

Bölüm 4

KONJENİTAL KALP HASTALIĞI İÇİN YENİDOĞAN NABIZ OKSİMETRE TARAMASI

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Konjenital kalp hastalığı (KKH), yenidoğanlarda en sık görülen doğumsal hastalıktır (1,2). Yaşamın ilk yılında cerrahi veya kateterizasyonla müdahale gerektiren kritik KKH (KKKH), tüm KKH'ların yaklaşık %25'ini oluşturur (3). KKKH'lı birçok yenidoğan semptomatik olup doğumdan hemen sonra teşhis edilirken, bir kısmı da taburculuktan sonra tanı alır (4,5). Kritik kalp lezyonlu bebeklerde, teşhisin gecikmesi ve bu hastaların tedavisinde uzmanlık sahibi bir üçüncü basamak merkeze zamanında yönlendirilmemesi morbidite ve mortalite riskini arttırır (6,7).

KKKH'nın erken tespiti için yenidoğan döneminde kullanılan en güvenilir yöntem nabız oksimetre taramasıdır (NOT).

1. TANIM VE HEDEFLenen LEZYONLAR

KKKH: İlk yaşta cerrahi veya kateterizasyonla müdahale gerektiren doğumsal kalp malformasyonlarıdır. Bu kategoride, duktal bağımlı ve siyanotik lezyonların yanı sıra, patent duktus arteriyozusa (PDA) bağımlı olmayan daha hafif KKH formları da yer alır. KKKH, tüm KKH'ların yaklaşık %25'ini oluşturur.

NOT'da Hedeflenen Lezyonlar:

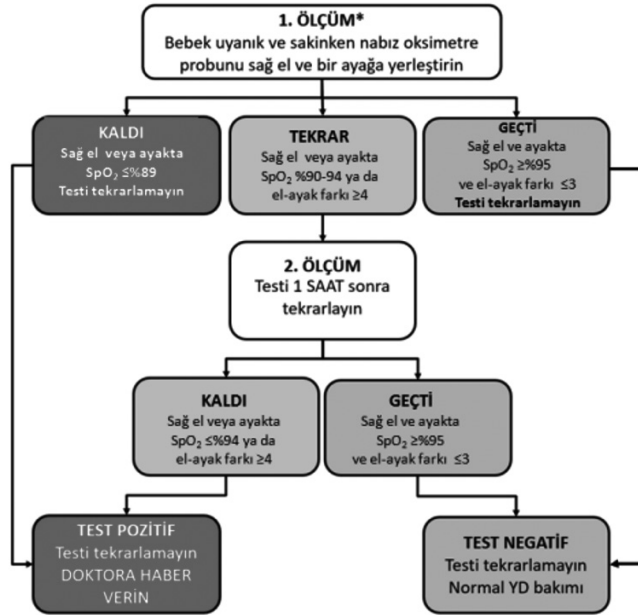
(A) İlk yaşta müdahale gerektiren lezyonlar

(B) Genellikle bir süreliğine veya sürekli hipoksemi ile seyreden defekler (8,9).

- Hipoplastik sol kalp sendromu
- Ventriküler septum bütünlüğü korunmuş pulmoner atrezi
- Ventriküler septal defektli pulmoner atrezi
- Fallot tetralojisi
- Total anormal pulmoner venöz dönüş
- Büyük arterlerin transpozisyonu

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- Sağlık hizmeti sağlayıcılarının ve ailelerin eğitimi: Personel ve aileler için eğitim materyalinin geliştirilmesi.
- Tarama sonucunun, diğer tarama testleri (örneğin; fetal ultrason), kardiyak olmayan durumlar, ekipman kalitesi, eğitim çabaları dahil tarama maliyeti ve geri ödeme bağlamında sürekli olarak değerlendirilmesi.
- KKKH taramasının uygulanması ülkeden ülkeye ve aynı ülke içinde farklı coğrafik bölgelerde değişiklik gösterir. Klinisyenler, uygulama alanları için uygun algoritma ve protokolleri belirlemek için yerel halk sağlığı kurumlarının yönergelerine başvurmaldırlar.



Şekil 1. T.C. Sağlık Bakanlığı Neonatal Kritik Doğumsal Kalp Hastalığı Tarama Şeması

Kaynak: Martin GR, et al. Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease. Pediatrics. 2020;146(1):e20191650

KAYNAKLAR

1. Tennant PW, Pearce MS, Bythell M, Rankin J. 20-year survival of children born with congenital anomalies: a population-based study. The Lancet. 2010 Feb;375(9715):649–56.
2. Bird TM, Hobbs CA, Cleves MA, Tilford JM, Robbins JM. National rates of birth defects among hospitalized newborns. Birth Defects Res A Clin Mol Teratol. 2006 Nov;76(11):762–9.

