



Articles related to CdLS go back to the 1980s and are divided by topic. To access a summary of an article, go to PubMed, then type the PMID code (indicated below after each article) in the Search for function at the top of the page.

- Anesthesia
- Behavior & Communications
- Clinical Features
- Genes
- Model Organisms (Animals)
- Prenatal
- Reviews
- Scientific Symposia

Anesthesia

- August DA, Sorhabi S. 2009. Is a difficult airway predictable in Cornelia de Lange syndrome? *Paediatr Anaesth* 19:707-9. PMID: 19638123

Behavior & Communication

- Arron K, Oliver C, Hall S, Sloneem J, Forman D, McClintock K. 2006. Effects of social context on social interaction and self-injurious behavior in Cornelia de Lange syndrome. *Am J Ment Retardation* 111:184-92. Erratum in: *Am J Ment Retard.* 2006 111:298. PMID: 16597185
- Goodban MT. 1993. Survey of speech and language skills with prognostic indicators in 116 patients with Cornelia de Lange syndrome. *Am J Med Genet* 47:1059-63. PMID: 8291523
- Hyman P, Oliver C. 2001. Causal explanations, concern and optimism regarding self-injurious behaviour displayed by individuals with Cornelia de Lange syndrome: the parents' perspective. *J Intellect Disabil Res* 45:326-34. PMID: 11489054
- Hyman P, Oliver C, Hall S. 2002. Self-injurious behavior, self-restraint, and compulsive behaviors in Cornelia de Lange syndrome. *Am J Ment Retardation* 107:146-54. PMID: 11853532



- Sarimski K. 2002. Analysis of intentional communication in severely handicapped children with Cornelia-de-Lange syndrome. *J Commun Disord* 35:483-500. PMID: 12443049
- Basile E, Villa L, Selicorni A, Molteni M. 2007. The behavioural phenotype of Cornelia de Lange Syndrome: a study of 56 individuals. *J Intellect Disabil Res* 51:671-81. PMID: 17845236
- Collis L, Moss J, Jutley J, Cornish K, Oliver C. 2008. Facial expression of affect in children with Cornelia de Lange syndrome. *J Intellect Disabil Res* 52:207-15. PMID: 18261020
- Hall SS, Arron K, Sloneem J, Oliver C. 2008. Health and sleep problems in Cornelia de Lange Syndrome: a case control study. *J Intellect Disabil Res* 52:458-68. PMID: 18341525
- Oliver C, Sloneem J, Hall S, Arron K. 2009. Self-injurious behaviour in Cornelia de Lange syndrome: 1. Prevalence and phenomenology. *J Intellect Disabil Res* 53:575-89. PMID: 19522788
- Oliver C, Arron K, Sloneem J, Hall S. 2008. Behavioural phenotype of Cornelia de Lange syndrome: case-control study. *Br J Psychiatry* 193:466-70. PMID: 19043149
- Sloneem J, Arron K, Hall SS, Oliver C. 2009. Self-injurious behaviour in Cornelia de Lange syndrome: 2. Association with environmental events. *J Intellect Disabil Res* 53:590-603. PMID: 19533839
- Richards C, Moss J, O'Farrell L, Kaur G, Oliver C. 2009. Social anxiety in Cornelia de Lange syndrome. *J Autism Dev Disord* 39:1155-62. PMID: 19330433
- Richman D, Belmont J, Kim M, Slavin C, Hayner A. 2009. Parenting Stress in Families of Children with Cornelia de Lange Syndrome and Down Syndrome. *J Dev Phys Disabil*, DOI 10.1007/s10882-009-9156-6.
- Contrasting age related changes in autism spectrum disorder phenomenology in Cornelia de Lange, Fragile X, and Cri du Chat syndromes: Results from a 2.5-year follow-up. Cochran L, Moss J, Nelson L, Oliver C. *Am J Med Genet C Semin Med Genet*. 2015 Jun;169(2):188-97. doi: 10.1002/ajmg.c.31438. Epub 2015 May 18. PMID: 25989416
- Autism traits in children and adolescents with Cornelia de Lange syndrome. Srivastava S, Landy-Schmitt C, Clark B, Kline AD, Specht M, Grados MA. *Am J Med Genet A*. 2014 Jun;164A(6):1400-10. doi: 10.1002/ajmg.a.36573. Epub 2014 Apr 9. PMID: 24718998
- Investigation of autistic features among individuals with mild to moderate Cornelia de Lange syndrome. Nakanishi M, Deardorff MA, Clark D, Levy SE, Krantz I, Pipan M. *Am J Med Genet A*. 2012 Aug;158A(8):1841-7. doi: 10.1002/ajmg.a.34014. Epub 2012 Jun 27. PMID: 22740374



