Daniel C Koboldt

List of Publications by Year
in descending order

25	360	840776 11	940533 16
papers	citations	h-index	g-index

25	25	25	665
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
2	Identification of Rare Variants Predisposing to Thyroid Cancer. Thyroid, 2019, 29, 946-955.	4.5	41
3	A Novel Dominant Mutation in SAG, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States. , 2017, 58, 2774.		31
4	Functional annotation of genomic variants in studies of late-onset Alzheimerâ€™s disease. Bioinformatics, 2018, 34, 2724-2731.	4.1	30
5	Quality control and integration of genotypes from two calling pipelines for whole genome sequence data in the Alzheimer's disease sequencing project. Genomics, 2019, 111, 808-818.	2.9	26
6	Somatic SLC35A2 mosaicism correlates with clinical findings in epilepsy brain tissue. Neurology: Genetics, 2020, 6, e460.	1.9	26
7	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	2.5	23
8	PTEN somatic mutations contribute to spectrum of cerebral overgrowth. Brain, 2021, 144, 2971-2978.	7.6	23
9	The Genotypic and Phenotypic Spectrum of BICD2 Variants in Spinal Muscular Atrophy. Annals of Neurology, 2020, 87, 487-496.	5.3	18
10	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
11	In-frame de novo mutation in BICD2 in two patients with muscular atrophy and arthrogryposis. Journal of Physical Education and Sports Management, 2018, 4, a003160.	1.2	14
12	Discovery of clinically relevant fusions in pediatric cancer. BMC Genomics, 2021, 22, 872.	2.8	13
13	Expanding the clinical history associated with syndromic Klippel-Feil: A unique case of comorbidity with medulloblastoma. European Journal of Medical Genetics, 2019, 62, 103701.	1.3	12
14	Genome sequencing identifies somatic BRAF duplication c.1794_1796dupTAC;p.Thr599dup in pediatric patient with low-grade ganglioglioma. Journal of Physical Education and Sports Management, 2018, 4, a002618.	1.2	7
15	Longâ€read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. Human Mutation, 2022, 43, 189-199.	2.5	7
16	Germline BAP1 Mutation in a Family With Multi-Generational Meningioma With Rhabdoid Features: A Case Series and Literature Review. Frontiers in Oncology, 2021, 11, 721712.	2.8	6
17	Inherited and de novo variants extend the etiology of TAOK1-associated neurodevelopmental disorder. Journal of Physical Education and Sports Management, 2022, , mcs.a006180.	1.2	6
18	Molecular classification of a complex structural rearrangement of the RB1 locus in an infant with sporadic, isolated, intracranial, sellar region retinoblastoma. Acta Neuropathologica Communications, 2021, 9, 61.	5.2	5

#	Article	IF	Citations
19	Early-onset Wilson disease caused by ATP7B exon skipping associated with intronic variant. Journal of Physical Education and Sports Management, 2020, 6, a005306.	1.2	4
20	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. Clinical Genetics, 2021, 100, 775-776.	2.0	4
21	Case report and review of the literature: immune dysregulation in a large familial cohort due to a novel pathogenicRELAvariant. Rheumatology, 2022, 62, 347-359.	1.9	4
22	Novel in-frame FLNB deletion causes Larsen syndrome in a three-generation pedigree. Journal of Physical Education and Sports Management, 2019, 5, a004176.	1.2	1
23	Expanding the phenotypic and molecular spectrum of NFS1 â€related disorders that cause functional deficiencies in mitochondrial and cytosolic ironâ€“sulfur cluster containing enzymes. Human Mutation, 2022, 43, 305-315.	2.5	1
24	Maternal mosaicism for a missense variant in the SMS gene that causes Snyderâ€“Robinson syndrome. Journal of Physical Education and Sports Management, 2021, 7, a006122.	1.2	0
25	Biallelic SEPSECS variants in two siblings with pontocerebellar hypoplasia type 2D underscore the relevance of splice-disrupting synonymous variants in disease.. Journal of Physical Education and Sports Management, 2022, , mcs.a006165.	1.2	0