Anne Gregor

List of Publications by Year
in descending order

12	489	1040056 9	1199594 12
papers	citations	h-index	g-index

13	13	13	1275
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	2.9	7
2	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	33
3	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
4	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. American Journal of Human Genetics, 2020, 107, 527-538.	6.2	53
5	Loss of PHF6 leads to aberrant development of human neuron-like cells. Scientific Reports, 2020, 10, 19030.	3.3	3
6	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in Drosophila melanogaster. Scientific Reports, 2020, 10, 1204.	3.3	8
7	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
8	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. American Journal of Human Genetics, 2018, 102, 44-57.	6.2	49
9	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
10	Clinical delineation of the PACS1â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
11	AlteredGPM6A/M6Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human andDrosophila. Human Mutation, 2014, 35, 1495-1505.	2.5	31
12	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	6.2	151