

CITATION REPORT

List of articles citing

De novo heterozygous mutations in SMC3 cause a range of Cornelia de Lange syndrome-overlapping phenotype

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Human Mutation, 2015, 36, 454-62.

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#	Paper	IF	Citations
64	Cohesin and human disease: lessons from mouse models. <i>Current Opinion in Cell Biology</i> , 2015 , 37, 9-17	9	27
63	Response to Dylan Mordaunt and Alisha McLauchlan. <i>Clinical Genetics</i> , 2015 , 88, 99-100	4	
62	Pathway Analysis Based on a Genome-Wide Association Study of Polycystic Ovary Syndrome. <i>PLoS ONE</i> , 2015 , 10, e0136609	3.7	27
61	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015 , 97, 922-32	11	70
60	Intellectual Disability: When the Hypertrichosis Is a Clue. <i>Journal of Pediatric Genetics</i> , 2015 , 4, 154-8	0.7	5
59	Deletion of 11q12.3-11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome. <i>Gene</i> , 2015 , 572, 130-134	3.8	8
58	A 6-year-old boy with Cornelia de Lange syndrome and Coats disease: case report and review of the literature. <i>Journal of AAPOS</i> , 2015 , 19, 474-8	1.3	4
57	Biochemical and structural characterization of HDAC8 mutants associated with Cornelia de Lange syndrome spectrum disorders. <i>Biochemistry</i> , 2015 , 54, 6501-13	3.2	32
56	Disorders of Transcriptional Regulation: An Emerging Category of Multiple Malformation Syndromes. <i>Molecular Syndromology</i> , 2016 , 7, 262-273	1.5	25
55	Expanding the clinical spectrum of the 'HDAC8-phenotype' - implications for molecular diagnostics, counseling and risk prediction. <i>Clinical Genetics</i> , 2016 , 89, 564-73	4	29
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52	Identification of a region in the coiled-coil domain of Smc3 that is essential for cohesin activity. <i>Nucleic Acids Research</i> , 2016 , 44, 6309-17	20.1	11
51	Chromosomal abnormalities and copy number variations in fetal left-sided congenital heart defects. <i>Prenatal Diagnosis</i> , 2016 , 36, 177-85	3.2	10
50	Molecular subtyping and improved treatment of neurodevelopmental disease. <i>Genome Medicine</i> , 2016 , 8, 22	14.4	12
49	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. <i>Human Genetics</i> , 2017 , 136, 307-320	6.3	40
48	Genotype-phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1566-1574	2.5	16

47	Two-step ATP-driven opening of cohesin head. <i>Scientific Reports</i> , 2017 , 7, 3266	4.9	14
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43	Zebrafish as a Model to Study Cohesin and Cohesinopathies. <i>Methods in Molecular Biology</i> , 2017 , 1515, 177-196	1.4	4
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41	A new twist in the coil: functions of the coiled-coil domain of structural maintenance of chromosome (SMC) proteins. <i>Current Genetics</i> , 2018 , 64, 109-116	2.9	15
40	Novel mosaic variants in two patients with Cornelia de Lange syndrome. <i>European Journal of Medical Genetics</i> , 2018 , 61, 680-684	2.6	6
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22	Chromatin Alterations in Neurological Disorders and Strategies of (Epi)Genome Rescue. <i>Pharmaceuticals</i> , 2021 , 14,	5.2	0
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5	A Novel Variant in RAD21 in Cornelia De Lange Syndrome Type 4: Case Report and Bioinformatic Analysis. 2023 , 14, 119		1
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1	Genetic and genomic analyses of <i>Drosophila melanogaster</i> models of chromatin modification disorders.		0