



DAFTAR PUSTAKA

- Abou Hassan, O. K., Fahed, A. C., Batrawi, M., Arabi, M., Refaat, M. M., DePalma, S. R., et al. 2015. NKX2-5 mutations in an inbred consanguineous population: genetic and phenotypic diversity. *Sci Rep*, 5, 8848.
- Akazawa, H. dan Komuro, I. 2005. Cardiac transcription factor Csx/Nkx2-5: Its role in cardiac development and diseases. *Pharmacol Ther*, 107, 252-68.
- Andersen, T. A., Troelsen Kde, L. dan Larsen, L. A. 2014. Of mice and men: molecular genetics of congenital heart disease. *Cell Mol Life Sci*, 71, 1327-52.
- Azhari, N., Shihata, M. S. dan Al-Fatani, A. 2004. Spontaneous closure of atrial septal defects within the oval fossa. *Cardiol Young*, 14, 148-55.
- Bartlett, H., Veenstra, G. J. dan Weeks, D. L. 2010. Examining the cardiac NK-2 genes in early heart development. *Pediatr Cardiol*, 31, 335-41.
- Baumgartner, H., De Backer, J., Babu-Narayan, S. V., Budts, W., Chessa, M., Diller, G. P., et al. 2021. 2020 ESC Guidelines for the management of adult congenital heart disease. *Eur Heart J*, 42, 563-645.
- Behiry, E. G., Al-Azzouny, M. A., Sabry, D., Behairy, O. G. dan Salem, N. E. 2019. Association of NKX2-5, GATA4, and TBX5 polymorphisms with congenital heart disease in Egyptian children. *Mol Genet Genomic Med*, 7, e612.
- Benson, D. W., Silberbach, G. M., Kavanaugh-McHugh, A., Cottrill, C., Zhang, Y., Riggs, S., et al. 1999. Mutations in the cardiac transcription factor NKX2.5 affect diverse cardiac developmental pathways. *J Clin Invest*, 104, 1567-73.
- Biesecker, L. G. 2020. Clinical Genomics- Genome Structure and Variation. *Goldman-Cecil Medicine*, 2-Volume Set. Twenty-Six ed.: Elsevier Inc.
- Blue, G. M., Kirk, E. P., Giannoulatou, E., Sholler, G. F., Dunwoodie, S. L., Harvey, R. P., et al. 2017. Advances in the Genetics of Congenital Heart Disease: A Clinician's Guide. *J Am Coll Cardiol*, 69, 859-870.
- Botto, L. D., Correa, A. dan Erickson, J. D. 2001. Racial and temporal variations in the prevalence of heart defects. *Pediatrics*, 107, E32.
- Bradley, E. A. dan Zaidi, A. N. 2020. Atrial Septal Defect. *Cardiol Clin*, 38, 317-324.
- Brickner, M. E., Hillis, L. D. dan Lange, R. A. 2000. Congenital heart disease in adults. First of two parts. *N Engl J Med*, 342, 256-63.
- Bruneau, B. G. 2008. The developmental genetics of congenital heart disease. *Nature*, 451, 943-8.
- Buckingham, M., Meilhac, S. dan Zaffran, S. 2005. Building the mammalian heart from two sources of myocardial cells. *Nat Rev Genet*, 6, 826-35.
- Cao, Y., Wang, J., Wei, C., Hou, Z., Li, Y., Zou, H., et al. 2016. Genetic variations of NKX2-5 in sporadic atrial septal defect and ventricular septal defect in Chinese Yunnan population. *Gene*, 575, 29-33.
- Caputo, S., Capozzi, G., Russo, M. G., Esposito, T., Martina, L., Cardaropoli, D., et al. 2005. Familial recurrence of congenital heart disease in patients with ostium secundum atrial septal defect. *Eur Heart J*, 26, 2179-84.



- Chen, L. T., Yang, T. B., Wang, T. T., Zheng, Z., Zhao, L. J., Ye, Z. W., *et al.* 2018. [Association of single nucleotide polymorphisms of transcription factors with congenital heart diseases in the Chinese population: a Meta analysis]. *Zhongguo Dang Dai Er Ke Za Zhi*, 20, 490-496.
