Anne De Paepe

List of Publications by Year
in descending order

60 papers	5,295 citations	147801 31 h-index	138484 58 g-index
63 all docs	63 docs citations	63 times ranked	7490 citing authors

#	Article	IF	Citations
1	Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	1,624
2	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	30.5	481
3	Exhaustive mutation analysis of theNF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. Human Mutation, 2000, 15, 541-555.	2.5	477
4	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
5	Bleeding and bruising in patients with Ehlersâ€“Danlos syndrome and other collagen vascular disorders. British Journal of Haematology, 2004, 127, 491-500.	2.5	190
6	Reference Values for Echocardiographic Assessment of the Diameter of the Aortic Root and Ascending Aorta Spanning All Age Categories. American Journal of Cardiology, 2014, 114, 914-920.	1.6	151
7	Novel Types of Mutation Responsible for the Dermatosparactic Type of Ehlersâ€“Danlos Syndrome (Type) Tj ETQq1 1 0.784314 rgBT /O 123, 656-663.	0.7	116
8	Dysautonomia and its underlying mechanisms in the hypermobility type of Ehlersâ€“Danlos syndrome. Seminars in Arthritis and Rheumatism, 2014, 44, 93-100.	3.4	116
9	Molecular cytogenetic and clinical findings inETV6/ABL1-positive leukemia. Genes Chromosomes and Cancer, 2001, 30, 274-282.	2.8	103
10	BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. Scientific Reports, 2016, 6, 30330.	3.3	82
11	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
12	Autonomic symptom burden in the hypermobility type of Ehlersâ€“Danlos syndrome: A comparative study with two other EDS types, fibromyalgia, and healthy controls. Seminars in Arthritis and Rheumatism, 2014, 44, 353-361.	3.4	81
13	CRISPR/Cas9-mediated homology-directed repair by ssODNs in zebrafish induces complex mutational patterns resulting from genomic integration of repair-template fragments. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	77
14	Zebrafish type I collagen mutants faithfully recapitulate human type I collagenopathies. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8037-E8046.	7.1	77
15	The natural history, including orofacial features of three patients with Ehlers-Danlos syndrome, dermatosparaxis type (EDS type VIIC). American Journal of Medical Genetics Part A, 2004, 131A, 18-28.	2.4	75
16	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
17	Loss of Type I Collagen Telopeptide Lysyl Hydroxylation Causes Musculoskeletal Abnormalities in a Zebrafish Model of Bruck Syndrome. Journal of Bone and Mineral Research, 2016, 31, 1930-1942.	2.8	65
18	Frequent allelic loss at 10q23 but low incidence ofPTEN mutations in merkel cell carcinoma. International Journal of Cancer, 2001, 92, 409-413.	5.1	63

#	Article	IF	Citations
19	Gene panel sequencing in heritable thoracic aortic disorders and related entities â€“ results of comprehensive testing in a cohort of 264 patients. Orphanet Journal of Rare Diseases, 2015, 10, 9.	2.7	62
20	RNF216 mutations as a novel cause of autosomal recessive Huntington-like disorder. Neurology, 2015, 84, 1760-1766.	1.1	59
21	Novel pathogenic COL11A1/COL11A2 variants in Stickler syndrome detected by targeted NGS and exome sequencing. Molecular Genetics and Metabolism, 2014, 113, 230-235.	1.1	48
22	Type III collagen affects dermal and vascular collagen fibrillogenesis and tissue integrity in a mutant Col3a1 transgenic mouse model. Matrix Biology, 2018, 70, 72-83.	3.6	48
23	Combined M-FISH and CGH analysis allows comprehensive description of genetic alterations in neuroblastoma cell lines. Genes Chromosomes and Cancer, 2001, 32, 126-135.	2.8	46
24	Intrinsic cardiomyopathy in Marfan syndrome: results from in-vivo and ex-vivo studies of the Fbn1C1039G/+ model and longitudinal findings in humans. Pediatric Research, 2015, 78, 256-263.	2.3	45
25	Defective Proteolytic Processing of Fibrillar Procollagens and Prodecorin Due to Biallelic BMP1 Mutations Results in a Severe, Progressive Form of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2015, 30, 1445-1456.	2.8	42
26	Ehlersâ€Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	40
27	Pharmacologic activation of wild-type p53 by nutlin therapy in childhood cancer. Cancer Letters, 2014, 344, 157-165.	7.2	39
28	Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534.	6.2	39
29	Expressed Repeat Elements Improve RT-qPCR Normalization across a Wide Range of Zebrafish Gene Expression Studies. PLoS ONE, 2014, 9, e109091.	2.5	38
30	Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891.	2.4	37
31	Orthostatic intolerance and fatigue in the hypermobility type of Ehlers-Danlos Syndrome. Rheumatology, 2016, 55, 1412-1420.	1.9	35
32	Bi-allelic AEBP1 mutations in two patients with Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2019, 28, 1853-1864.	2.9	29
33	Ehlers-Danlos Syndrome, Hypermobility Type, Is Linked to Chromosome 8p22-8p21.1 in an Extended Belgian Family. Disease Markers, 2015, 2015, 1-9.	1.3	28
34	Clinical utility gene card for: Hereditary thoracic aortic aneurysm and dissection including next-generation sequencing-based approaches. European Journal of Human Genetics, 2016, 24, 146-150.	2.8	28
35	Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831.	4.1	28
36	Genetic analysis of osteogenesis imperfecta in the <scp>P</scp>alestinian population: molecular screening of 49 affected families. Molecular Genetics & Genomic Medicine, 2018, 6, 15-26.	1.2	26

