

Laurie H Seaver

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

247
citations

1307594

7
h-index

1125743

13
g-index

15
all docs

15
docs citations

15
times ranked

698
citing authors

#	ARTICLE	IF	CITATIONS
1	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
2	IV Spectrinopathies Cause Profound Intellectual Disability, Congenital Hypotonia, and Motor Axonal Neuropathy. American Journal of Human Genetics, 2018, 102, 1158-1168.	6.2	57
3	The interface of genomic information with the electronic health record: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1431-1436.	2.4	28
4	Virus-induced genetics revealed by multidimensional precision medicine transcriptional workflow applicable to COVID-19. Physiological Genomics, 2020, 52, 255-268.	2.3	21
5	Lethal NARS2-Related Disorder Associated With Rapidly Progressive Intractable Epilepsy and Global Brain Atrophy. Pediatric Neurology, 2018, 89, 26-30.	2.1	20
6	Evolution of the phenotype of craniosynostosis with dental anomalies syndrome and report of IL11RA variant population frequencies in a Crouzon-like autosomal recessive syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 668-673.	1.2	12
7	Focused Revision: ACMG practice resource: Genetic evaluation of short stature. Genetics in Medicine, 2021, 23, 813-815.	2.4	11
8	Points to consider to avoid unfair discrimination and the misuse of genetic information: A statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 512-520.	2.4	8
9	Expanding the molecular spectrum and the neurological phenotype related to CAMTA1 variants. Clinical Genetics, 2021, 99, 259-268.	2.0	6
10	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
11	Perinatal Manifestations of DARS2-Associated Leukoencephalopathy With Brainstem and Spinal Cord Involvement and Lactate Elevation (LBSL). Child Neurology Open, 2021, 8, 2329048X2110191.	1.1	3
12	PTPN4 germline variants result in aberrant neurodevelopment and growth. Human Genetics and Genomics Advances, 2021, 2, 100033.	1.7	2
13	N-methyl-D-aspartate (NMDA) receptor genetics: The power of paralog homology and protein dynamics in defining dominant genetic variants. American Journal of Medical Genetics, Part A, 2022, 188, 556-568.	1.2	2
14	Reply to Finsterer Regarding Lethal NARS2-Related Disorder Associated With Rapidly Progressive Intractable Epilepsy and Global Brain Atrophy. Pediatric Neurology, 2019, 93, 65.	2.1	1
15	Response to Hamosh et al.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0