- Chung, I. M. dan Rajakumar, G. 2016. Genetics of Congenital Heart Defects: The NKX2-5 Gene, a Key Player. *Genes (Basel)*, 7.
- Cross, S. 2013. *Underwood's Pathology*, Elsevier Health Sciences.
- Dahlan, S. 2019. *Besar Sampel dalam Penelitian Kedokteran dan Kesehatan*, Jakarta, Epidemiologi Indonesia.
- Destefano, G. M. dan Christiano, A. M. 2021. 54 - Basic Principles of Genetics. *Dermatology: 2-Volume Set*. Fourth Edition ed.: Elsevier Ltd.
- Dinarti, L. K., Hartopo, A. B., Kusuma, A. D., Satwiko, M. G., Hadwiono, M. R., Pradana, A. D., *et al.* 2020. The COngenital HeART Disease in adult and Pulmonary Hypertension (COHARD-PH) registry: a descriptive study from single-center hospital registry of adult congenital heart disease and pulmonary hypertension in Indonesia. *BMC Cardiovasc Disord*, 20, 163.
- Ebert, D. H., Finn, C. T. dan Scd, J. W. S. 2020. Genetics and Psychiatry. *Massachusetts General Hospital Comprehensive Clinical Psychiatry*, 677-701.e3.
- Ebert, D. H., Finn, C. T., Stoler, J. M. dan Smoller, J. W. 2010. Genetics and psychiatry. *Massachusetts General Hospital Handbook of General Hospital Psychiatry*, 6th edn. Elsevier, Inc.: Philadelphia, PA.
- Edwards, J. J. dan Gelb, B. D. 2016. Genetics of congenital heart disease. *Curr Opin Cardiol*, 31, 235-41.
- Ejim, E. C., Anisiuba, B. C., Ike, S. O. dan Essien, I. O. 2011. Atrial septal defects presenting initially in adulthood: patterns of clinical presentation in enugu, South-East Nigeria. *J Trop Med*, 2011, 251913.
- El Bouchikhi, I., Bouguenouch, L., Zohra Moufid, F., Houssaini, M. I., Belhassan, K., Samri, I., *et al.* 2017. NKX2-5 molecular screening and assessment of variant rate and risk factors of secundum atrial septal defect in a Moroccan population. *Anatol J Cardiol*, 17, 217-223.
- Ellesoe, S. G., Johansen, M. M., Bjerre, J. V., Hjortdal, V. E., Brunak, S. dan Larsen, L. A. 2016. Familial Atrial Septal Defect and Sudden Cardiac Death: Identification of a Novel NKX2-5 Mutation and a Review of the Literature. *Congenit Heart Dis*, 11, 283-90.
- Erickson, R. P. dan Wynshaw-Boris, A. J. 2016. Epstein's Inborn Errors of Development: The Molecular Basis of Clinical Disorders of Morphogenesis.
- Evans, S. M. 1999. Vertebrate tinman homologues and cardiac differentiation. *Semin Cell Dev Biol*, 10, 73-83.
- Fagerberg, L., Hallstrom, B. M., Oksvold, P., Kampf, C., Djureinovic, D., Odeberg, J., *et al.* 2014. Analysis of the human tissue-specific expression by genome-wide integration of transcriptomics and antibody-based proteomics. *Mol Cell Proteomics*, 13, 397-406.
- Fahed, A. C., Gelb, B. D., Seidman, J. G. dan Seidman, C. E. 2013. Genetics of congenital heart disease: the glass half empty. *Circ Res*, 112, 707-20.



- Ford, S. M., Watanabe, M. dan Wikenheiser, J. 2015. Cardiac embryology. *Fanaroff & Martin's Neonatal-Perinatal Medicine: Diseases of the Fetus and Infant. 10th ed. Philadelphia, PA: Elsevier Saunders.*
- Gelernter-Yaniv, L. dan Lorber, A. 2007. The familial form of atrial septal defect. *Acta Paediatr*, 96, 726-30.