- Characteristics of autism spectrum disorder in Cornelia de Lange syndrome. Moss J, Howlin P, Magiati I, Oliver C. *J Child Psychol Psychiatry*. 2012 Aug;53(8):883-91. doi: 10.1111/j.1469-7610.2012.02540.x. Epub 2012 Apr 10. PMID: 2249001
- A longitudinal follow-up study of affect in children and adults with Cornelia de Lange syndrome. Nelson L, Moss J, Oliver C. *Am J Intellect Dev Disabil*. 2014 May;119(3):235-52. doi: 10.1352/1944-7558-119.3.235. PMID: 24871792

Clinical Features

- Hawley PP, Jackson LG, Kurnit DM. 1985. Sixty-four patients with Brachmann-de Lange syndrome: a survey. *Am J Med Genet* 20:453-9. PMID: 3993674
- Opitz JM. 1985. The Brachmann-de Lange syndrome. *Am J Med Genet* 22:89-102. PMID: 3901753
- Ireland M, Donnai D, Burn J. 1993. Brachmann-de Lange syndrome. Delineation of the clinical phenotype. *Am J Med Genet* 47:959-63. PMID: 8291539
- Jackson L, Kline AD, Barr MA, Koch S. 1993. de Lange syndrome: a clinical review of 310 individuals. *Am J Med Genet* 47:940-46. PMID: 8291537
- Kline AD, Barr M, Jackson LG. 1993. Growth manifestations in Brachmann-de Lange syndrome. *Am J Med Genet* 47: 1042-49. PMID: 8291521
- Kline AD, Stanley C, Belevich J, Brodsky K, Barr M, Jackson LG. 1993. Developmental data on individuals with Brachmann-de Lange syndrome. *Am J Med Genet* 47: 1053-58. PMID: 7507292
- van Allen MI, Filippi G, Siegel-Bartelt J, Yong SL, McGillivray B, Zuker RM, Smith CR, Magee JF, Ritchie S, Toi A, et al. 1993. Clinical variability within Brachmann-de Lange syndrome: a proposed classification system. *Am J Med Genet* 47:947-58. PMID: 8291538
- Wagnanski-Jaffe T, Shin J, Perruzza E, Abdolell M, Jackson LG, Levin AV. 2005. Ophthalmologic findings in the Cornelia de Lange Syndrome. *J AAPOS* 9:407-15. PMID: 16213388



- Kline AD, Grados M, Sponseller P, Levy HP, Blagowidow N, Schoedel C, Rampolla J, Clemens DK, Krantz I, Kimball A, Pichard C, Tuchman D. 2007. Natural history of aging in Cornelia de Lange syndrome. *Am J Med Genet C Semin Med Genet* 45C:248-60. PMID: 17640042
- Kline AD, Krantz ID, Sommer A, Kliewer M, Jackson LG, FitzPatrick DR, Levin AV, Selicorni A. 2007. Cornelia de Lange syndrome: clinical review, diagnostic and scoring systems, and anticipatory guidance. *Am J Med Genet A* 143A:1287-96. PMID: 17640042
- Barisic I, Tokic V, Loane M, Bianchi F, Calzolari E, Garne E, Wellesley D, Dolk H; EUROCAT Working Group. 2008. Descriptive epidemiology of Cornelia de Lange syndrome in Europe. *Am J Med Genet A* 146A:51-9. PMID: 18074387
- Hunter AG, Collins JS, Deardorff MA, Krantz ID. 2009. Detailed assessment of the ear in Cornelia de Lange syndrome: comparison with a control sample using the new dysmorphology guidelines. *Am J Med Genet A* 149A:2181-92. PMID: 19764039
- Castronovo P, Gervasini C, Cereda A, Masciadri M, Milani D, Russo S, Selicorni A, Larizza L. 2009. Premature chromatid separation is not a useful diagnostic marker for Cornelia de Lange syndrome. *Chromosome Res* 17:763-71. PMID: 19690971
- Immunologic features of Cornelia de Lange syndrome. Jyonouchi S, Orange J, Sullivan KE, Krantz I, Deardorff M. *Pediatrics*. 2013 Aug;132(2):e484-9. doi: 10.1542/peds.2012-3815. Epub 2013 Jul 1. PMID: 2382169
- Congenital heart disease in Cornelia de Lange syndrome: phenotype and genotype analysis. Chatfield KC, Schrier SA, Li J, Clark D, Kaur M, Kline AD, Deardorff MA, Jackson LS, Goldmuntz E, Krantz ID. *Am J Med Genet A*. 2012 Oct;158A(10):2499-505. doi: 0.1002/ajmg.a.35582. Epub 2012 Sep 10. PMID: 22965847
- Insomnia in Cornelia de Lange syndrome. Rajan R, Benke JR, Kline AD, Levy HP, Kimball A, Mettel TL, Boss EF, Ishman SL. *Int J Pediatr Otorhinolaryngol*. 2012 Jul;76(7):972-5. doi: 10.1016/j.ijporl.2012.03.008. Epub 2012 Apr 13. PMID: 2250344
- Audiological findings, genotype and clinical severity score in Cornelia de Lange syndrome. Marchisio P, Selicorni A, Bianchini S, Milani D, Baggi E, Cerutti M, Larizza L, Principi N, Esposito S. *Int J Pediatr Otorhinolaryngol*. 2014 Jul;78(7):1045-8. doi: 0.1016/j.ijporl.2014.03.038. Epub 2014 Apr 8. PMID: 24774220
- Causes of death and autopsy findings in a large study cohort of individuals with Cornelia de Lange syndrome and review of the literature. Schrier SA, Sherer I, Deardorff MA, Clark D, Audette L, Gillis L, Kline AD, Ernst L, Loomes K, Krantz