3. Oster ME, Lee KA, Honein MA, Riehle-Colarusso T, Shin M, Correa A. Temporal Trends in Survival Among Infants With Critical Congenital Heart Defects. *Pediatrics*. 2013 May 1;131(5):e1502-8.
4. Wren C, Reinhardt Z, Khawaja K. Twenty-year trends in diagnosis of life-threatening neonatal cardiovascular malformations. *Arch Dis Child Fetal Neonatal Ed*. 2008 Jan;93(1):F33-5.
5. Peterson C, Ailes E, Riehle-Colarusso T, Oster ME, Olney RS, Cassell CH, et al. Late Detection of Critical Congenital Heart Disease Among US Infants. *JAMA Pediatr*. 2014 Apr 1;168(4):361.
6. de-Wahl Granelli A, Wennergren M, Sandberg K, Mellander M, Bejлум C, Ingas L, et al. Impact of pulse oximetry screening on the detection of duct dependent congenital heart disease: a Swedish prospective screening study in 39 821 newborns. *BMJ*. 2009 Jan 8;338(jan08 2):a3037-a3037.
7. Eckersley L, Sadler L, Parry E, Finucane K, Gentles TL. Timing of diagnosis affects mortality in critical congenital heart disease. *Arch Dis Child*. 2016 Jun;101(6):516-20.
8. Mahle WT, Newburger JW, Matherne GP, Smith FC, Hoke TR, Koppel R, et al. Role of Pulse Oximetry in Examining Newborns for Congenital Heart Disease: A Scientific Statement from the AHA and AAP. *Pediatrics*. 2009 Aug 1;124(2):823-36.
9. Oster ME, Aucott SW, Glidewell J, Hackell J, Kochilas L, Martin GR, et al. Lessons Learned From Newborn Screening for Critical Congenital Heart Defects. *Pediatrics*. 2016 May 1;137(5).
10. Khoshnood B, Lelong N, Houyel L, Thieulin AC, Jouannic JM, Magnier S, et al. Prevalence, timing of diagnosis and mortality of newborns with congenital heart defects: a population-based study. *Heart*. 2012 Nov 15;98(22):1667-73.
11. Ailes EC, Gilboa SM, Honein MA, Oster ME. Estimated Number of Infants Detected and Missed by Critical Congenital Heart Defect Screening. *Pediatrics*. 2015 Jun 1;135(6):1000-8.
12. Lees MH. Cyanosis of the newborn infant. *J Pediatr*. 1970 Sep;77(3):484-98.
13. Schultz AH, Localio AR, Clark BJ, Ravishankar C, Videon N, Kimmel SE. Epidemiologic Features of the Presentation of Critical Congenital Heart Disease: Implications for Screening. *Pediatrics*. 2008 Apr 1;121(4):751-7.
14. Kuehl KS, Loffredo CA, Ferencz C. Failure to Diagnose Congenital Heart Disease in Infancy. *Pediatrics*. 1999 Apr 1;103(4):743-7.
15. Schultz AH, Localio AR, Clark BJ, Ravishankar C, Videon N, Kimmel SE. Epidemiologic Features of the Presentation of Critical Congenital Heart Disease: Implications for Screening. *Pediatrics*. 2008 Apr 1;121(4):751-7.
16. Chang RKR, Gurvitz M, Rodriguez S. Missed Diagnosis of Critical Congenital Heart Disease. *Arch Pediatr Adolesc Med*. 2008 Oct 6;162(10):969.
17. Peterson C, Dawson A, Grosse SD, Riehle-Colarusso T, Olney RS, Tanner JP, et al. Hospitalizations, costs, and mortality among infants with critical congenital heart disease: How important is timely detection? *Birth Defects Res A Clin Mol Teratol*. 2013 Oct;97(10):664-72.
18. Zhao Q ming, Ma X jing, Ge X ling, Liu F, Yan W li, Wu L, et al. Pulse oximetry with clinical assessment to screen for congenital heart disease in neonates in China: a prospective study. *The Lancet*. 2014 Aug;384(9945):747-54.

