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	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
2	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
3	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
4	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
5	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
6	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
7	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
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	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	2.8	56
10	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
11	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
12	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	2.5	3
13	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
14	A novel MED12 mutation: Evidence for a fourth phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2377-2382.	1.2	31
15	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
16	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	2.5	37
17	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
18	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

#	Article	IF	Citations
19	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
20	The clustering of functionally related genes contributes to CNV-mediated disease. Genome Research, 2015, 25, 802-813.	5.5	31
21	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
22	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
23	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
24	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
25	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
26	Clinical Significance of De Novo and Inherited Copy-Number Variation. Human Mutation, 2013, 34, 1679-1687.	2.5	100
27	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	6.2	78
28	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