ID, Jackson LG. Am J Med Genet A. 2011 Dec;155A(12):3007-24. doi: 10.1002/ajmg.a.34329. Epub 2011 Nov 8. Review. PMID: 22069164

Genes

HDAC8

- Exome sequencing identifies a de novo mutation in HDAC8 associated with Cornelia de Lange syndrome. Feng L, Zhou D, Zhang Z, Liu Y, Yang Y. J Hum Genet. 2015 Mar;60(3):165. doi: 10.1038/jhg.2014.113. No abstract available. PMID: 25805374
- HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Deardorff MA, Bando M, Nakato R, et al. Nature. 2012 Sep 13;489(7415):313-7. doi: 10.1038/nature11316. PMID: 22885700
- Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Kaiser FJ, Ansari M, Braunholz D, et al. Hum Mol Genet. 2014 Jun 1;23(11):2888-900. doi: 10.1093/hmg/ddu002. Epub 2014 Jan 8. PMID: 24403048

NIPBL

- Krantz ID, McCallum J, DeScipio C, Kaur M, Gillis LA, Yaeger D, Jukofsky L, Wasserman N, Bottani A, Morris CA, Nowaczyk MJ, Toriello H, Bamshad MJ, Carey JC, Rappaport E, Kawauchi S, Lander AD, Calof AL, Li HH, Devoto M, Jackson LG. 2004. Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. Nat Genet 36:631-5. PMID: 15146186
- Tonkin ET, Wang TJ, Lisgo S, Bamshad MJ, Strachan T. 2004. NIPBL, encoding a homolog of fungal Scc2-type sister chromatid cohesion proteins and fly Nipped-B, is mutated in Cornelia de Lange syndrome. Nat Genet 36:636-41. PMID: 15146185
- Borck G, Redon R, Sanlaville D, Rio M, Prieur M, Lyonnet S, Vekemans M, Carter NP, Munnich A, Colleaux L, Cormier-Daire V. 2004. NIPBL mutations and genetic heterogeneity in Cornelia de Lange syndrome. J Med Genet 41:e128. PMID: 15591270
- Gillis LA, McCallum J, Kaur M, DeScipio C, Yaeger D, Mariani A, Kline AD, Li HH, Devoto M, Jackson LG, Krantz ID. 2004. NIPBL mutational analysis in 120 individuals with Cornelia de Lange syndrome and evaluation of genotype-phenotype correlations. Am J Hum Genet 75:610-23. PMID: 15318302
- Miyake N, Visser R, Kinoshita A, Yoshiura K, Niikawa N, Kondoh T, Matsumoto N, Harada N, Okamoto N, Sonoda T, Naritomi K, Kaname T, Chinen Y, Tonoki H,



Kurosawa K. 2005. Four novel NIPBL mutations in Japanese patients with Cornelia de Lange syndrome. *Am J Med Genet A* 135:103–5. PMID: 15723327

