Antonie D Kline

List of Publications by Year
in descending order

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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	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
2	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
3	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
4	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
5	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
6	Factors affecting quality of life in children and adolescents with hypermobile Ehlersâ€Ðanlos syndrome/hypermobility spectrum disorders. American Journal of Medical Genetics, Part A, 2019, 179, 561-569.	1.2	39
7	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
8	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
9	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
10	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
11	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
12	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
13	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
14	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
15	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
16	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
17	Improvement in hearing loss over time in Cornelia de Lange syndrome. International Journal of Pediatric Otorhinolaryngology, 2016, 87, 203-207.	1.0	9
18	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

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19	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
20	Cornelia de Lange Syndrome: A Variable Disorder of Cohesin Pathology. Current Genetic Medicine Reports, 2015, 3, 74-81.	1.9	0
21	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
22	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
23	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
24	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
25	Newborn screening for X-linked adrenoleukodystrophy: Further evidence high throughput screening is feasible. Molecular Genetics and Metabolism, 2014, 111, 55-57.	1.1	51
26	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
27	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
28	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
29	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
30	Characterization of sleep disturbance in Cornelia de Lange Syndrome. International Journal of Pediatric Otorhinolaryngology, 2011, 75, 215-218.	1.0	21
31	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
32	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
33	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
34	SMC1Aexpression and mechanism of pathogenicity in probands with X-Linked Cornelia de Lange syndrome. Human Mutation, 2009, 30, 1535-1542.	2.5	46
35	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
36	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

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37	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
38	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
39	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
40	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
41	Growth manifestations in the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1042-1049.	2.4	74
42	Developmental data on individuals with the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1053-1058.	2.4	54
43	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