- Geva, T., Martins, J. D. dan Wald, R. M. 2014. Atrial septal defects. *Lancet*, 383, 1921-32.
- González-Castro, T. B., Tovilla-Zárate, C. A., López-Narvaez, M. L., Juárez-Rojop, I. E., Calderón-Colmenero, J., Sandoval, J. P., et al. 2020. Association between congenital heart disease and NKX2. 5 gene polymorphisms: systematic review and meta-analysis. *Biomarkers in Medicine*, 14, 1747-1757.
- Gunal, N., Gul, S. dan Kahramanyol, O. 1997. Familial atrial septal defect with prolonged atrioventricular conduction. *Acta Paediatr Jpn*, 39, 634-6.
- Herlong, J. R. 2020. 62 - Introductory Embryology. *Caffey's Pediatric Diagnostic Imaging, 2-Volume Set*. Thirteenth ed.: Elsevier Inc.
- Hosoda, T., Komuro, I., Shiojima, I., Hiroi, Y., Harada, M., Murakawa, Y., et al. 1999. Familial atrial septal defect and atrioventricular conduction disturbance associated with a point mutation in the cardiac homeobox gene CSX/NKX2-5 in a Japanese patient. *Jpn Circ J*, 63, 425-6.
- Ismail, M. T., Hidayati, F., Krisdinarti, L., Noormanto, N., Nugroho, S. dan Wahab, A. S. 2015. Epidemiological profile of congenital heart disease in a national referral hospital. *ACI (Acta Cardiologia Indonesiana)*, 1.
- Juan, W., Xingyuan, L. dan Yiqing, Y. 2011. Novel NKX2-5 mutations responsible for congenital heart disease. *Heart*, 97, A205-A205.
- Kalayinia, S., Biglari, A., Rokni-Zadeh, H., Mahdavi, M., Rabbani, B., Maleki, M., et al. 2018. The Nkx2-5 Gene Mutations Related to Congenital Heart Diseases in Iranian Patients Population. *International Cardiovascular Research Journal*, 12.
- Kalayinia, S., Ghasemi, S. dan Mahdieh, N. 2019. A comprehensive in silico analysis, distribution and frequency of human Nkx2-5 mutations; A critical gene in congenital heart disease. *J Cardiovasc Thorac Res*, 11, 287-299.
- Kasahara, H. dan Benson, D. W. 2004. Biochemical analyses of eight NKX2.5 homeodomain missense mutations causing atrioventricular block and cardiac anomalies. *Cardiovasc Res*, 64, 40-51.
- Kendler, K. S. 1987. Sporadic vs familial classification given etiologic heterogeneity: I. Sensitivity, specificity, and positive and negative predictive value. *Genet Epidemiol*, 4, 313-30.
- Kennelly, P. J. dan Rodwell, V. W. 2009. Asam Amino dan Peptida. In: MURRAY, R. K., GRANNER, D. K. & RODWELL, V. W. (eds.) *Biokimia Harper*. 27 ed. Jakarta: Penerbit Buku Kedokteran EGC.
- Khatami, M., Mazidi, M., Taher, S., Heidari, M. M. dan Hadadzadeh, M. 2018. Novel Point Mutations in the NKX2.5 Gene in Pediatric Patients with Non-Familial Congenital Heart Disease. *Medicina (Kaunas)*, 54.
- Kodo, K. dan Yamagishi, H. 2017. Current insights into genetics of congenital heart diseases: GATA and T-box cardiac transcription factors as the hotspot



- pathogenesis. *Journal of pediatric cardiology and cardiac surgery*, 1, 18-27.
- Krasuski, R. A. 2019. Congenital Heart Disease in the Adult. *Current Clinical Medicine*, 2010, 212-222.e1.
- Lindsey, J. B. dan Hillis, L. D. 2007. Clinical update: atrial septal defect in adults. *The Lancet*, 369, 1244-1246.
- Luna-Zurita, L., Stirnimann, C. U., Glatt, S., Kaynak, B. L., Thomas, S., Baudin, F., et al. 2016. Complex Interdependence Regulates Heterotypic Transcription Factor Distribution and Coordinates Cardiogenesis. *Cell*, 164, 999-1014.
