# Anne De Paepe

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4375465/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 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 | Phenotypic and Molecular Heterogeneity in Mandibulofacial Dysostoses: A Case Series From India. Cleft Palate-Craniofacial Journal, 2022, 59, 1346-1351. | 0.9 | 3 |
| 2 | 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 |
| 3 | A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131. | 2.4 | 17 |
| 4 | <scp>VEGFA</scp> variants as prognostic markers for the retinopathy in pseudoxanthoma elasticum. Clinical Genetics, 2020, 98, 74-79. | 2.0 | 8 |
| 5 | 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 |
| 6 | Bi-allelic AEBP1 mutations in two patients with Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2019, 28, 1853-1864. | 2.9 | 29 |
| 7 | Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245. | 2.4 | 66 |
| 8 | 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 |
| 9 | 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 |
| 10 | Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831. | 4.1 | 28 |
| 11 | 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 |
| 12 | 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 |
| 13 | Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227. | 6.2 | 82 |
| 14 | 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 |
| 15 | Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052. | 30.5 | 481 |
| 16 | BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. Scientific Reports, 2016, 6, 30330. | 3.3 | 82 |
| 17 | Orthostatic intolerance and fatigue in the hypermobility type of Ehlers-Danlos Syndrome. Rheumatology, 2016, 55, 1412-1420. | 1.9 | 35 |
| 18 | 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 |

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|---|---|---|---|
| 19 | 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 |
| 20 | Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891. | 2.4 | 37 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | 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 |
| 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 | 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 |
| 27 | Genetics of the Ehlersâ€“Danlos syndrome: more than collagen disorders. Expert Opinion on Orphan Drugs, 2015, 3, 379-392. | 0.8 | 3 |
| 28 | Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2015, 135, 992-998. | 0.7 | 25 |
| 29 | RNF216 mutations as a novel cause of autosomal recessive Huntington-like disorder. Neurology, 2015, 84, 1760-1766. | 1.1 | 59 |
| 30 | 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 |
| 31 | Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534. | 6.2 | 39 |
| 32 | 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 |
| 33 | 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 |
| 34 | Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749. | 2.5 | 9 |
| 35 | Expressed Repeat Elements Improve RT-qPCR Normalization across a Wide Range of Zebrafish Gene Expression Studies. PLoS ONE, 2014, 9, e109091. | 2.5 | 38 |
| 36 | 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 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | 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 |
| 40 | Pharmacologic activation of wild-type p53 by nutlin therapy in childhood cancer. Cancer Letters, 2014, 344, 157-165. | 7.2 | 39 |
| 41 | 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 |
| 42 | 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 |
| 43 | Reply to the letter to the editor by Marc Williams. Genetics in Medicine, 2011, 13, 77-78. | 2.4 | 0 |
| 44 | 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 |
| 45 | MicroRNA Profiling of EVI1 Deregulated Myeloid Leukemia.. Blood, 2007, 110, 4146-4146. | 1.4 | 0 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | 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 |
| 50 | 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 |
| 51 | Molecular cytogenetic and clinical findings inETV6/ABL1-positive leukemia. Genes Chromosomes and Cancer, 2001, 30, 274-282. | 2.8 | 103 |
| 52 | Molecular cytogenetic and clinical findings in ETV6ABL1â€positive leukemia. Genes Chromosomes and Cancer, 2001, 30, 274-282. | 2.8 | 3 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | 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 |
| 57 | Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37. | 2.4 | 1,624 |
| 58 | Ehlersâ€Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37. | 2.4 | 40 |
| 59 | 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 |
| 60 | 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 |