- Bhuiyan ZA, Klein M, Hammond P, van Haeringen A, Mannens MM, Van Berckelaer-Onnes I, Hennekam RC. 2006. Genotype-phenotype correlations of 39 patients with Cornelia De Lange syndrome: the Dutch experience. *J Med Genet* 43:568-75. PMID: 16236812
- Borck G, Zarhrate M, Cluzeau C, Bal E, Bonnefont JP, Munnich A, Cormier-Daire V, Colleaux L. 2006. Father-to-daughter transmission of Cornelia de Lange syndrome caused by a mutation in the 5' untranslated region of the NIPBL Gene. *Hum Mutat* 27:731-5. PMID: 16799922
- Yan J, Saifi GM, Wierzba TH, Withers M, Bien-Willner GA, Limon J, Stankiewicz P, Lupski JR, Wierzba J. 2006. Mutational and genotype-phenotype correlation analyses in 28 Polish patients with Cornelia de Lange syndrome. *Am J Med Genet A* 140:1531-41. PMID: 16770807
- Nipbl and mediator cooperatively regulate gene expression to control limb development. Muto A, Ikeda S, Lopez-Burks ME, et al. *PLoS Genet*. 2014 Sep 25;10(9):e1004671. doi: 10.1371/journal.pgen.1004671. eCollection 2014 Sep. PMID: 25255084
- Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Ansari M, Poke G, Ferry Q, et al. *J Med Genet*. 2014 Oct;51(10):659-68. doi: 10.1136/jmedgenet-2014-102573. Epub 2014 Aug 14. PMID: 25125236

RAD21

- RAD21 mutations cause a human cohesinopathy. Deardorff MA, Wilde JJ, Albrecht M, et al. *Am J Hum Genet*. 2012 Jun 8;90(6):1014-27. doi: 10.1016/j.ajhg.2012.04.019. Epub 2012 May 24. PMID: 22633399

SMC1A & SMC3

- Musio A, Selicorni A, Focarelli ML, Gervasini C, Milani D, Russo S, Vezzoni P, Larizza L. 2006. X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. *Nat Genet* 38:528-30. PMID: 16604071
- Borck G, Zarhrate M, Bonnefont JP, Munnich A, Cormier-Daire V, Colleaux L. 2007. Incidence and clinical features of X-linked Cornelia de Lange syndrome due to SMC1L1 mutations. *Hum Mutat* 28:205-6. PMID: 17221863
- Deardorff MA, Kaur M, Yaeger D, Rampuria A, Korolev S, Pie J, Gil-Rodríguez C, Arnedo M, Loeys B, Kline AD, Wilson M, Lillquist K, Siu V, Ramos FJ, Musio A, Jackson LS, Dorsett D, Krantz ID. 2007. Mutations in cohesin complex members SMC3



and SMC1A cause a mild variant of Cornelia de Lange syndrome with predominant mental retardation. *Am J Hum Genet* 80:485-94. PMID: 17273969

- De novo heterozygous mutations in SMC3 cause a range of Cornelia de Lange syndrome-overlapping phenotypes. Gil-Rodríguez MC, Deardorff MA, Ansari M, et al. *J. Hum Mutat.* 2015 Apr;36(4):454-62. doi: 10.1002/humu.22761. Epub 2015 Mar 17. PMID: 25655089

Unknown Genomic Imbalance

- Schoumans J, Wincent J, Barbaro M, Djureinovic T, Maguire P, Forsberg L, Staaf J, Thuresson AC, Borg A, Nordgren A, Malm G, Anderlid BM. 2007. Comprehensive mutational analysis of a cohort of Swedish Cornelia de Lange syndrome patients. *Eur J Hum Genet* 15:143-9. PMID: 17106445
- Gervasini C, Pfundt R, Castronovo P, Russo S, Roversi G, Masciadri M, Milani D, Zampino G, Selicorni A, Schoenmakers EF, Larizza L. 2008. Search for genomic imbalances in a cohort of 24 Cornelia de Lange patients negative for mutations in the NIPBL and SMC1L1 genes. *Clin Genet* 74:531-8. PMID: 18798846