19. Garg LF, Van Naarden Braun K, Knapp MM, Anderson TM, Koppel RI, Hirsch D, et al. Results From the New Jersey Statewide Critical Congenital Heart Defects Screening Program. *Pediatrics*. 2013 Aug 1;132(2):e314–23.
20. Abouk R, Grosse SD, Ailes EC, Oster ME. Association of US State Implementation of Newborn Screening Policies for Critical Congenital Heart Disease With Early Infant Cardiac Deaths. *JAMA*. 2017 Dec 5;318(21):2111.
21. Sakai-Bizmark R, Kumamaru H, Marr EH, Bedel LEM, Mena LA, Baghaee A, et al. Pulse Oximetry Screening: Association of State Mandates with Emergency Hospitalizations. *Pediatr Cardiol*. 2023 Jan 23;44(1):67–74.
22. Singh Y, Chen SE. Impact of pulse oximetry screening to detect congenital heart defects: 5 years' experience in a UK regional neonatal unit. *Eur J Pediatr*. 2022 Feb 7;181(2):813–21.
23. Plana MN, Zamora J, Suresh G, Fernandez-Pineda L, Thangaratinam S, Ewer AK. Pulse oximetry screening for critical congenital heart defects. *Cochrane Database of Systematic Reviews*. 2018 Mar 1;2018(3).
24. Powell R, Pattison HM, Bhoyar A, Furnston AT, Middleton LJ, Daniels JP, et al. Pulse oximetry screening for congenital heart defects in newborn infants: an evaluation of acceptability to mothers. *Arch Dis Child Fetal Neonatal Ed*. 2013 Jan;98(1):F59–63.
25. Mahle WT, Martin GR, Beekman RH, Morrow WR, Rosenthal GL, Snyder CS, et al. Endorsement of Health and Human Services Recommendation for Pulse Oximetry Screening for Critical Congenital Heart Disease. *Pediatrics*. 2012 Jan 1;129(1):190–2.
26. Martin GR, Ewer AK, Gaviglio A, Hom LA, Saarinen A, Sontag M, et al. Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease. *Pediatrics*. 2020 Jul 1;146(1).
27. Mouledoux J, Guerra S, Ballweg J, Li Y, Walsh W. A novel, more efficient, staged approach for critical congenital heart disease screening. *Journal of Perinatology*. 2017 Mar 10;37(3):288–90.
28. Diller CL, Kelleman MS, Kupke KG, Quarry SC, Kochilas LK, Oster ME. A Modified Algorithm for Critical Congenital Heart Disease Screening Using Pulse Oximetry. *Pediatrics*. 2018 May 1;141(5).
29. Valmari P. Should pulse oximetry be used to screen for congenital heart disease? *Arch Dis Child Fetal Neonatal Ed*. 2007 May 1;92(3):F219–24.
30. Thangaratinam S, Daniels J, Ewer AK, Zamora J, Khan KS. Accuracy of pulse oximetry in screening for congenital heart disease in asymptomatic newborns: a systematic review. *Arch Dis Child Fetal Neonatal Ed*. 2007 May 1;92(3):F176–80.
31. Reich JD, Connolly B, Bradley G, Littman S, Koepfel W, Lewycky P, et al. Reliability of a Single Pulse Oximetry Reading as a Screening Test for Congenital Heart Disease in Otherwise Asymptomatic Newborn Infants: The Importance of Human Factors. *Pediatr Cardiol*. 2008 Mar 12;29(2):371–6.
32. Fouzas S, Priftis KN, Anthracopoulos MB. Pulse Oximetry in Pediatric Practice. *Pediatrics*. 2011 Oct 1;128(4):740–52.
33. Ewer AK, Middleton LJ, Furnston AT, Bhoyar A, Daniels JP, Thangaratinam S, et al. Pulse oximetry screening for congenital heart defects in newborn infants (PulseOx): a test accuracy study. *The Lancet*. 2011 Aug;378(9793):785–94.
34. Thangaratinam S, Brown K, Zamora J, Khan KS, Ewer AK. Pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies: a systematic review and meta-analysis. *The Lancet*. 2012 Jun;379(9835):2459–64.

