



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
- 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
- Kliewer 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
- Lee W, McNie B, Chaiworapongsa T, Conoscenti G, Kalache KD, Vetraino 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



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Aug;158A(8):1848-56. doi: 10.1002/ajmg.a.35410. Epub 2012 Jun 27. Review. PMID: 22740382

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