Anneke T Vulto-Van Silfhout

List of Publications by Year
in descending order

28 papers	2,596 citations	394421 19 h-index	501196 28 g-index
30 all docs	30 docs citations	30 times ranked	5917 citing authors

#	Article	IF	Citations
1	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
3	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
4	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
5	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	6.2	110
6	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
7	Clinical Significance of De Novo and Inherited Copy-Number Variation. Human Mutation, 2013, 34, 1679-1687.	2.5	100
8	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
9	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	6.2	78
10	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
11	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	6.2	59
12	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	2.8	56
13	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	2.5	37
14	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	2.8	34
15	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
16	The clustering of functionally related genes contributes to CNV-mediated disease. Genome Research, 2015, 25, 802-813.	5.5	31
17	A novel MED12 mutation: Evidence for a fourth phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2377-2382.	1.2	31
18	An update on ECARUCA, the European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations. European Journal of Medical Genetics, 2013, 56, 471-474.	1.3	28

#	Article	IF	Citations
19	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
20	Personalized genetic counseling for Stargardt disease: Offspring risk estimates based on variant severity. American Journal of Human Genetics, 2022, 109, 498-507.	6.2	23
21	Establishing the phenotypic spectrum of ZTTK syndrome by analysis of 52 individuals with variants in SON. European Journal of Human Genetics, 2022, 30, 271-281.	2.8	19
22	Gene Networks Underlying Convergent and Pleiotropic Phenotypes in a Large and Systematically-Phenotyped Cohort with Heterogeneous Developmental Disorders. PLoS Genetics, 2015, 11, e1005012.	3.5	14
23	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
24	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
25	Severe Positional Central Sleep Apnea in an Asymptomatic Adult With a PHOX2B Frameshift Mutation. Journal of Clinical Sleep Medicine, 2018, 14, 1427-1430.	2.6	4
26	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	2.5	3
27	Constraint and conservation of pairedâ€type homeodomains predicts the clinical outcome of missense variants of uncertain significance. Human Mutation, 2020, 41, 1407-1424.	2.5	2
28	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1