Scott E Hickey

List of Publications by Year
in descending order

27	724	759233 12	552781 26
papers	citations	h-index	g-index

29	29	29	1763
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cerebral organoids containing an AUTS2 missense variant model microcephaly. Brain, 2023, 146, 387-404.	7.6	11
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	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
4	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
5	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
6	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
7	Bleeding Severity and Phenotype in 22q11.2 Deletion Syndromeâ€”A Cross-Sectional Investigation. Journal of Pediatrics, 2021, 235, 220-225.	1.8	2
8	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. Clinical Genetics, 2021, 100, 775-776.	2.0	4
9	CNTN6 copy number variations: Uncertain clinical significance in individuals with neurodevelopmental disorders. European Journal of Medical Genetics, 2020, 63, 103636.	1.3	6
10	Genotype-phenotype correlation: Inheritance and variant-type infer pathogenicity in IQSEC2 gene. European Journal of Medical Genetics, 2020, 63, 103735.	1.3	12
11	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
12	Impact of Interdisciplinary Team Care for Children With 22q11.2 Deletion Syndrome. Cleft Palate-Craniofacial Journal, 2020, 57, 1362-1369.	0.9	9
13	Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. Genetics in Medicine, 2020, 22, 986-1004.	2.4	53
14	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
15	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26
16	Yield of additional genetic testing after chromosomal microarray for diagnosis of neurodevelopmental disability and congenital anomalies: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 1105-1113.	2.4	57
17	Partial tetrasomy 11q resulting from an intrachromosomal triplication of a 22 Mb region of chromosome 11. American Journal of Medical Genetics, Part A, 2017, 173, 1056-1060.	1.2	2
18	A case of constitutional trisomy 3 mosaicism in a teenage patient with mild phenotype. European Journal of Medical Genetics, 2016, 59, 569-572.	1.3	2

#	Article	IF	Citations
19	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
20	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
21	17p13.3 microduplication including CRK leads to overgrowth and elevated growth factors: A case report. European Journal of Medical Genetics, 2016, 59, 512-516.	1.3	17
22	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. Human Molecular Genetics, 2016, 25, 1255-1270.	2.9	30
23	Atypical breakpoint in a t(6;17) translocation case of acampomelic campomelic dysplasia. European Journal of Medical Genetics, 2014, 57, 315-318.	1.3	6
24	Duplication of the Xq27.3â€“q28 region, including the FMR1 gene, in an Xâ€linked hypogonadism, gynecomastia, intellectual disability, short stature, and obesity syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2294-2299.	1.2	19
25	Multigeneration family with short stature, developmental delay, and dysmorphic features due to 4q27-q28.1 microdeletion. European Journal of Medical Genetics, 2013, 56, 521-525.	1.3	3
26	A case of an atypically large proximal 15q deletion as cause for Praderâ€“Willi syndrome arising from a de novo unbalanced translocation. European Journal of Medical Genetics, 2013, 56, 510-514.	1.3	3
27	ACMG Practice Guideline: lack of evidence for MTHFR polymorphism testing. Genetics in Medicine, 2013, 15, 153-156.	2.4	124