



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
- Wygnanski-Jaffe T, Shin J, Perruzza E, Abdoell 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