

Valentina Stanley

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9760482/publications.pdf>

Version: 2024-02-01

27
papers

881
citations

471509

17
h-index

552781

26
g-index

30
all docs

30
docs citations

30
times ranked

2224
citing authors

#	ARTICLE	IF	CITATIONS
1	Loss of tubulin deglutamylase <sc>CCP</sc> 1 causes infantile-onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
2	Biallelic loss of human CTNNA2, encoding β -N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
3	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
4	Zika Virus Protease Cleavage of Host Protein Septin-2 Mediates Mitotic Defects in Neural Progenitors. Neuron, 2019, 101, 1089-1098.e4.	8.1	55
5	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
6	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyrosis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
7	Novel congenital disorder of α -linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
8	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
9	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
10	A homozygous founder mutation in α -TRAPPC6B associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. Journal of Medical Genetics, 2018, 55, 48-54.	3.2	37
11	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	8.1	31
12	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
13	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
14	Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15.	28.9	27
15	Somatic mosaicism reveals clonal distributions of neocortical development. Nature, 2022, 604, 689-696.	27.8	26
16	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and congenital features (COFG). Tj ETQq0 0 0 rgBT # Overlock 15 Tf 50 1	6.2	25
17	Mutations in LNPB, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 296-304.	6.2	24
18	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

#	ARTICLE	IF	CITATIONS
19	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. <i>New England Journal of Medicine</i> , 2021, 385, 1292-1301.	27.0	23
20	Biallelic loss of function variants in <i>SYT2</i> cause a treatable congenital onset presynaptic myasthenic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2272-2283.	1.2	20
21	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 105, 844-853.	6.2	17
22	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	1.2	17
23	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. <i>American Journal of Human Genetics</i> , 2021, 108, 134-147.	6.2	15
24	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. <i>American Journal of Human Genetics</i> , 2021, 108, 2017-2023.	6.2	9
25	Biallelic <i>FRA10AC1</i> variants cause a neurodevelopmental disorder with growth retardation. <i>Brain</i> , 2022, 145, 1551-1563.	7.6	9
26	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. <i>Brain</i> , 2020, 143, e31-e31.	7.6	6
27	Unbiased mosaic variant assessment in sperm: a cohort study to test predictability of transmission. <i>ELife</i> , 0, 11, .	6.0	5