome and a conotruncal heart defect. *Developmental Medicine and Child Neurology*, **2005**, 47, 797-802	3.3	25
44	The motor profile of primary school-age children with a 22q11.2 deletion syndrome (22q11.2DS) and an age- and IQ-matched control group. *Child Neuropsychology*, **2009**, 15, 532-42	2.7	24
43	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. *American Journal of Human Genetics*, **2020**, 106, 26-40	11	24
42	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. *Genome Research*, **2019**, 29, 1389-1401	9.7	21
41	A pilot study on immuno-psychiatry in the 22q11.2 deletion syndrome: A role for Th17 cells in psychosis?. *Brain, Behavior, and Immunity*, **2018**, 70, 88-95	16.6	21
40	Cerebellar hypoplasia in a patient with velo-cardio-facial syndrome. *Developmental Medicine and Child Neurology*, **1996**, 38, 949-53	3.3	21
39	The importance of understanding cognitive trajectories: the case of 22q11.2 deletion syndrome. *Current Opinion in Psychiatry*, **2016**, 29, 133-7	4.9	19
38	Malformations of the middle and inner ear on CT imaging in 22q11 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2975-2983	2.5	19
37	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. *Human Molecular Genetics*, **2018**, 27, 1150-1163	5.6	18
36	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2172-2181	2.5	18
35	Neurodevelopmental outcome in 22q11.2 deletion syndrome and management. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2160-2166	2.5	17
34	Polyhydramnios as a prenatal symptom of the DiGeorge/velo-cardio-facial syndrome **1998**, 18, 68-72		17
33	Presenting symptoms in adults with the 22q11 deletion syndrome. *European Journal of Medical Genetics*, **2014**, 57, 157-62	2.6	16
32	The cognitive spectrum in velo-cardio-facial syndrome **2005**, 147-164		15

31	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
30	Cognitive deficits in childhood, adolescence and adulthood in 22q11.2 deletion syndrome and association with psychopathology. *Translational Psychiatry*, **2020**, 10, 53	8.6	12
29	Autism and genetics: high incidence of specific genetic syndromes in 21 autistic adolescents and adults living in two residential homes in Belgium. *American Journal of Medical Genetics Part A*, **1996**, 67, 315-6		12
28	3 generation pedigree with paternal transmission of the 22q11.2 deletion syndrome: Intrafamilial phenotypic variability. *European Journal of Medical Genetics*, **2015**, 58, 244-8	2.6	11
27	Atypical language characteristics and trajectories in children with 22q11.2 deletion syndrome. *Journal of Communication Disorders*, **2018**, 75, 37-56	1.9	11
26	Numerical magnitude processing impairments in genetic syndromes: a cross-syndrome comparison of Turner and 22q11.2 deletion syndromes. *Developmental Science*, **2017**, 20, e12458	4.5	11
25	The Coffin-Siris syndrome: data on mental development, language, behavior and social skills in 12 children. *Clinical Genetics*, **1995**, 48, 177-82	4	11
24	Referential communication abilities in children with 22q11.2 deletion syndrome. *International Journal of Speech-Language Pathology*, **2017**, 19, 490-502	2.1	10
23	High prevalence of fatigue in adults with a 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 858-867	2.5	9
22	Parental report on socio-communicative behaviours in children with 22q11.2 deletion syndrome. *Journal of Intellectual and Developmental Disability*, **2017**, 42, 162-172	1.9	9
21	Lower cortisol levels and attenuated cortisol reactivity to daily-life stressors in adults with 22q11.2 deletion syndrome. *Psychoneuroendocrinology*, **2019**, 106, 85-94	5	9
20	Exploratory study on cognitive abilities and social responsiveness in children with 22q11.2 deletion syndrome (22q11DS) and children with idiopathic intellectual disability (IID). *Research in Developmental Disabilities*, **2018**, 81, 89-102	2.7	9
19	Deep Phenotyping of Development, Communication and Behaviour in Phelan-McDermid Syndrome. *Molecular Syndromology*, **2020**, 10, 294-305	1.5	9
18	A catalog of hemizygous variation in 127 22q11 deletion patients. *Human Genome Variation*, **2016**, 3, 15065	1.8	8
17	Psychotic disorder associated with 22q11.2 duplication syndrome. *Psychiatry Research*, **2016**, 236, 206-207	3.9	8
16	Low prevalence of substance use in people with 22q11.2 deletion syndrome. *British Journal of Psychiatry*, **2019**, 215, 661-667	5.4	8
15	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6
14	Affective and psychotic reactivity to daily-life stress in adults with 22q11DS: a study using the experience sampling method. *Journal of Neurodevelopmental Disorders*, **2020**, 12, 30	4.6	4

List of Publications

13	A normative chart for cognitive development in a genetically selected population. *Neuropsychopharmacology*, **2021**,	8.7	4
12	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. *Human Molecular Genetics*, **2019**, 28, 3724-3733	5.6	4
11	Developmental course of conversational behaviour of children with 22q11.2 deletion syndrome and Williams syndrome. *First Language*, **2017**, 37, 583-611	1.5	3
10	Vestibular dysfunction is a manifestation of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 448-454	2.5	3
9	Recent developments in Phelan-McDermid syndrome research: an update on cognitive development, communication and psychiatric disorders. *Current Opinion in Psychiatry*, **2021**, 34, 118-122	4.9	2
8	Blood brain barrier permeability increases with age in individuals with 22q11.2 deletion syndrome. *World Journal of Biological Psychiatry*, **2021**, 1-26	3.8	1
7	Polyhydramnios as a prenatal symptom of the DiGeorge/velo-cardio-facial syndrome **1998**, 18, 68		1
6	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology.. *American Journal of Psychiatry*, **2022**, 179, 189-203	11.9	1
5	Resilience and quality of life in young adults with a 22q11.2 deletion syndrome: a patient‘s perspective. *European Child and Adolescent Psychiatry*, **2021**, 1	5.5	0
4	Stressed parents, happy parents. An assessment of parenting stress and family quality of life in families with a child with Phelan-McDermid syndrome. *Journal of Applied Research in Intellectual Disabilities*, **2021**, 34, 1076-1088	2.2	0
3	Early motor development in young children with 22q.11 deletion syndrome and a conotruncal heart defect. *Developmental Medicine and Child Neurology*, **2007**, 47, 797-802	3.3	
2	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. *Clinical Dysmorphology*, **2020**, 29, 210-213	0.9	
1	22q11.2 Deletion Syndrome **2021**, 163-194		