

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3325142/publications.pdf Version: 2024-02-01

		933447	1281871	
12	1,021	10	11	
papers	citations	h-index	g-index	
10	10	10	2542	
13	13	13	2543	
all docs	docs citations	times ranked	citing authors	

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#	Article	IF	CITATIONS
1	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	8.2	312
2	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
3	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. Blood Advances, 2019, 3, 2962-2979.	5.2	110
4	Isolation and Molecular Characterization of Circulating Melanoma Cells. Cell Reports, 2014, 7, 645-653.	6.4	91
5	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
6	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	2.9	54
7	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	8.2	34
8	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	6.4	24
9	How I curate: applying American Society of Hematology-Clinical Genome Resource Myeloid Malignancy Variant Curation Expert Panel rules for RUNX1 variant curation for germline predisposition to myeloid malignancies. Haematologica, 2020, 105, 870-887.	3.5	23
10	Utilizing ClinGen geneâ€disease validity and dosage sensitivity curations to inform variant classification. Human Mutation, 2022, 43, 1031-1040.	2.5	20
11	Revision of <i>RUNX1</i> variant curation rules. Blood Advances, 2022, 6, 4726-4730.	5.2	7
12	Myeloid Malignancy Variant Curation Expert Panel: An ASH-Sponsored Clingen Expert Panel to Optimize and Validate Acmg/AMP Variant Interpretation Guidelines for Genes Associated with Inherited Myeloid Neoplasms. Blood, 2018, 132, 5849-5849.	1.4	0