

BÖLÜM



SIK GÖRÜLEN KEMİK DİSPLAZİLERİ

Yaşar TÜRK¹

Vaka 1: Akondroplazi

Vaka 2: Tanatoforik displazi

Vaka 3: Osteogenezis İmperfekta

Vaka 4: Osteopetrozis

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Osteopetrotik kemik kalitesi kemik iliği kavitesinin bozulması ve iatrojenik kırık riskini artırır. Bu potansiyel tuzakların önlenmesi, aralıklı periyotlarla soğuk normal salin irrigasyonu, düşük hızda delme, sık matkap ucu değişiklikleri ve çekiç kullanımından kaçınmayı gerektirir (24-26). Enfeksiyon ve yeniden yaranma riskinin daha yüksek olması nedeniyle 1 hafta ve 1, 3, 6, 9 ve 12. ayda yakın takip önerilmiştir (24).

Tedavi ve yaklaşım

İnfantil osteopetroz olumsuz sonuç nedeniyle tedavi gerektirir (27). D vitamini (kalsitriol) uyuyan osteoklastları uyurarak kemik rezorpsiyonuna yardımcı olur (28). Gama interferon ile tedavi uzun vadeli faydalar sağlamıştır. Kalsitriol ile kombinasyon tedavisi tek başına kalsitriolden daha üstündür. Eritropoietin anemiyi düzeltmek için kullanılabilir. Kortikosteroidler ayrıca anemiyi tedavi etmek ve kemik emilimini uyarmak için kullanılmıştır. Bazen kırıklar nedeniyle cerrahi tedavi gerekebilir.

KAYNAKÇA

1. Moreland LW. (2004) Rheumatology and immunology therapy, A to Z essentials. Springer Verlag. ISBN:3540206256.
2. Xue Y, Sun A, Mekikian PB, et al. FGFR3 mutation frequency in 324 cases from the International Skeletal Dysplasia Registry. *Mol Genet Genomic Med.* 2014;2(6):497-503. doi:10.1002/mgg3.96
3. Pauli RM, Legare JM. [(1998) Updated 2018 May 10]. Achondroplasia. In M.P Adam, H.H. Ardinger, R.A. Pagon, et al. (Eds), GeneReviews® (pp. 1993-2020) [Internet]. Seattle (WA): University of Washington, Seattle.
4. Dighe M, Fligner C, Cheng E, et al. Fetal skeletal dysplasia: an approach to diagnosis with illustrative cases *Radiographics.* 2008;28(4):1061-1077. doi:10.1148/rg.284075122
5. Miller E, Blaser S, Shannon P, et al. Brain and bone abnormalities of thanatophoric dwarfism. *AJR.* 2009;192(1):48-51. doi:10.2214/AJR.08.1524
6. Burrows PE, Stannard MW, Pearrow J, et al. Early antenatal sonographic recognition of thanatophoric dysplasia with cloverleaf skull deformity. *AJR.* 1984;143(4):841-843. doi:10.2214/ajr.143.4.841
7. Cunniff C; American Academy of Pediatrics Committee on Genetics. Prenatal screening and diagnosis for pediatricians. *Pediatrics.* 2004;114(3):889-894. doi:10.1542/peds.2004-1368
8. Blaas HG, Vogt C, Eik-Nes SH. Abnormal gyration of the temporal lobe and megalencephaly are typical features of thanatophoric dysplasia and can be visualized prenatally by ultrasound. *Ultrasound Obstet Gynecol.* 2012;40(2):230-234. doi:10.1002/uog.11127
9. Wang L, Takai Y, Baba K, et al. Can biparietal diameter-to-femur length ratio be a useful sonographic marker for screening thanatophoric dysplasia since the first trimester? A literature review of case reports and a retrospective study based on 10,293 routine fetal biometry measurements. *Taiwan J Obstet Gynecol.* 2017;56(3):374-378. doi:10.1016/j.tjog.2017.04.021
10. Ulla M, Aiello H, Cobos MP, et al. Prenatal diagnosis of skeletal dysplasias: contribution of three-dimensional computed tomography. *Fetal Diagn Ther.* 2011;29(3):238-247. doi:10.1159/000322212
11. MacDonald IM, Hunter AG, MacLeod PM, et al. Growth and development in thanatophoric dysplasia. *Am J Med Genet.* 1989;33(4):508-512. doi:10.1002/ajmg.1320330420
12. Minch CM, Kruse RW. Osteogenesis imperfecta: a review of basic science and diagnosis. *Orthopedics.* 1998;21(5):558-569.
13. Redon JY, Gloaguen D, Collet M, et al. L'ostéogénèse imparfaite. Réflexions sur le diagnostic prénatal (à propos de deux cas) [Osteogenesis imperfecta. Reflections after the prenatal diagnosis of 2 cases]. *J Gynecol Obstet Biol Reprod.* 1993;22(2):173-178.
14. Renaud A, Aucourt J, Weill J, et al. Radiographic features of osteogenesis imperfecta. *Insights Imaging.* 2013;4(4):417-429. doi:10.1007/s13244-013-0258-4