35. Kochilas LK, Menk JS, Saarinen A, Gaviglio A, Lohr JL. A Comparison of Retesting Rates Using Alternative Testing Algorithms in the Pilot Implementation of Critical Congenital Heart Disease Screening in Minnesota. *Pediatr Cardiol.* 2015 Mar 11;36(3):550-4.
36. Hoke TR, Donohue PK, Bawa PK, Mitchell RD, Pathak A, Rowe PC, et al. Oxygen Saturation as a Screening Test for Critical Congenital Heart Disease: A Preliminary Study. *Pediatr Cardiol.* 2002 Jul 24;23(4):403-9.
37. Oster ME, Colarusso T, Glidewell J. Screening for Critical Congenital Heart Disease: A Matter of Sensitivity. *Pediatr Cardiol.* 2013 Jan 21;34(1):203-4.
38. Wright J, Kohn M, Niermeyer S, Rausch CM. Feasibility of Critical Congenital Heart Disease Newborn Screening at Moderate Altitude. *Pediatrics.* 2014 Mar 1;133(3):e561-9.
39. Paranka MS, Brown JM, White RD, Park M V., Kelleher AS, Clark RH. The impact of altitude on screening for critical congenital heart disease. *Journal of Perinatology.* 2018 May 29;38(5):530-6.
40. Han LM, Klewer SE, Blank KM, Seckeler MD, Barber BJ. Feasibility of Pulse Oximetry Screening for Critical Congenital Heart Disease at 2643-Foot Elevation. *Pediatr Cardiol.* 2013 Dec 16;34(8):1803-7.
41. Hoffman JIE. Is Pulse Oximetry Useful for Screening Neonates for Critical Congenital Heart Disease at High Altitudes? *Pediatr Cardiol.* 2016 Jun 18;37(5):812-7.
42. Cawsey MJ, Noble S, Cross-Sudworth F, Ewer AK. Feasibility of pulse oximetry screening for critical congenital heart defects in homebirths. *Arch Dis Child Fetal Neonatal Ed.* 2016 Jul;101(4):F349-51.
43. Evers PD, Vernon MM, Schultz AH. Critical Congenital Heart Disease Screening Practices Among Licensed Midwives in Washington State. *J Midwifery Womens Health.* 2015 Mar 17;60(2):206-10.
44. Manja V, Mathew B, Carrion V, Lakshminrusimha S. Critical congenital heart disease screening by pulse oximetry in a neonatal intensive care unit. *Journal of Perinatology.* 2015 Jan 24;35(1):67-71.
45. Van Naarden Braun K, Grazel R, Koppel R, Lakshminrusimha S, Lohr J, Kumar P, et al. Evaluation of critical congenital heart defects screening using pulse oximetry in the neonatal intensive care unit. *Journal of Perinatology.* 2017 Oct 27;37(10):1117-23.
46. Murphy D, Pak Y, Cleary JP. Pulse Oximetry Overestimates Oxyhemoglobin in Neonates with Critical Congenital Heart Disease. *Neonatology.* 2016;109(3):213-8.
47. Goetz EM, Magnuson KM, Eickhoff JC, Porte MA, Hokanson JS. Pulse oximetry screening for critical congenital heart disease in the neonatal intensive care unit. *Journal of Perinatology.* 2016 Jan 5;36(1):52-6.
48. Kemper AR, Mahle WT, Martin GR, Cooley WC, Kumar P, Morrow WR, et al. Strategies for Implementing Screening for Critical Congenital Heart Disease. *Pediatrics.* 2011 Nov 1;128(5):e1259-67.
49. Peterson C, Grosse SD, Oster ME, Olney RS, Cassell CH. Cost-Effectiveness of Routine Screening for Critical Congenital Heart Disease in US Newborns. *Pediatrics.* 2013 Sep 1;132(3):e595-603.
50. Kochilas LK, Lohr JL, Bruhn E, Borman-Shoap E, Gams BL, Pylipow M, et al. Implementation of Critical Congenital Heart Disease Screening in Minnesota. *Pediatrics.* 2013 Sep 1;132(3):e587-94.

51. Roberts TE, Barton PM, Auguste PE, Middleton LJ, Furmston AT, Ewer AK. Pulse oximetry as a screening test for congenital heart defects in newborn infants: a cost-effectiveness analysis. *Arch Dis Child*. 2012 Mar;97(3):221–6.
52. Grosse S, Peterson C, Abouk R, Glidewell J, Oster M. Cost and Cost-Effectiveness Assessments of Newborn Screening for Critical Congenital Heart Disease Using Pulse Oximetry: A Review. *Int J Neonatal Screen*. 2017 Dec 14;3(4):34.
53. Reeder MR, Kim J, Nance A, Krikov S, Feldkamp ML, Randall H, et al. Evaluating cost and resource use associated with pulse oximetry screening for critical congenital heart disease: Empiric estimates and sources of variation. *Birth Defects Res A Clin Mol Teratol*. 2015 Nov;103(11):962–71.
54. Glidewell J, Grosse SD, Riehle-Colarusso T, Pinto N, Hudson J, Daskalov R, et al. Actions in Support of Newborn Screening for Critical Congenital Heart Disease — United States, 2011–2018. *MMWR Morb Mortal Wkly Rep*. 2019 Feb 8;68(5):107–11.
55. Manzoni P, Martin GR, Sanchez Luna M, Mestrovic J, Simeoni U, Zimmermann L, et al. Pulse oximetry screening for critical congenital heart defects: a European consensus statement. *Lancet Child Adolesc Health*. 2017 Oct;1(2):88–90.
56. Narayan IC, Blom NA, Ewer AK, Vento M, Manzoni P, te Pas AB. Aspects of pulse oximetry screening for critical congenital heart defects: when, how and why? *Arch Dis Child Fetal Neonatal Ed*. 2016 Mar;101(2):F162–7.
57. T.C. Sağlık Bakanlığı Halk Sağlığı Genel Müdürlüğü Neonatal Kritik Doğumsal Kalp Hastalıkları Tarama Rehberi, Sağlık Bakanlığı Yayın No: 1211, 1. Baskı Ankara, 2021 (ISBN: 978-975-590-827-4)