- Marelli, A. J. 2012. Congenital Heart Disease in Adults. In: GOLDMAN, L. & SCHAFER, A. I. (eds.) *GOLDMAN'S CECIL MEDICINE*. Philadelphia: Elsevier Inc.
- Marelli, A. J., Ionescu-Ittu, R., Mackie, A. S., Guo, L., Dendukuri, N. dan Kaouache, M. 2014. Lifetime prevalence of congenital heart disease in the general population from 2000 to 2010. *Circulation*, 130, 749-56.
- Marín-García, J. 2011. Post-genomic cardiology.
- Mark, M., Rijli, F. M. dan Chambon, P. 1997. Homeobox genes in embryogenesis and pathogenesis. *Pediatr Res*, 42, 421-9.
- McMahon, C. J., Feltes, T. F., Fraley, J. K., Bricker, J. T., Grifka, R. G., Tortoriello, T. A., et al. 2002. Natural history of growth of secundum atrial septal defects and implications for transcatheter closure. *Heart*, 87, 256-9.
- Moorman, A., Webb, S., Brown, N. A., Lamers, W. dan Anderson, R. H. 2003. Development of the heart: (1) formation of the cardiac chambers and arterial trunks. *Heart*, 89, 806-14.
- Mozaffarian, D., Benjamin, E. J., Go, A. S., Arnett, D. K., Blaha, M. J., Cushman, M., et al. 2015. Heart disease and stroke statistics--2015 update: a report from the American Heart Association. *Circulation*, 131, e29-322.
- Musunuru, K. dan Kathiresan, S. 2016. 8 - Principles of Cardiovascular Genetics. *Braunwald's Heart Disease*, 10/e, 64-74.
- Nakashima, Y., Ono, K., Yoshida, Y., Kojima, Y., Kita, T., Tanaka, M., et al. 2009. The search for Nkx2-5-regulated genes using purified embryonic stem cell-derived cardiomyocytes with Nkx2-5 gene targeting. *Biochemical and Biophysical Research Communications*, 390, 821-826.
- Nikiforova, M. N. dan Nikiforov, Y. E. 2011. Chapter 2 - Molecular Anatomic Pathology: Principles, Techniques, and Application to Immunohistologic Diagnosis. In: DABBS, D. J. (ed.) *Diagnostic Immunohistochemistry (Third Edition)*. Philadelphia: W.B. Saunders.
- Nora, J. J. dan Meyer, T. C. 1966. Familial nature of congenital heart diseases. *Pediatrics*, 37, 329-34.
- Nurulloh, M. I., Trimarsanto, H., Anggraito, Y. U., Peniati, E. dan Susanti, R. 2019. Simulasi Metode Statistik untuk Seleksi Single Nucleotide Polymorphism. 2019, 7, 7.
- Nyboe, C., Olsen, M. S., Nielsen-Kudsk, J. E. dan Hjortdal, V. E. 2015. Atrial fibrillation and stroke in adult patients with atrial septal defect and the long-term effect of closure. *Heart*, 101, 706-11.



- Nyboe, C., Olsen, M. S., Nielsen-Kudsk, J. E., Johnsen, S. P. dan Hjortdal, V. E. 2014. Risk of pneumonia in adults with closed versus unclosed atrial septal defect (from a nationwide cohort study). *Am J Cardiol*, 114, 105-10.
- Ouyang, P., Saarel, E., Bai, Y., Luo, C., Lv, Q., Xu, Y., et al. 2011. A de novo mutation in NKX2.5 associated with atrial septal defects, ventricular noncompaction, syncope and sudden death. *Clin Chim Acta*, 412, 170-5.
- Pabst, S., Wollnik, B., Rohmann, E., Hintz, Y., Glänzer, K., Vetter, H., et al. 2008. A novel stop mutation truncating critical regions of the cardiac transcription factor NKX2-5 in a large family with autosomaldominant inherited congenital heart disease. *Clinical Research in Cardiology*, 97, 39-42.
