Eduardo Calpena

List of Publications by Year
in descending order

15	345	1040056 9	940533 16
papers	citations	h-index	g-index

17	17	17	960
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
2	Unexpected role of SIX1 variants in craniosynostosis: expanding the phenotype of SIX1-related disorders. Journal of Medical Genetics, 2021, , jmedgenet-2020-107459.	3.2	5
3	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
4	Dissection of contiguous gene effects for deletions around ERF on chromosome 19. Human Mutation, 2021, 42, 811-817.	2.5	2
5	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. Genetics in Medicine, 2021, 23, 2360-2368.	2.4	13
6	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	2.4	31
7	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
8	amplimap: a versatile tool to process and analyze targeted NGS data. Bioinformatics, 2019, 35, 5349-5350.	4.1	9
9	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
10	A de novo substitution in BCL11B leads to loss of interaction with transcriptional complexes and craniosynostosis. Human Molecular Genetics, 2019, 28, 2501-2513.	2.9	23
11	De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder. American Journal of Human Genetics, 2019, 104, 709-720.	6.2	41
12	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
13	Disruption ofTWIST1translation by 5â€² UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365.	2.5	10
14	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
15	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37