#	Article	IF	Citations
37	Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2015, 135, 992-998.	0.7	25
38	Molecular Docking Simulations Provide Insights in the Substrate Binding Sites and Possible Substrates of the ABCC6 Transporter. PLoS ONE, 2014, 9, e102779.	2.5	25
39	Pseudoxanthoma elasticum: Similar autosomal recessive subtype in Belgian and Afrikaner families. American Journal of Medical Genetics Part A, 1991, 38, 16-20.	2.4	24
40	Occipital horn syndrome and classical Menkes Syndrome caused by deep intronic mutations, leading to the activation of ATP7A pseudo-exon. European Journal of Human Genetics, 2014, 22, 517-521.	2.8	22
41	A homozygous pathogenic missense variant broadens the phenotypic and mutational spectrum of CREB3L1-related osteogenesis imperfecta. Human Molecular Genetics, 2019, 28, 1801-1809.	2.9	21
42	An Exploratory Caseâ€Control Study on the Impact of <scp>IL</scp>â€1 Gene Polymorphisms on Early Implant Failure. Clinical Implant Dentistry and Related Research, 2016, 18, 234-240.	3.7	19
43	Chromosome 2 short arm translocations revealed by M-FISH analysis of neuroblastoma cell lines. Medical and Pediatric Oncology, 2000, 35, 538-540.	1.0	17
44	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
45	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	2.4	14
46	Tissueâ€specific mosaicism for a lethal osteogenesis imperfecta COL1A1 mutation causes mild OI/EDS overlap syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1047-1050.	1.2	13
47	7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. European Journal of Medical Genetics, 2016, 59, 502-506.	1.3	10
48	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749.	2.5	9
49	<scp>VEGFA</scp> variants as prognostic markers for the retinopathy in pseudoxanthoma elasticum. Clinical Genetics, 2020, 98, 74-79.	2.0	8
50	Substitution of valine for glycine 793 in type III procollagen in Ehlers-Danlos syndrome type IV. Human Mutation, 1995, 5, 179-181.	2.5	6
51	Attitudes of cystic fibrosis patients and their parents towards direct-to-consumer genetic testing for carrier status. Personalized Medicine, 2015, 12, 99-107.	1.5	6
52	Exhaustive mutation analysis of the NF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. Human Mutation, 2000, 15, 541.	2.5	6
53	Exhaustive mutation analysis of the NF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. , 2000, 15, 541.		4
54	Genetics of the Ehlersâ€“Danlos syndrome: more than collagen disorders. Expert Opinion on Orphan Drugs, 2015, 3, 379-392.	0.8	3

#	Article	IF	Citations
55	Congenital contractural arachnodactyly due to a novel splice site mutation in the FBN2 gene. Journal of Pediatric Genetics, 2015, 03, 163-166.	0.7	3
56	Molecular cytogenetic and clinical findings in ETV6ABL1â€positive leukemia. Genes Chromosomes and Cancer, 2001, 30, 274-282.	2.8	3
57	Phenotypic and Molecular Heterogeneity in Mandibulofacial Dysostoses: A Case Series From India. Cleft Palate-Craniofacial Journal, 2022, 59, 1346-1351.	0.9	3
58	Reply to the letter to the editor by Marc Williams. Genetics in Medicine, 2011, 13, 77-78.	2.4	0
59	MicroRNA Profiling of EVI1 Deregulated Myeloid Leukemia.. Blood, 2007, 110, 4146-4146.	1.4	0
60	Downregulation of MiR-449a Is Essential for the Survival of EVI1 Positive Leukemic Cells through Modulation of NOTCH1 and BCL2.. Blood, 2009, 114, 361-361.	1.4	0