- Palomino Doza, J., Salguero-Bodes, R., de la Parte, M. dan Arribas-Ynsaurriaga, F. 2018. Association Between Mutations in the NKX2.5 Homeobox, Atrial Septal Defects, Ventricular Noncompaction and Sudden Cardiac Death. *Rev Esp Cardiol (Engl Ed)*, 71, 53-55.
- Pashmforoush, M., Lu, J. T., Chen, H., Amand, T. S., Kondo, R., Pradervand, S., et al. 2004. Nkx2-5 pathways and congenital heart disease; loss of ventricular myocyte lineage specification leads to progressive cardiomyopathy and complete heart block. *Cell*, 117, 373-86.
- Perera, J. L., Johnson, N. M., Judge, D. P. dan Crosson, J. E. 2014. Novel and highly lethal NKX2.5 missense mutation in a family with sudden death and ventricular arrhythmia. *Pediatr Cardiol*, 35, 1206-12.
- Pinho, E., Gomes, A. A., Silva, M. J., Torres, T. P., Coelho, A., Almeida, P. B., et al. 2013. Atrial Septal Defect in a Very Old Woman. *Cardiol Res*, 4, 41-44.
- Posch, M. G., Perrot, A., Schmitt, K., Mittelhaus, S., Esenwein, E. M., Stiller, B., et al. 2008. Mutations in GATA4, NKX2.5, CRELD1, and BMP4 are infrequently found in patients with congenital cardiac septal defects. *Am J Med Genet A*, 146A, 251-3.
- Randolph, T. R. 2008. Pathophysiology of compound heterozygotes involving hemoglobinopathies and thalassemias. *Clinical Laboratory Science*, 21, 240.
- Reamon-Buettner, S. M. dan Borlak, J. 2010. NKX2-5: an update on this hypermutable homeodomain protein and its role in human congenital heart disease (CHD). *Hum Mutat*, 31, 1185-94.
- Reamon-Buettner, S. M., Hecker, H., Spanel-Borowski, K., Craatz, S., Kuenzel, E. dan Borlak, J. 2004. Novel NKX2-5 mutations in diseased heart tissues of patients with cardiac malformations. *Am J Pathol*, 164, 2117-25.
- Rouine-Rapp, K., Russell, I. A. dan Foster, E. 2012. Congenital heart disease in the adult. *Int Anesthesiol Clin*, 50, 16-39.
- Rowan-Hull, A. 2013. Human organogenesis. *Textbook of Clinical Embryology*. Cambridge University Press, Cambridge, 118-132.
- Satwiko, M. G., Gharini, P. P. dan Hartopo, A. B. 2019. Variasi Genetika NKX2-5 dan GATA4 pada Kejadian Defek Septum Atrium Familial di RSUP Dr Sardjito Yogyakarta. Master, Universitas Gadjah Mada.
- Schott, J. J., Benson, D. W., Basson, C. T., Pease, W., Silberbach, G. M., Moak, J. P., et al. 1998. Congenital heart disease caused by mutations in the transcription factor NKX2-5. *Science*, 281, 108-11.



- Scott, D. A. dan Lee, B. 2020. Chapter 96 The Human Genome. In: KLEIGMAN, R. M., BEHRMAN, R. E., JENSON, H. B. & STANTON, B. M. D. (eds.) *Nelson textbook of pediatrics*. 21 ed.: Elsevier Health Sciences.
- Slagle, C. E. dan Conlon, F. L. 2016. Emerging Field of Cardiomics: High-Throughput Investigations into Transcriptional Regulation of Cardiovascular Development and Disease. *Trends Genet*, 32, 707-716.
- Steele, P. M., Fuster, V., Cohen, M., Ritter, D. G. dan McGoon, D. C. 1987. Isolated atrial septal defect with pulmonary vascular obstructive disease--long-term follow-up and prediction of outcome after surgical correction. *Circulation*, 76, 1037-42.