Cohesin in Human Gene Expression & DNA Repair

- Vrouwe MG, Elghalbzouri-Maghrani E, Meijers M, Schouten P, Godthelp BC, Bhuiyan ZA, Redeker EJ, Mannens MM, Mullenders LH, Pastink A, Darroudi F. 2007. Increased DNA damage sensitivity of Cornelia de Lange syndrome cells: evidence for impaired recombinational repair. *Hum Mol Genet* 16:1478-87. PMID: 17468178
- Wendt KS, Yoshida K, Itoh T, Bando M, Koch B, Schirghuber E, Tsutsumi S, Nagae G, Ishihara K, Mishihiro T, Yahata K, Imamoto F, Aburatani H, Nakao M, Imamoto N, Maeshima K, Shirahige K, Peters JM. 2008. Cohesin mediates transcriptional insulation by CCCTC-binding factor. *Nature* 451:796-801. PMID: 18235444
- Liu J, Zhang Z, Bando M, Itoh T, Deardorff MA, Clark D, Kaur M, Tandy S, Kondoh T, Rappaport E, Spinner NB, Vega H, Jackson LG, Shirahige K, Krantz ID. 2009. Transcriptional dysregulation in NIPBL and cohesin mutant human cells. *PLoS Biol* 7:e1000119. PMID: 19468298
- Revenkova E, Focarelli ML, Susani L, Paulis M, Bassi MT, Mannini L, Frattini A, Delia D, Krantz I, Vezzoni P, Jessberger R, Musio A. 2009. Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. *Hum Mol Genet* 18:418-27. PMID: 18996922



- Liu J, Feldman R, Zhang Z, Deardorff MA, Haverfield EV, Kaur M, Li JR, Clark D, Kline AD, Waggoner DJ, Das S, Jackson LG, Krantz ID. 2009. SMC1A expression and mechanism of pathogenicity in probands with X-Linked Cornelia de Lange syndrome. *Hum Mutat.* 2009 Jul 15. [Epub ahead of print] PMID: 19701948
- Liu J, Feldman R, Zhang Z, Deardorff MA, Haverfield EV, Kaur M, Li JR, Clark D, Kline AD, Waggoner DJ, Das S, Jackson LG, Krantz ID. 2009. SMC1A expression and mechanism of pathogenicity in probands with X-Linked Cornelia de Lange syndrome. *Hum Mutat.* 2009 Jul 15. [Epub ahead of print] PMID: 19701948
- Gard S, Light W, Xiong B, Bose T, McNairn AJ, Harris B, Fleharty B, Seidel C, Brickner JH, Gerton JL. Cohesinopathy mutations disrupt the subnuclear organization of chromatin. *J Cell Biol.* 2009 Nov 16;187(4):455-62. PMID: 19948494. Free news summary: <http://jcb.rupress.org/cgi/cont>

Model Organisms (Animals)

Drosophila (fruit fly)

- Rollins RA, Morcillo P, Dorsett D. 1999. Nipped-B, a *Drosophila* homologue of chromosomal adherins, participates in activation by remote enhancers in the cut and Ultrabithorax genes. *Genetics* 152:577-93. PMID: 10353901
- Rollins RA, Korom M, Aulner N, Martens A, Dorsett D. 2004. *Drosophila* nipped-B protein supports sister chromatid cohesion and opposes the stromalin/Scs3 cohesion factor to facilitate long-range activation of the cut gene. *Mol Cell Biol* 24:3100-11. PMID: 15060134
- Dorsett D, Eissenberg JC, Misulovin Z, Martens A, Redding B, McKim K. 2005. Effects of sister chromatid cohesion proteins on cut gene expression during wing development in *Drosophila*. *Development* 132:4743-53. PMID: 16207752
- Hallson G, Syrzycka M, Beck SA, Kennison JA, Dorsett D, Page SL, Hunter SM, Keall R, Warren WD, Brock HW, Sinclair DA, Honda BM. 2008. The *Drosophila* cohesin subunit Rad21 is a trithorax group (trxG) protein. *Proc Natl Acad Sci U S A* 105:12405-10. PMID: 18713858
- Misulovin Z, Schwartz YB, Li XY, Kahn TG, Gause M, MacArthur S, Fay JC, Eisen MB, Pirrotta V, Biggin MD, Dorsett D. 2008. Association of cohesin and Nipped-B with transcriptionally active regions of the *Drosophila melanogaster* genome. *Chromosoma* 117:89-102. PMID: 17965872



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.

- Pauli A, Althoff F, Oliveira RA, Heidmann S, Schuldiner O, Lehner CF, Dickson BJ, Nasmyth K. 2008. Cell-type-specific TEV protease cleavage reveals cohesin functions in *Drosophila* neurons. *Dev Cell* 14:239-51. PMID: 18267092
- Schuldiner O, Berdnik D, Levy JM, Wu JS, Luginbuhl D, Gontang AC, Luo L. 2008. piggyBac-based mosaic screen identifies a postmitotic function for cohesin in regulating developmental axon pruning. *Dev Cell* 14:227-38. PMID: 18267091
- Schaaf CA, Misulovin Z, Sahota G, Siddiqui AM, Schwartz YB, Kahn TG, Pirrotta V, Gause M, Dorsett D. 2009. Regulation of the *Drosophila* Enhancer of split and invected-engrailed gene complexes by sister chromatid cohesion proteins. *PLoS One* 4:e6202. PMID: 19587787

