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	Williams syndrome: From genotype through to the cognitive phenotype. American Journal of Medical Genetics Part A, 2000, 97, 164-171.	2.4	289
2	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
3	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	21.4	255
4	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
5	GTF2IRD1 in Craniofacial Development of Humans and Mice. Science, 2005, 310, 1184-1187.	12.6	183
6	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	2.8	147
7	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
8	LIMâ€“kinase deleted in Williams syndrome. Nature Genetics, 1996, 13, 272-273.	21.4	136
9	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
10	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	1.3	83
11	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
12	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
13	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
14	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
15	A Complete Physical Contig and Partial Transcript Map of the Williams Syndrome Critical Region. Genomics, 1999, 58, 138-145.	2.9	41
16	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
17	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
18	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

#	Article	IF	Citations
19	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
20	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
21	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
22	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
23	Nine novel L1 CAM mutations in families with X-linked hydrocephalus. Human Mutation, 1997, 9, 512-518.	2.5	16
24	How clinicians add to knowledge of development. Lancet, The, 2003, 362, 477-484.	13.7	14
25	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
26	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	2.9	12
27	Dysmorphology and the ESHG. European Journal of Human Genetics, 2017, 25, S33-S34.	2.8	2
28	Genetic contribution to neurodevelopmental disability: an overview. Developmental Medicine and Child Neurology, 2008, 45, 6-6.	2.1	1