- Szabo, L., Morey, R., Palpant, N. J., Wang, P. L., Afari, N., Jiang, C., et al. 2015. Statistically based splicing detection reveals neural enrichment and tissue-specific induction of circular RNA during human fetal development. *Genome Biol*, 16, 126.
- Tanghøj, G., Lindam, A., Liuba, P., Sjöberg, G. dan Naumburg, E. 2020. Atrial Septal Defect in Children: The Incidence and Risk Factors for Diagnosis. *Congenital Heart Disease*, 15, 287-299.
- Ungerleider, G. D., Yarrabolu, T. R. dan Stewart, R. D. 2020. 47 - Atrial Septal Defects. *Critical Heart Disease in Infants and Children*, 572-586.e1.
- van der Linde, D., Konings, E. E., Slager, M. A., Witsenburg, M., Helbing, W. A., Takkenberg, J. J., et al. 2011. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. *J Am Coll Cardiol*, 58, 2241-7.
- Vecht, J. A., Saso, S., Rao, C., Dimopoulos, K., Grapsa, J., Terracciano, C. M., et al. 2010. Atrial septal defect closure is associated with a reduced prevalence of atrial tachyarrhythmia in the short to medium term: a systematic review and meta-analysis. *Heart*, 96, 1789-97.
- Wang, H., Liu, Y., Li, Y., Wang, W., Li, L., Meng, M., et al. 2019. Analysis of NKX2-5 in 439 Chinese Patients with Sporadic Atrial Septal Defect. *Med Sci Monit*, 25, 2756-2763.
- Wang, Z., Zou, L., Zhong, R., Zhu, B., Chen, W., Shen, N., et al. 2013. Associations between two genetic variants in NKX2-5 and risk of congenital heart disease in Chinese population: a meta-analysis. *PLoS One*, 8, e70979.
- Warnmark, A., Treuter, E., Wright, A. P. dan Gustafsson, J. A. 2003. Activation functions 1 and 2 of nuclear receptors: molecular strategies for transcriptional activation. *Mol Endocrinol*, 17, 1901-9.
- Watanabe, Y., Benson, D. W., Yano, S., Akagi, T., Yoshino, M. dan Murray, J. C. 2002. Two novel frameshift mutations in NKX2.5 result in novel features including visceral inversus and sinus venosus type ASD. *J Med Genet*, 39, 807-11.
- Webb, G. dan Gatzoulis, M. A. 2006. Atrial septal defects in the adult: recent progress and overview. *Circulation*, 114, 1645-53.
- Wiggs, J. L. 2018. Fundamentals of human genetics. *Ophthalmology E-Book*, 1.
- Xuan Tuan, H., The Phuoc Long, P., Duy Kien, V., Manh Cuong, L., Van Son, N. dan Dalla-Pozza, R. 2019. Trends in the Prevalence of Atrial Septal Defect



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PERBEDAAN VARIASI GENETIKA NKX2-5 DEFEK SEPTUM ATRIUM FAMILIAL DENGAN
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SARDJITO

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and Its Associated Factors among Congenital Heart Disease Patients in Vietnam. *J Cardiovasc Dev Dis*, 7.

- Yuan, F., Qiu, X. B., Li, R. G., Qu, X. K., Wang, J., Xu, Y. J., *et al.* 2015. A novel NKX2-5 loss-of-function mutation predisposes to familial dilated cardiomyopathy and arrhythmias. *Int J Mol Med*, 35, 478-86.
- Zhang, W., Li, X., Shen, A., Jiao, W., Guan, X. dan Li, Z. 2009. Screening NKX2.5 mutation in a sample of 230 Han Chinese children with congenital heart diseases. *Genet Test Mol Biomarkers*, 13, 159-62.
- Zhao, Q. M., Ma, X. J., Jia, B. dan Huang, G. Y. 2013. Prevalence of congenital heart disease at live birth: an accurate assessment by echocardiographic screening. *Acta Paediatr*, 102, 397-402.
- Zipes, D. P., Libby, P., Bonow, R. O., Mann, D. L. dan Tomaselli, G. F. 2018. Braunwald's Heart Disease E-Book: A Textbook of Cardiovascular Medicine.