# Antonie D Kline

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6158449/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 43 papers | 2,760 citations | 304743 22 h-index | 276875 41 g-index |
| 53 all docs | 53 docs citations | 53 times ranked | 3696 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494. | 6.2 | 445 |
| 2 | NIPBL Mutational Analysis in 120 Individuals with Cornelia de Lange Syndrome and Evaluation of Genotype-Phenotype Correlations. American Journal of Human Genetics, 2004, 75, 610-623. | 6.2 | 277 |
| 3 | Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352. | 6.2 | 230 |
| 4 | Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance. American Journal of Medical Genetics, Part A, 2007, 143A, 1287-1296. | 1.2 | 223 |
| 5 | Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666. | 16.3 | 223 |
| 6 | De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473. | 6.2 | 124 |
| 7 | Natural history of aging in Cornelia de Lange syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 248-260. | 1.6 | 122 |
| 8 | Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900. | 2.9 | 120 |
| 9 | Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. American Journal of Medical Genetics, Part A, 2010, 152A, 1641-1653. | 1.2 | 75 |
| 10 | Growth manifestations in the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1042-1049. | 2.4 | 74 |
| 11 | De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462. | 2.5 | 72 |
| 12 | Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334. | 1.2 | 68 |
| 13 | Causes of death and autopsy findings in a large study cohort of individuals with Cornelia de Lange syndrome and review of the literature. American Journal of Medical Genetics, Part A, 2011, 155, 3007-3024. | 1.2 | 60 |
| 14 | GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants inHNRNPK. Human Mutation, 2015, 36, 1009-1014. | 2.5 | 56 |
| 15 | Developmental data on individuals with the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1053-1058. | 2.4 | 54 |
| 16 | Newborn screening for X-linked adrenoleukodystrophy: Further evidence high throughput screening is feasible. Molecular Genetics and Metabolism, 2014, 111, 55-57. | 1.1 | 51 |
| 17 | SMC1Aexpression and mechanism of pathogenicity in probands with X-Linked Cornelia de Lange syndrome. Human Mutation, 2009, 30, 1535-1542. | 2.5 | 46 |
| 18 | Congenital heart disease in Cornelia de Lange syndrome: Phenotype and genotype analysis. American Journal of Medical Genetics, Part A, 2012, 158A, 2499-2505. | 1.2 | 46 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154. | 2.5 | 45 |
| 20 | Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158. | 1.2 | 40 |
| 21 | Factors affecting quality of life in children and adolescents with hypermobile Ehlersâ€Danlos syndrome/hypermobility spectrum disorders. American Journal of Medical Genetics, Part A, 2019, 179, 561-569. | 1.2 | 39 |
| 22 | Autism traits in children and adolescents with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1400-1410. | 1.2 | 27 |
| 23 | Phenotypic spectrum of Auâ€“Kline syndrome: a report of six new cases and review of the literature. European Journal of Human Genetics, 2018, 26, 1272-1281. | 2.8 | 26 |
| 24 | Isolated NIBPL missense mutations that cause Cornelia de Lange syndrome alter MAU2 interaction. European Journal of Human Genetics, 2012, 20, 271-276. | 2.8 | 24 |
| 25 | Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. Human Molecular Genetics, 2018, 27, 3002-3011. | 2.9 | 24 |
| 26 | Characterization of sleep disturbance in Cornelia de Lange Syndrome. International Journal of Pediatric Otorhinolaryngology, 2011, 75, 215-218. | 1.0 | 21 |
| 27 | De Novo Mutations of CCNK Cause a Syndromic Neurodevelopmental Disorder with Distinctive Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 448-455. | 6.2 | 17 |
| 28 | Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464. | 2.5 | 17 |
| 29 | Cornelia de Lange syndrome: Correlation of brain MRI findings with behavioral assessment. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 190-197. | 1.6 | 14 |
| 30 | Confirmation of a cryptic unbalanced translocation using whole chromosome fluorescencein situ hybridization. American Journal of Medical Genetics Part A, 1992, 44, 477-481. | 2.4 | 12 |
| 31 | Pain and sleep quality in children with nonâ€vascular Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part A, 2018, 176, 1858-1864. | 1.2 | 12 |
| 32 | Cornelia de Lange syndrome: Further delineation of phenotype, cohesin biology and educational focus, 5th Biennial Scientific and Educational Symposium abstracts. American Journal of Medical Genetics, Part A, 2014, 164, 1384-1393. | 1.2 | 9 |
| 33 | Improvement in hearing loss over time in Cornelia de Lange syndrome. International Journal of Pediatric Otorhinolaryngology, 2016, 87, 203-207. | 1.0 | 9 |
| 34 | Further Characterization of SMC1A Loss of Function Epilepsy Distinct From Cornelia de Lange Syndrome. Journal of Child Neurology, 2022, 37, 390-396. | 1.4 | 5 |
| 35 | A de novo complex karyotype with two independent balanced translocations and a double inversion of chromosome 6 presenting with multiple congenital anomalies. American Journal of Medical Genetics Part A, 2004, 129A, 124-129. | 2.4 | 4 |
| 36 | Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance Am J Med Genet Part A 143A:1287-1296. American Journal of Medical Genetics, Part A, 2008, 146A, 2713-2713. | 1.2 | 3 |

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|---|---|---|---|
| 37 | PARS2-associated mitochondrial disease: A case report of a patient with prolonged survival and literature review. Molecular Genetics and Metabolism Reports, 2020, 24, 100613. | 1.1 | 3 |
| 38 | Repetitive and Self-injurious Behaviors in Children with Cornelia de Lange Syndrome. Journal of Autism and Developmental Disorders, 2021, 51, 1748-1758. | 2.7 | 2 |
| 39 | Cornelia de Lange syndrome and the Cohesin complex: Abstracts from the 9th Biennial Scientific and Educational Virtual Symposium 2020. American Journal of Medical Genetics, Part A, 2022, 188, 1005-1014. | 1.2 | 1 |
| 40 | Clinical, molecular, and animal model studies in Cornelia de Lange syndrome and the cohesinopathies: Abstracts from the 3rd Scientific Cornelia de Lange Syndrome Symposium, 2008. , 2009, 149A, 1615-1622. | | 0 |
| 41 | Cornelia de Lange Syndrome: A Variable Disorder of Cohesin Pathology. Current Genetic Medicine Reports, 2015, 3, 74-81. | 1.9 | 0 |
| 42 | Buggies, villi, cornelia, and genes: My extended mentorship with LG Jackson. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 83-85. | 1.6 | 0 |
| 43 | Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i. | 1.2 | 0 |