Valentina Stanley

List of Publications by Year
in descending order

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27	881	471509 17	552781 26
papers	citations	h-index	g-index

30	30	30	2224
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	7.6	9
2	Somatic mosaicism reveals clonal distributions of neocortical development. Nature, 2022, 604, 689-696.	27.8	26
3	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	8.1	31
4	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147.	6.2	15
5	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	1.2	17
6	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
7	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. New England Journal of Medicine, 2021, 385, 1292-1301.	27.0	23
8	Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15.	28.9	27
9	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	6.2	9
10	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	1.2	20
11	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
12	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
13	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
14	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
15	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
16	Zika Virus Protease Cleavage of Host Protein Septin-2 Mediates Mitotic Defects in Neural Progenitors. Neuron, 2019, 101, 1089-1098.e4.	8.1	55
17	Bi-allelic Mutations in FAM149B1 Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2019, 104, 731-737.	6.2	23
18	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 57	3.2	15

#	Article	IF	Citations
19	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
20	A homozygous founder mutation in TRAPPC6B associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. Journal of Medical Genetics, 2018, 55, 48-54.	3.2	37
21	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
22	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
23	Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 296-304.	6.2	24
24	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
25	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
26	Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2017, 101, 441-450.	6.2	43
27	Unbiased mosaic variant assessment in sperm: a cohort study to test predictability of transmission. ELife, 0, 11, .	6.0	5