

Christelle Golzio

List of Publications by Year in descending order

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39
papers

4,966
citations

172457

29
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289244

40
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44
all docs

44
docs citations

44
times ranked

10511
citing authors

#	ARTICLE	IF	CITATIONS
1	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. <i>Cell</i> , 2014, 158, 263-276.	28.9	637
2	The ciliary gene RPGRI1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. <i>Nature Genetics</i> , 2007, 39, 875-881.	21.4	442
3	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. <i>Nature Genetics</i> , 2009, 41, 359-364.	21.4	364
4	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. <i>Nature</i> , 2012, 485, 363-367.	27.8	363
5	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. <i>Cell</i> , 2012, 150, 533-548.	28.9	347
6	Loss-of-Function Mutation in the Dioxygenase-Encoding FTO Gene Causes Severe Growth Retardation and Multiple Malformations. <i>American Journal of Human Genetics</i> , 2009, 85, 106-111.	6.2	340
7	<i>CHD8</i> regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E4468-77.	7.1	297
8	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. <i>Nature Genetics</i> , 2012, 44, 450-455.	21.4	226
9	Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene STRA6. <i>American Journal of Human Genetics</i> , 2007, 80, 1179-1187.	6.2	174
10	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. <i>American Journal of Human Genetics</i> , 2013, 92, 210-220.	6.2	135
11	Mutational, functional, and expression studies of the <i>TCF4</i> gene in Pitt-Hopkins syndrome. <i>Human Mutation</i> , 2009, 30, 669-676.	2.5	126
12	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. <i>American Journal of Human Genetics</i> , 2016, 99, 174-187.	6.2	124
13	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 245-257.	6.2	111
14	Identification of cis-suppression of human disease mutations by comparative genomics. <i>Nature</i> , 2015, 524, 225-229.	27.8	106
15	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. <i>Genetics</i> , 2014, 198, 723-733.	2.9	92
16	Phenotypic spectrum of <i>STRA6</i> mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. <i>Human Mutation</i> , 2009, 30, E673-E681.	2.5	89
17	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	6.2	86
18	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. <i>American Journal of Human Genetics</i> , 2013, 93, 798-811.	6.2	82

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19	Novel bone morphogenetic protein signaling through Smad2 and Smad3 to regulate cancer progression and development. <i>FASEB Journal</i> , 2014, 28, 1248-1267.	0.5	80
20	<i>Rbm8a</i> Haploinsufficiency Disrupts Embryonic Cortical Development Resulting in Microcephaly. <i>Journal of Neuroscience</i> , 2015, 35, 7003-7018.	3.6	75
21	Endoglin mediates fibronectin/ $\alpha 5\beta 1$ integrin and TGF- $\beta 2$ pathway crosstalk in endothelial cells. <i>EMBO Journal</i> , 2012, 31, 3885-3900.	7.8	73
22	Endoglin interacts with VEGFR2 to promote angiogenesis. <i>FASEB Journal</i> , 2018, 32, 2934-2949.	0.5	56
23	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , 2015, 96, 784-796.	6.2	53
24	Genetic architecture of reciprocal CNVs. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 240-248.	3.3	51
25	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. <i>Scientific Reports</i> , 2018, 8, 10779.	3.3	50
26	<i>In Vivo</i> Modeling of the Morbid Human Genome using <i>Danio rerio</i> . <i>Journal of Visualized Experiments</i> , 2013, , e50338.	0.3	49
27	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. <i>American Journal of Human Genetics</i> , 2015, 96, 519-531.	6.2	47
28	ISL1 Directly Regulates FGF10 Transcription during Human Cardiac Outflow Formation. <i>PLoS ONE</i> , 2012, 7, e30677.	2.5	46
29	Endoglin regulates PI3-kinase/Akt trafficking and signaling to alter endothelial capillary stability during angiogenesis. <i>Molecular Biology of the Cell</i> , 2012, 23, 2412-2423.	2.1	41
30	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. <i>Nature Communications</i> , 2020, 11, 2441.	12.8	37
31	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. <i>Human Molecular Genetics</i> , 2019, 28, 1474-1486.	2.9	32
32	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. <i>American Journal of Human Genetics</i> , 2017, 101, 564-577.	6.2	30
33	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. <i>Molecular Psychiatry</i> , 2019, 24, 1748-1768.	7.9	26
34	Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic mutation in IFT140 that causes Mainzer-Saldino syndrome. <i>Human Genomics</i> , 2017, 11, 16.	2.9	22
35	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. <i>American Journal of Human Genetics</i> , 2020, 106, 438-452.	6.2	17
36	Matthew-Wood syndrome: Report of two new cases supporting autosomal recessive inheritance and exclusion of FGF10 and FGFR2. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 219-228.	1.2	12

#	ARTICLE	IF	CITATIONS
37	Phosphorylation of Threonine 794 on Tie1 by Rac1/PAK1 Reveals a Novel Angiogenesis Regulatory Pathway. PLoS ONE, 2015, 10, e0139614.	2.5	8
38	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. Prenatal Diagnosis, 2006, 26, 1201-1205.	2.3	5
39	Mitochondrial Copy Number as a Biomarker for Autism?. Pediatrics, 2016, 137, e20160049.	2.1	3