Karola RehnstrÃ¶m

List of Publications by Year
in descending order

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22	8,459	394421	677142
papers	citations	19 h-index	22 g-index

23	23	23	18770
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
2	Identification of common genetic risk variants for autism spectrum disorder. Nature Genetics, 2019, 51, 431-444.	21.4	1,538
3	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
4	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	21.4	943
5	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	28.9	935
6	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
7	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	2.5	279
8	Loss-of-function nuclear factor ÎºB subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. Journal of Allergy and Clinical Immunology, 2018, 142, 1285-1296.	2.9	185
9	Deletion of TOP3Î², a component of FMRP-containing mRNPs, contributes to neurodevelopmental disorders. Nature Neuroscience, 2013, 16, 1228-1237.	14.8	144
10	Analysis of four neuroligin genes as candidates for autism. European Journal of Human Genetics, 2005, 13, 1285-1292.	2.8	136
11	Identification of Small Exonic CNV from Whole-Exome Sequence Data and Application to Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 93, 607-619.	6.2	136
12	The Genome-wide Patterns of Variation Expose Significant Substructure in a Founder Population. American Journal of Human Genetics, 2008, 83, 787-794.	6.2	132
13	The Genetic Structure of the Swedish Population. PLoS ONE, 2011, 6, e22547.	2.5	67
14	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics, 2013, 21, 659-665.	2.8	64
15	Characterization of a novel cation transporter ATPase gene (ATP13A4) interrupted by 3q25â€“q29 inversion in an individual with language delay. Genomics, 2005, 86, 182-194.	2.9	52
16	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
17	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
18	A Genome-Wide Analysis of Populations from European Russia Reveals a New Pole of Genetic Diversity in Northern Europe. PLoS ONE, 2013, 8, e58552.	2.5	32

#	Article	IF	Citations
19	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
20	Fine mapping of Xq11.1â€q21.33 and mutation screening of RPS6KA6, ZNF711, ACSL4, DLG3, and IL1RAPL2 for autism spectrum disorders (ASD). Autism Research, 2011, 4, 228-233.	3.8	18
21	Allelic variants in HTR3C show association with autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 741-746.	1.7	15
22	Phenotype mining in CNV carriers from a population cohort â€. Human Molecular Genetics, 2011, 20, 2686-2695.	2.9	13