Christelle Golzio

List of Publications by Year
in descending order

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39 papers	4,966 citations	172457 29 h-index	289244 40 g-index
44 all docs	44 docs citations	44 times ranked	10511 citing authors

#	Article	IF	Citations
1	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. Nature Communications, 2020, 11, 2441.	12.8	37
2	De Novo Frameshift Variants in the Neuronal Splicing Factor NOVA2 Result in a Common C-Terminal Extension and Cause a Severe Form of Neurodevelopmental Disorder. American Journal of Human Genetics, 2020, 106, 438-452.	6.2	17
3	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. Human Molecular Genetics, 2019, 28, 1474-1486.	2.9	32
4	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
5	Endoglin interacts with VEGFR2 to promote angiogenesis. FASEB Journal, 2018, 32, 2934-2949.	0.5	56
6	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. Scientific Reports, 2018, 8, 10779.	3.3	50
7	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
8	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. American Journal of Human Genetics, 2017, 101, 564-577.	6.2	30
9	Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic mutation in IFT140 that causes Mainzer-Saldino syndrome. Human Genomics, 2017, 11, 16.	2.9	22
10	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	6.2	124
11	Mitochondrial Copy Number as a Biomarker for Autism?. Pediatrics, 2016, 137, e20160049.	2.1	3
12	Phosphorylation of Threonine 794 on Tie1 by Rac1/PAK1 Reveals a Novel Angiogenesis Regulatory Pathway. PLoS ONE, 2015, 10, e0139614.	2.5	8
13	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
14	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
15	Rbm8aHaploinsufficiency Disrupts Embryonic Cortical Development Resulting in Microcephaly. Journal of Neuroscience, 2015, 35, 7003-7018.	3.6	75
16	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
17	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	6.2	47
18	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. Genetics, 2014, 198, 723-733.	2.9	92

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19	Novel bone morphogenetic protein signaling through Smad2 and Smad3 to regulate cancer progression and development. FASEB Journal, 2014, 28, 1248-1267.	0.5	80
20	CHD8 regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4468-77.	7.1	297
21	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
22	Genetic architecture of reciprocal CNVs. Current Opinion in Genetics and Development, 2013, 23, 240-248.	3.3	51
23	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. American Journal of Human Genetics, 2013, 93, 798-811.	6.2	82
24	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	6.2	135
25	In Vivo Modeling of the Morbid Human Genome using Danio rerio. Journal of Visualized Experiments, 2013, , e50338.	0.3	49
26	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	21.4	226
27	Endoglin regulates PI3-kinase/Akt trafficking and signaling to alter endothelial capillary stability during angiogenesis. Molecular Biology of the Cell, 2012, 23, 2412-2423.	2.1	41
28	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
29	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	27.8	363
30	Endoglin mediates fibronectin/Î±5Î²1 integrin and TGF-Î² pathway crosstalk in endothelial cells. EMBO Journal, 2012, 31, 3885-3900.	7.8	73
31	ISL1 Directly Regulates FGF10 Transcription during Human Cardiac Outflow Formation. PLoS ONE, 2012, 7, e30677.	2.5	46
32	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
33	Phenotypic spectrum ofSTRA6mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. Human Mutation, 2009, 30, E673-E681.	2.5	89
34	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
35	Loss-of-Function Mutation in the Dioxygenase-Encoding FTO Gene Causes Severe Growth Retardation and Multiple Malformations. American Journal of Human Genetics, 2009, 85, 106-111.	6.2	340
36	Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene STRA6. American Journal of Human Genetics, 2007, 80, 1179-1187.	6.2	174

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37	Matthew-Wood syndrome: Report of two new cases supporting autosomal recessive inheritance and exclusion ofFGF10 andFGFR2. American Journal of Medical Genetics, Part A, 2007, 143A, 219-228.	1.2	12
38	The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nature Genetics, 2007, 39, 875-881.	21.4	442
39	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. Prenatal Diagnosis, 2006, 26, 1201-1205.	2.3	5