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	Longâ€read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. Human Mutation, 2022, 43, 189-199.	2.5	7
2	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
3	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
4	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
5	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
6	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
7	PTEN somatic mutations contribute to spectrum of cerebral overgrowth. Brain, 2021, 144, 2971-2978.	7.6	23
8	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
9	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
10	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. Clinical Genetics, 2021, 100, 775-776.	2.0	4
11	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
12	Discovery of clinically relevant fusions in pediatric cancer. BMC Genomics, 2021, 22, 872.	2.8	13
13	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
14	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
15	Somatic SLC35A2 mosaicism correlates with clinical findings in epilepsy brain tissue. Neurology: Genetics, 2020, 6, e460.	1.9	26
16	The Genotypic and Phenotypic Spectrum of BICD2 Variants in Spinal Muscular Atrophy. Annals of Neurology, 2020, 87, 487-496.	5.3	18
17	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
18	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	2.5	23

#	Article	IF	Citations
19	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
20	Identification of Rare Variants Predisposing to Thyroid Cancer. Thyroid, 2019, 29, 946-955.	4.5	41
21	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
22	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
23	Functional annotation of genomic variants in studies of late-onset Alzheimerâ€™s disease. Bioinformatics, 2018, 34, 2724-2731.	4.1	30
24	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
25	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