

Loss-of-function HDAC8 mutations cause a phenotypic syndrome-like features, ocular hypertelorism, large fontanelles

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	1.5	141
2	A commentary on exome sequencing identifies a de novo mutation in HDAC8 associated with Cornelia de Lange syndrome. <i>Journal of Human Genetics</i> , 2014, 59, 479-479.	1.1	1
3	De Novo variants in the KMT2A (MLL) gene causing atypical Wiedemann-Steiner syndrome in two unrelated individuals identified by clinical exome sequencing. <i>BMC Medical Genetics</i> , 2014, 15, 49.	2.1	49
4	Exome sequencing identifies a de novo mutation in HDAC8 associated with Cornelia de Lange syndrome. <i>Journal of Human Genetics</i> , 2014, 59, 536-539.	1.1	22
5	Compromised Structure and Function of HDAC8 Mutants Identified in Cornelia de Lange Syndrome Spectrum Disorders. <i>ACS Chemical Biology</i> , 2014, 9, 2157-2164.	1.6	56
6	SMC1B is present in mammalian somatic cells and interacts with mitotic cohesin proteins. <i>Scientific Reports</i> , 2015, 5, 18472.	1.6	24
7	Cohesin and human disease: lessons from mouse models. <i>Current Opinion in Cell Biology</i> , 2015, 37, 9-17.	2.6	35
8	HDAC8 deficiency causes an X-linked dominant disorder with a wide range of severity. <i>Clinical Genetics</i> , 2015, 88, 98-98.	1.0	4
9	Response to Dylan Mordaunt and Alisha McLauchlan. <i>Clinical Genetics</i> , 2015, 88, 99-100.	1.0	0
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11	<i>Drosophila</i> Nipped-B Mutants Model Cornelia de Lange Syndrome in Growth and Behavior. <i>PLoS Genetics</i> , 2015, 11, e1005655.	1.5	33
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14	De Novo Heterozygous Mutations in SMC3 Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. <i>Human Mutation</i> , 2015, 36, 454-462.	1.1	72
15	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	1.4	37
16	HDAC8: a multifaceted target for therapeutic interventions. <i>Trends in Pharmacological Sciences</i> , 2015, 36, 481-492.	4.0	210
17	Novel SMC1A frameshift mutations in children with developmental delay and epilepsy. <i>European Journal of Medical Genetics</i> , 2015, 58, 562-568.	0.7	26
18	Biochemical and Structural Characterization of HDAC8 Mutants Associated with Cornelia de Lange Syndrome Spectrum Disorders. <i>Biochemistry</i> , 2015, 54, 6501-6513.	1.2	41

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20	Histone Deacetylases in Bone Development and Skeletal Disorders. <i>Physiological Reviews</i> , 2015, 95, 1359-1381.	13.1	122
21	Cornelia de Lange syndrome. <i>Clinical Genetics</i> , 2015, 88, 1-12.	1.0	157
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