

# Antonie D Kline

## List of Publications by Year in descending order

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43  
papers

2,760  
citations

346980

22  
h-index

312153

41  
g-index

53  
all docs

53  
docs citations

53  
times ranked

3939  
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. <i>American Journal of Human Genetics</i> , 2007, 80, 485-494.	2.6	445
2	NIPBL Mutational Analysis in 120 Individuals with Cornelia de Lange Syndrome and Evaluation of Genotype-Phenotype Correlations. <i>American Journal of Human Genetics</i> , 2004, 75, 610-623.	2.6	277
3	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
4	Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1287-1296.	0.7	223
5	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. <i>Nature Reviews Genetics</i> , 2018, 19, 649-666.	7.7	223
6	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. <i>American Journal of Human Genetics</i> , 2015, 96, 462-473.	2.6	124
7	Natural history of aging in Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2007, 145C, 248-260.	0.7	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. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
9	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1641-1653.	0.7	75
10	Growth manifestations in the Brachmann-de Lange syndrome. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 1042-1049.	2.4	74
11	<i>De Novo</i> Heterozygous Mutations in <i>SMC3</i> Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. <i>Human Mutation</i> , 2015, 36, 454-462.	1.1	72
12	Noonan syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2323-2334.	0.7	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. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 3007-3024.	0.7	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 in <i>HNRNPK</i> . <i>Human Mutation</i> , 2015, 36, 1009-1014.	1.1	56
15	Developmental data on individuals with the Brachmann-de Lange syndrome. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 1053-1058.	2.4	54
16	Newborn screening for X-linked adrenoleukodystrophy: Further evidence high throughput screening is feasible. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 55-57.	0.5	51
17	<i>SMC1A</i> expression and mechanism of pathogenicity in probands with X-Linked Cornelia de Lange syndrome. <i>Human Mutation</i> , 2009, 30, 1535-1542.	1.1	46
18	Congenital heart disease in Cornelia de Lange syndrome: Phenotype and genotype analysis. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2499-2505.	0.7	46

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19	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. <i>Human Mutation</i> , 2016, 37, 148-154.	1.1	45
20	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	0.7	40
21	Factors affecting quality of life in children and adolescents with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 561-569.	0.7	39
22	Autism traits in children and adolescents with Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1400-1410.	0.7	27
23	Phenotypic spectrum of AuKline syndrome: a report of six new cases and review of the literature. <i>European Journal of Human Genetics</i> , 2018, 26, 1272-1281.	1.4	26
24	Isolated NIBPL missense mutations that cause Cornelia de Lange syndrome alter MAU2 interaction. <i>European Journal of Human Genetics</i> , 2012, 20, 271-276.	1.4	24
25	Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. <i>Human Molecular Genetics</i> , 2018, 27, 3002-3011.	1.4	24
26	Characterization of sleep disturbance in Cornelia de Lange Syndrome. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2011, 75, 215-218.	0.4	21
27	De Novo Mutations of CCNK Cause a Syndromic Neurodevelopmental Disorder with Distinctive Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 448-455.	2.6	17
28	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. <i>Human Mutation</i> , 2020, 41, 449-464.	1.1	17
29	Cornelia de Lange syndrome: Correlation of brain MRI findings with behavioral assessment. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 190-197.	0.7	14
30	Confirmation of a cryptic unbalanced translocation using whole chromosome fluorescence in situ hybridization. <i>American Journal of Medical Genetics Part A</i> , 1992, 44, 477-481.	2.4	12
31	Pain and sleep quality in children with nonvascular Ehlers-Danlos syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1858-1864.	0.7	12
32	Cornelia de Lange syndrome: Further delineation of phenotype, cohesin biology and educational focus, 5th Biennial Scientific and Educational Symposium abstracts. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1384-1393.	0.7	9
33	Improvement in hearing loss over time in Cornelia de Lange syndrome. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2016, 87, 203-207.	0.4	9
34	Further Characterization of <i>SMC1A</i> Loss of Function Epilepsy Distinct From Cornelia de Lange Syndrome. <i>Journal of Child Neurology</i> , 2022, 37, 390-396.	0.7	5
35	A de novo complex karyotype with two independent balanced translocations and a double inversion of chromosome 6 presenting with multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , 2004, 129A, 124-129.	2.4	4
36	Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance <i>Am J Med Genet Part A</i> 143A:1287-1296. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2713-2713.	0.7	3

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37	PARS2-associated mitochondrial disease: A case report of a patient with prolonged survival and literature review. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 24, 100613.	0.4	3
38	Repetitive and Self-injurious Behaviors in Children with Cornelia de Lange Syndrome. <i>Journal of Autism and Developmental Disorders</i> , 2021, 51, 1748-1758.	1.7	2
39	Cornelia de Lange syndrome and the Cohesin complex: Abstracts from the 9th Biennial Scientific and Educational Virtual Symposium 2020. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1005-1014.	0.7	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. <i>Current Genetic Medicine Reports</i> , 2015, 3, 74-81.	1.9	0
42	Buggies, villi, cornelia, and genes: My extended mentorship with LG Jackson. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 83-85.	0.7	0
43	Cover Image, Volume 173A, Number 9, September 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	0.7	0