

10. 文 献

- 1. Gaffney G, Manning N, Boyd PA, et al: Prenatal sonographic diagnosis of skeletal dysplasias: a report of the diagnostic and prognostic accuracy in 35 cases. *Prenat Diagn* 1998;18:357-362
- 2. Connor JM, Connor RA, Sweet EM, et al: Lethal neonatal chondrodysplasia in the West of Scotland 1970-1983 with a description of a thanatophoric, dysplasialike, autosomal recessive disorder, Glasgow variant. *Am J Med Genet* 1985;22:243-253
- 3. Al Gazali LI, Bakir M, Hamid Z, et al: Birth prevalence and pattern of osteochondrodysplasias in an inbred high risk population. *Birth Defects Part A Clin Mol Teratol* 2003;67:125-132
- 4. Camera G, Mastroiacov P: Birth prevalence of skeletal dysplasias in the Italian multicentric monitoring system for birth defects. In: Papadatos CJ, Bartosocas CS eds. *Skeletal Dysplasias*, Alan R. Liss, New York, 1982, p441-444
- 5. Superti-Furga A, Unger S: Nosology and classification of genetic skeletal disorders: 2006 revision. *Am J Med Genet A* 2007;143:1-18
- 6. 西村玄 :骨系統疾患の分類と画像診断 . 小児科診療 2006;69:1271-1278
- 7. Spranger J, Maroteaux P: The lethal osteochondrodysplasias. *Adv Hum Genet* 1990;19:1-103
- 8. Graf R: The diagnosis of congenital hip-joint dislocation by the ultrasound compound treatment. *Arch Orthop Trauma Surg* 1980;97:117-133
- 9. Filly RA, Golbus MS, Carey JC, et al: Short-limbed dwarfism: ultrasonographic diagnosis by mensuration of fetal femoral length. *Radiology* 1981;138:653-656
- 10. Kurtz AB, Needleman L, Wapner RJ, et al: Usefulness of a short femur in the in utero detection of skeletal dysplasias. *Radiology* 1990;177:197-200
- 11. Merz E, Kim-Kern MS, Pehl S: Ultrasonic mensuration of fetal limb bones in the second and third trimesters. *J Clin Ultrasound* 1987;15:175-183
- 12. Romero R, Athanasiadis AP, Jeanty P: Fetal skeletal anomalies. *Radiol Clin North Am* 1990;28:75-99
- 13. Alanay Y, Krakow D, Rimoin DL, et al: Angulated femurs and the skeletal dysplasias: experience of the International Skeletal Dysplasia Registry (1988-2006). *Am J Med Genet Part A* 2007;143A:1159-1168
- 14. 澤井英明 : Personal communication
- 15. Pazzaglia UE, Beluffi G: Radiology and histopathology of the bent limbs in campomelic dysplasia: implication in the aetiology of the disease and review of theories. *Pediatr Radiol* 1987;17:50-55
- 16. Brasseur-Daudruy M, Ickowicz V, Degre S, et al: Hyperechoic metaphyses in hypophosphatasia: what does it mean? *Pediatr Radiol* 2008;38:340-343

- 17. Spirt BA, Oliphant M, Gottlieb RH, et al: Prenatal sonographic evaluation of short-limbed dwarfism: an algorithmic approach. Radiographics 1990;10:217-236
- 18. Meizner I: Fetal skeletal malformations revisited: steps in the diagnostic approach. Ultrasound Obstet Gynecol 1997;10:303-306
- 19. Pattarelli P, Pretorius DH, Edwards DK: Intrauterine growth restriction mimicking skeletal dysplasia on antenatal sonography. J Ultrasound Med 1990;9:737-739
- 20. Hernandez J, Salas S, Pastor I, et al: Chondrodysplasia punctata associated with Down's syndrome. Presentation of a case and review of the literature. An Esp Pediatr 1988;28:145-147
- 21. Kurtz AB, Filly RA, Wapner RJ, et al: In utero analysis of heterozygous achondroplasia: variable time of onset as detected by femur length measurements. J Ultrasound Med 1986;5:137-140
- 22. O ' Rahilly R: Morphologic patterns in limb deficiencies and duplication. Am J Anat 1951;89 :135-193
- 23. Wessels MW, Den Hollander NS, Cohen-Overbeek TE, et al: Prenatal diagnosis and confirmation of the acrofacial dysostosis syndrome type Rodriguez. Am J Med Genet 2002;113:97 -100
- 24. Stoll C, Langer B, Gasser B, et al: Sporadic case of dyssegmental dysplasia with antenatal presentation. Genet Couns 1998;9:125-30
- 25. Guzman ER, Day-Salvatore D, Westover T, et al; Prenatal ultrasonographic demonstration of the trident hand in heterozygous achondroplasia. J Ultrasound Med 1994;13:63-66
- 26. 室月淳 , 荒木慶彦 , 山下徹 , 他 : Achondrogenesis type II の 1 例 . 山形県病医誌 1988;22 :101-104
- 27. Barnes AM, Chang W, Morello R, et al: Deficiency of cartilage-associated protein in recessive lethal osteogenesis imperfecta. N Engl J Med 2006;355:2757-2764
- 28. Cabral WA, Chang W, Barnes AM, et al: Prolyl 3-hydroxylase 1 deficiency causes a recessive metabolic bone disorder resembling lethal/severe osteogenesis imperfecta. Nat Genet 2007;39:359 -365
- 29. Sillence DO, Senn A, Danks DM: Genetic heterogeneity in osteogenesis imperfecta. J Med Genet 1979;16:101-116
- 30. Senanayake H, Anandakumar C, de Silva MV. : Mid-trimester fracture of femur in a normal fetus. J Obstet Gynecol Res 2003;29:186-188
- 31. Angels E, Wellnitz CV, Goodsitt MM, et al: Radiation dose to the fetus for pregnant patients undergoing multidetector CT image: Monte Carlo simulations estimating fetal dose for a range of gestational age and patient size. Radiologoy 2008;249:220-227
- 32. Johnson A, Callan NA, Bhutani VK, et al: Ultrasonic ratio of fetal thoracic to abdominal circumference: an association with fetal pulmonary hypoplasia. Am J Obstet Gynecol 1987;157:764 -769
- 33. Rahemtullah A, McGillivray B, Wilson RD: Suspected skeletal dysplasia: femur length to abdominal circumference ratio can be used in ultrasonographic prediction of fetal outcome. Am J Obstet Gynecol 1997;177:864-869

- 34. 加部一彦 , 横尾京子 , 野辺明子編 : 障害をもつ子を産むということ . 19人の体験 . 東京 , 中央法規出版 , 1999
 - 35. チャールズ・RK・ハインド : いかに " 深刻な診断 " を伝えるか . 誠実なインフォームド・コンセントのために . 岡安大仁監訳 . 高野和也訳 . 東京 , 人間と歴史社 , 2000 , p196
 - 36. 池川志郎 : Personal communication
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11. 胎児骨系統疾患に関する参考図書へ

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