15. Grissom LE, Harcke HT. Radiographic features of bisphosphonate therapy in pediatric patients. *Pediatr Radiol.* 2003;33(4):226–229. doi:10.1007/s00247-003-0865-1
16. Goldman AB, Davidson D, Pavlov H, et al. “Popcorn” calcific cations: a prognostic sign in osteogenesis imperfecta. *Radiology.* 1980;136(2):351–358. doi:10.1148/radiology.136.2.7403509
17. Jones D, Hosalkar H, Jones S. The orthopaedic management of osteogenesis imperfecta. *Clin Orthop Relat Res.* 2002;16:374–88.
18. Zeitlin L, Fassier F, Glorieux FH. Modern approach to children with osteogenesis imperfecta. *J Pediatr Orthop B.* 2003;12(2):77–87. doi:10.1097/01.bpb.0000049567.52224.fa
19. Forin V. L’ostéogénèse imparfaite en pédiatrie: traitement médical et de rééducation [Paediatric osteogenesis imperfecta: medical and physical treatment]. *Arch Pediatr.* 2008;15(5):792–793. doi:10.1016/S0929-693X(08)71913-4
20. Matar HE, James LA. A challenging paediatric pathological femur fracture in pyknodysostosis (osteopetrosis acro-osteolytica): lessons learnt. *BMJ Case Rep.* 2014;2014:bcr2014207730. Published 2014 Nov 20. doi:10.1136/bcr-2014-207730
21. Gerritsen EJ, Vossen JM, van Loo IH, et al. Autosomal recessive osteopetrosis: variability of findings at diagnosis and during the natural course. *Pediatrics.* 1994;93(2):247–253.
22. Elster AD, Theros EG, Key LL, et al. Cranial imaging in autosomal recessive osteopetrosis. Part I. Facial bones and calvarium. *Radiology.* 1992;183(1):129–135. doi:10.1148/radiology.183.1.1549658
23. Silver IA, Murrills RJ, Etherington DJ. Microelectrode studies on the acid microenvironment beneath adherent macrophages and osteoclasts. *Exp Cell Res.* 1988;175(2):266–276. doi:10.1016/0014-4827(88)90191-7
24. Farfán MA, Olarte CM, Pesantez RF, et al. Recommendations for fracture management in patients with osteopetrosis: case report. *Arch Orthop Trauma Surg.* 2015;135(3):351–356. doi:10.1007/s00402-014-2144-z
25. Amit S, Shehkar A, Vivek M, et al. Fixation of Subtrochanteric Fractures in Two Patients with Osteopetrosis Using a Distal Femoral Locking Compression Plate of the Contralateral Side. *Eur J Trauma Emerg Surg.* 2010;36(3):263–269. doi:10.1007/s00068-009-8237-7
26. Bhargava A, Vagela M, Lennox CM. “Challenges in the management of fractures in osteopetrosis”! Review of literature and technical tips learned from long-term management of seven patients. *Injury.* 2009;40(11):1167–1171. doi:10.1016/j.injury.2009.02.009
27. Proceedings and abstracts of the First International Symposium on Osteopetrosis: biology and therapy. October 23–24, 2003. Bethesda, Maryland, USA. *J Bone Miner Res.* 2004;19(8):1356–1375. doi:10.1002/jbmr.5650190822
28. Key L, Carnes D, Cole S, et al. Treatment of congenital osteopetrosis with high-dose calcitriol. *N Engl J Med.* 1984;310(7):409–415. doi:10.1056/NEJM198402163100701