C. Elegans & Xenopus (worm and frog)

- Seitan VC, Banks P, Laval S, Majid NA, Dorsett D, Rana A, Smith J, Bateman A, Krpic S, Hostert A, Rollins RA, Erdjument-Bromage H, Tempst P, Benard CY, Hekimi S, Newbury SF, Strachan T. 2006. Metazoan Scc4 homologs link sister chromatid cohesion to cell and axon migration guidance. *PLoS Biol* 4:e242. PMID: 16802858

Zebrafish

- Horsfield JA, Anagnostou SH, Hu JK, Cho KH, Geisler R, Lieschke G, Crosier KE, Crosier PS. 2007. Cohesin-dependent regulation of Runx genes. *Development* 134:2639-49. PMID: 17567667
- Mönnich M, Banks S, Eccles M, Dickinson E, Horsfield J. 2009. Expression of cohesin and condensin genes during zebrafish development supports a non-proliferative role for cohesin. *Gene Expr Patterns*. Aug 31. [Epub ahead of print] PMID: 19723591
- Muto A, Calof AL, Lander AD, and Schilling, T.F. (2011) Multifactorial origins of heart and gut defects in *nipbl*-deficient zebrafish, a model of Cornelia de Lange Syndrome. *PLoS Biology* Oct;9(10):e1001181. Epub 2011 Oct 25. ([comment: PLoS Biol. 2011 Oct;9\(10\):e1001180](#). PMID: 22011921)
- L-leucine partially rescues translational and developmental defects associated with zebrafish models of Cornelia de Lange syndrome. Xu B, Sowa N, Cardenas ME, et al. J

REACHING OUT. PROVIDING HELP. GIVING HOPE.

CdLS Foundation | 302 West Main Street, #100 | Avon, CT 06001 | www.CdLSusa.org



Hum Mol Genet. 2015 Mar 15;24(6):1540-55. doi: 10.1093/hmg/ddu565. Epub 2014 Nov 6. PMID: 25378554.

Mouse

- Zhang B, Jain S, Song H, Fu M, Heuckeroth RO, Erlich JM, Jay PY, Milbrandt J. 2007. Mice lacking sister chromatid cohesion protein PDS5B exhibit developmental abnormalities reminiscent of Cornelia de Lange syndrome. *Development* 134:3191-201. PMID: 17652350
- Parelho V, Hadjur S, Spivakov M, Leleu M, Sauer S, Gregson HC, Jarmuz A, Canzonetta C, Webster Z, Nesterova T, Cobb BS, Yokomori K, Dillon N, Aragon L, Fisher AG, Merkenschlager M. 2008. Cohesins functionally associate with CTCF on mammalian chromosome arms. *Cell* 132:422-33. PMID: 18237772
- Zhang B, Chang J, Fu M, Huang J, Kashyap R, Salavaggione E, Jain S, Shashikant K, Deardorff MA, Uzielli ML, Dorsett D, Beebe DC, Jay PY, Heuckeroth RO, Krantz I, Milbrandt J. 2009. Dosage effects of cohesin regulatory factor PDS5 on mammalian development: implications for cohesinopathies. *PLoS ONE* 4:e5232. PMID: 19412548
- Kawauchi S, Calof AL, Santos R, Lopez-Burks ME, Young CM, Hoang MP, Chua A, Lao T, Lechner MS, Daniel JA, Nussenzweig A, Kitzes L, Yokomori K, Hallgrímsson B, Lander AD. 2009. Multiple organ system defects and transcriptional dysregulation in the Nipbl(+/-) mouse, a model of Cornelia de Lange Syndrome. *PLoS Genet* 5:e1000650. PMID: 19763162
- Chien R, Zeng W, Kawauchi S, Santos R, Gregson HC, Schmiesing JA, Newkirk D, Kong X, Ball AR J, Calof AL, Lander AD, Groudine M & Yokomori K (2011) Cohesin mediates chromatin interactions that regulate mammalian b-globin expression. *J. Biol. Chem.* 286:DOI 10.1074. PMID: PMC3093862

Prenatal

- Westergaard JG, Chemnitz J, Teisner B, Poulsen HK, Ipsen L, Beck B, Grudzinskas JG. Pregnancy-associated plasma protein A: a possible marker in the classification and prenatal diagnosis of Cornelia de Lange syndrome. *Prenat Diagn* 3:225-32. PMID: 6194522
- Bruner JP, Hsia YE. 1990. Prenatal findings in Brachmann-de Lange syndrome. *Obstet Gynecol* 76:966-8. PMID: 1699187



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.

