Dian Donnai

List of Publications by Year
in descending order

28 papers	2,413 citations	361413 20 h-index	526287 27 g-index
33 all docs	33 docs citations	33 times ranked	3564 citing authors

#	Article	IF	Citations
1	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
2	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
3	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	2.9	12
4	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
5	Dysmorphology and the ESHG. European Journal of Human Genetics, 2017, 25, S33-S34.	2.8	2
6	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
7	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	1.3	83
8	Leriâ€™s pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassingGDF6andSDC2and provides insight into systemic sclerosis pathogenesis. Annals of the Rheumatic Diseases, 2015, 74, 1249-1256.	0.9	22
9	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
10	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	6.2	34
11	Donnaiâ€“Barrow syndrome (DBS/FOAR) in a child with a homozygous LRP2 mutation due to complete chromosome 2 paternal isodisomy. American Journal of Medical Genetics, Part A, 2008, 146A, 1842-1847.	1.2	55
12	Genetic contribution to neurodevelopmental disability: an overview. Developmental Medicine and Child Neurology, 2008, 45, 6-6.	2.1	1
13	Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. Nature Genetics, 2007, 39, 957-959.	21.4	284
14	Williamsâ€“Beuren Syndrome: More or less? Segmental duplications and deletions in the Williamsâ€“Beuren syndrome region provide new insights into language development. European Journal of Human Genetics, 2006, 14, 507-508.	2.8	12
15	Autosomal dominant inheritance of Williams???Beuren syndrome in a father and son with haploinsufficiency for FKBP6. Clinical Dysmorphology, 2005, 14, 61-65.	0.3	23
16	GTF2IRD1 in Craniofacial Development of Humans and Mice. Science, 2005, 310, 1184-1187.	12.6	183
17	Isolation and characterisation of GTF2IRD2, a novel fusion gene and member of the TFII-I family of transcription factors, deleted in Williamsâ€“Beuren syndrome. European Journal of Human Genetics, 2004, 12, 551-560.	2.8	51
18	How clinicians add to knowledge of development. Lancet, The, 2003, 362, 477-484.	13.7	14

#	Article	IF	Citations
19	A novel mutation in the IHH gene causes brachydactyly type A1: a 95-year-old mystery resolved. Human Genetics, 2002, 111, 368-375.	3.8	42
20	Williams syndrome: From genotype through to the cognitive phenotype. American Journal of Medical Genetics Part A, 2000, 97, 164-171.	2.4	289
21	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	2.8	147
22	A transcription factor involved in skeletal muscle gene expression is deleted in patients with Williams syndrome. European Journal of Human Genetics, 1999, 7, 737-747.	2.8	33
23	Williams Syndrome: Use of Chromosomal Microdeletions as a Tool to Dissect Cognitive and Physical Phenotypes. American Journal of Human Genetics, 1999, 64, 118-125.	6.2	245
24	A Complete Physical Contig and Partial Transcript Map of the Williams Syndrome Critical Region. Genomics, 1999, 58, 138-145.	2.9	41
25	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	21.4	255
26	Nine novel L1 CAM mutations in families with X-linked hydrocephalus. Human Mutation, 1997, 9, 512-518.	2.5	16
27	LIMâ€“kinase deleted in Williams syndrome. Nature Genetics, 1996, 13, 272-273.	21.4	136
28	Diaphragmatic hernia, exomphalos, absent corpus callosum, hypertelorism, myopia, and sensorineural deafness: A newly recognized autosomal recessive disorder?. American Journal of Medical Genetics Part A, 1993, 47, 679-682.	2.4	77