- Drolshagen LF, Durmon G, Berumen M, Burks DD. 1992. Prenatal ultrasonographic appearance of "Cornelia de Lange" syndrome. *J Clin Ultrasound* 20:470-4. PMID: 1324952
- Kliever MA, Kahler SG, Hertzberg BS, Bowie JD. 1993. Fetal biometry in the Brachmann-de Lange syndrome. *Am J Med Genet* 47:1035-41. PMID: 8291520
- Manouvrier S, Espinasse M, Vaast P, Boute O, Farre I, Dupont F, Puech F, Gosselin B, Farriaux JP. 1996. Brachmann-de Lange syndrome: pre- and postnatal findings. *Am J Med Genet* 62:268-73. Ackerman J, Gilbert-Barness E. 1997. Brachmann-de Lange syndrome. *Am J Med Genet* 68:367-8. PMID: 8882785
- Ranzini AC, Day-Salvatore D, Farren-Chavez D, McLean DA, Greco R. 1997. Prenatal diagnosis of de Lange syndrome. *J Ultrasound Med* 16:755-8. PMID: 9360240
- Enns GM, Cox VA, Goldstein RB, Gibbs DL, Harrison MR, Golabi M. 1998. Congenital diaphragmatic defects and associated syndromes, malformations, and chromosome anomalies: a retrospective study of 60 patients and literature review. *Am J Med Genet* 79:215-25. PMID: 9788565
- Aitken DA, Ireland M, Berry E, Crossley JA, Macri JN, Burn J, Connor JM. 1999. Second-trimester pregnancy associated plasma protein-A levels are reduced in Cornelia de Lange syndrome pregnancies. *Prenat Diagn* 19:706-10. PMID: 10451512
- Boog G, Sagot F, Winer N, David A, Nomballais MF. 1999. Brachmann-de Lange syndrome: a cause of early symmetric fetal growth delay. *Eur J Obstet Gynecol Reprod Biol* 85:173-7. PMID: 10584631
- Sekimoto H, Osada H, Kimura H, Kamiyama M, Arai K, Sekiya S. 2000. Prenatal findings in Brachmann-de Lange syndrome. *Arch Gynecol Obstet* 263:182-4. PMID: 10834327
- Urban M, Hartung J. 2001. Ultrasonographic and clinical appearance of a 22-week-old fetus with Brachmann-de Lange syndrome. *Am J Med Genet* 102:73-5. PMID: 11471176
- Huang WH, Porto M. 2002. Abnormal first-trimester fetal nuchal translucency and Cornelia De Lange syndrome. *Obstet Gynecol* 99:956-8. PMID: 11975974

REACHING OUT. PROVIDING HELP. GIVING HOPE.

CdLS Foundation | 302 West Main Street, #100 | Avon, CT 06001 | www.CdLSusa.org



- Lee W, McNie B, Chaiworapongsa T, Conoscenti G, Kalache KD, Vettraino IM, Romero R, Comstock CH. 2002. Three-dimensional ultrasonographic presentation of micrognathia. *J Ultrasound Med* 21:775-81. PMID: 12099566
- Marino T, Wheeler PG, Simpson LL, Craigo SD, Bianchi DW. 2002. Fetal diaphragmatic hernia and upper limb anomalies suggest Brachmann-de Lange syndrome. *Prenat Diagn* 22:144-7. PMID: 11857622
- Arbuzova S, Nikolenko M, Krantz D, Hallahan T, Macri J. 2003. Low first-trimester pregnancy-associated plasma protein-A and Cornelia de Lange syndrome. *Prenat Diagn* 23:864. PMID: 14558036
- Le Vaillant C, Quere MP, David A, Berlivet M, Boog G. 2004. Prenatal diagnosis of a 'minor' form of Brachmann-de Lange syndrome by three-dimensional sonography and three-dimensional computed tomography. *Fetal Diagn Ther*. 19:155-9. PMID: 14764961
- Lalatta F, Russo S, Gentilin B, Spaccini L, Boschetto C, Cavalleri F, Masciadri M, Gervasini C, Bentivegna A, Castronovo P, Larizza L. 2007. Prenatal/neonatal pathology in two cases of Cornelia de Lange syndrome harboring novel mutations of NIPBL. *Genet Med* 9:188-94. PMID: 17413424
- Chong K, Keating S, Hurst S, Summers A, Berger H, Seaward G, Martin N, Friedberg T, Chitayat D. 2009. Cornelia de Lange syndrome (CdLS): prenatal and autopsy findings. *Prenat Diagn* 29:489-94. PMID: 19242925
- Sepulveda W, Wong AE, Dezerega V. 2009. Brachmann-de Lange Syndrome: prenatal diagnosis with 2- and 3-dimensional sonography. *J Ultrasound Med* 28:401-4. PMID: 19244081
- Identification of a prenatal profile of Cornelia de Lange syndrome (CdLS): a review of 53 CdLS pregnancies. Clark DM, Sherer I, Deardorff MA. *Am J Med Genet A*. 2012 Aug;158A(8):1848-56. doi: 10.1002/ajmg.a.35410. Epub 2012 Jun 27. Review. PMID: 22740382

Reviews

- Dorsett D. 2004. Adherin: key to the cohesin ring and Cornelia de Lange syndrome. *Curr Biol* 14:R834-6. PMID: 15458660
- Strachan T. 2005. Cornelia de Lange Syndrome and the link between chromosomal function, DNA repair and developmental gene regulation. *Curr Opin Genet Dev* 15:258-64. PMID: 15917200
- Dorsett D. 2007. Roles of the sister chromatid cohesion apparatus in gene expression, development, and human syndromes. *Chromosoma* 116:1-13. PMID: 16819604



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.

- Dorsett D. 2008. Running rings around chromosomes to trim axons and target dendrites. *Dev Cell* 14:156-8. PMID: 18267084
- Gause M, Schaaf CA, Dorsett D. 2008. Cohesin and CTCF: cooperating to control chromosome conformation? *Bioessays* 30:715-8. PMID: 18623068
- Liu J, Krantz ID. 2008. Cohesin and human disease. *Annu Rev Genomics Hum Genet* 9:303-20. PMID: 18767966
- McNairn AJ, Gerton JL. 2008. Cohesinopathies: One ring, many obligations. *Mutat Res* 647:103-11. PMID: 18786550
- Onn I, Heidinger-Pauli JM, Guacci V, Unal E, Koshland DE. 2008. Sister chromatid cohesion: a simple concept with a complex reality. *Annu Rev Cell Dev Biol* 24:105-29. PMID: 18616427
- Peters JM, Tedeschi A, Schmitz J. 2008. The cohesin complex and its roles in chromosome biology. *Genes Dev* 22:3089-114. PMID: 19056890
- Dorsett D, Krantz ID. 2009. On the molecular etiology of Cornelia de Lange syndrome. *Ann N Y Acad Sci* 1151:22-37. PMID: 19154515
- Dorsett D. 2009. Cohesin, gene expression and development: lessons from *Drosophila*. *Chromosome Res* 17:185-200. PMID: 19308700
- Wendt KS, Peters JM. 2009. How cohesin and CTCF cooperate in regulating gene expression. *Chromosome Res* 17:201-14. PMID: 19308701
- Mannini L, Liu J, Krantz ID, Musio A. 2009. Spectrum and consequences of SMC1A mutations: The unexpected involvement of a core component of cohesin in human disease. *Hum Mutat*. Sep 23. [Epub ahead of print] PMID: 19842212
- Liu J, Krantz ID. 2009. Cornelia de Lange syndrome, cohesin, and beyond. *Clin Genet* 76:303-14. PMID: 19793304

Scientific Symposia

- Kline AD et al. 2007. Scientific Abstract Submissions presented at the Second National Scientific Symposium in conjunction with the Cornelia de Lange Syndrome Foundation

REACHING OUT. PROVIDING HELP. GIVING HOPE.

CdLS Foundation | 302 West Main Street, #100 | Avon, CT 06001 | www.CdLSusa.org



25th National Meeting. Am J Med Genet 143A:1297-1305.

<http://www3.interscience.wiley.com/cgi-bin/fulltext/114219440/PDFSTART>

- Kline AD. 2009. 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. Am J Med Genet A 149A:1615-1622.
<http://www3.interscience.wiley.com/cgi-bin/fulltext/122513511/PDFSTART>
- Clinical, developmental and molecular update on Cornelia de Lange syndrome and the cohesin complex: Abstracts from the 2014 Scientific and Educational Symposium. Kline AD, Calof AL, Lander AD, Gerton JL, et al. Am J Med Genet A. 2015 Jun;167(6):1179-92. doi: 10.1002/ajmg.a.37056. Epub 2015 Apr 21. PMID: 25899772
-