

DAFTAR PUSTAKA

1. Manson JM, Carr MC. Molecular Epidemiology of Hypospadias: Review of Genetic and Environmental Risk Factors. *Birth Defects Research (Part A)*.2003;67:825–836.
2. Paulozzi LJ, Erickson D, Jackson RJ. Hypospadias trends in two US surveillance systems. *Pediatrics*. 1997; 100(5):831-834.
3. Brouwers MM, Feitz WFJ, Roelofs L, Kiemeney L, De Gier R, Roeleveld N. Risk factor for hypospadias. *Eur J Pediatr*. 2006 ; 166:671-678
4. Brouwers MM, Van Der Zanden LFM, De Gier RPE, Barten EJ, Zielhuis GA, Feitz WFJ, et al. Hypospadias : risk factor pattern and different phenotypes. *BJU International*. 2009;105:255-262.
5. Jin L, Ye R, Zheng J, Hong S, Ren A. Secular trends of hypospadias prevalence and factors associated with it in southeast China during 1993-2005. *Birth Defect Res A Clin Mol Teratol*. 2010;88(6):458-65
6. Kojima Y, Kohri K, Hayashi Y. Genetic pathway of external genitalia formation and molecular etiology of hypospadias. *Journal of Pediatric Urology*.2010;6:346-354.
7. Wang Y, Li Q, Xu J, Liu Q, Wang W, Lin Y, et al. Mutation analysis of five candidate genes in Chinese patients with hypospadias. *European Journal of Human Genetics*.2004;12:706-712.
8. Van der Zanden LFM, van Rooij IALM, Feitz WFJ, Franke B, Knoers NVAM, Roeleveld N. Aetiology of hypospadias: a systematic review of genes and environment. *Human Reproduction Update*.2012;18:260–283.
9. Van der Zanden LFM, van Rooij IALM, Feitz WFJ, Vermeulen SHHM, Kiemeney LALM, Knoers NVAM, Roeleveld N, Franke B. Genetics of hypospadias: are single-nucleotide polymorphisms in SRD5A2, ESR1, ESR2, ATF3 really associated with the malformation? *J Clin Endocrinol Metab* 2010;95:2384–2390.

10. Wang R, Dong Z, Wang W, Xiao Y, Ni J, Wang D. Mutation analysis of the SRD5A2, AR, and SF-1 genes in 52 Chinese boys with hypospadias. *J Pediatr Endocr Met.* 2013. [Cited 2013 Aug 8].
11. Zhang M, Yang J, Zhang H, Ning G, Li X, Sun S. A Novel SRD5A2 Mutation with Loss of Function Identified in Chinese Patients with Hypospadias. *Horm Res Paediatr* 2011;76:44–49.
12. Samtani R, Bajpai M, Vashisht K, Ghosh PK, Saraswathy KN. Hypospadias risk and polymorphism in SRD5A2 and CYP17 genes: case control study among Indian children. *The Journal of urology.* 2011;185:2334-2339.
13. Sata F, Kurahashi N, Ban S, Moriya K, Tanaka KD, Ishizuka M, et al. Genetic polymorphisms of 17 β -hydroxysteroid dehydrogenase 3 and the risk of hypospadias. *J Sex Med.* 2010;7:2729-2738.
14. Makridakis N, Ross RK, Pike MC, Chang L, Stanczyk FZ, Kolonel LN, et al. A Prevalent Missense Substitution that modulates activity of Prostatic Steroid 5 α -reductase. *Cancer Res* 1997;57:1020-1022.
15. Li J, Coated RJ, Gwinn M, Khoury MJ. Steroid 5- α -reductase type 2 (SRD5A2) gene polymorphisms and risk of prostate cancer: A HuGe Review. *American Journal of Epidemiology.* 2010;171:1-13.
16. Mieusset R, Soulie M. Hypospadias: Psychosocial, Sexual, and Reproductive Consequences in Adult Life. *Journal of Andrology.* 2005. 26(2):163-169.
17. Bracka A. A long term view of hypospadias. *Br J Plast Surg.* 1989;42:251-255.
18. O'Brien KLO, Varghese AC, Agarwal A. The genetic causes of male factor infertility: A review. *Fertility and Sterility.* 2010;93(1):1-12.
19. Tateno T, Sasagawa I, Ashida J, Nakada T, Ogata T. Absence of Y chromosome microdeletions in Patients with Isolated Hypospadias. *Fertility and Sterility.* 2000.74(2):399-400.
20. Marzuki NS, Suciati LP, Dewi M, Tridjaja B. Two novel mutation of SRD5A2 gene in Indonesian siblings with clinical 5- α reductase deficiency. *Journal of Pediatric Endocrinology and Metabolism.* 2011;23(12). [Abstract].

21. Wang Ming, Baskin LS. Endocrine Disruptors, Genital Development, and Hypospadias. *Journal of Andrology*. 2008; 29(5):499-504
22. Baskin LS, Himes K, Colborn T. Hypospadias and Endocrine Disruption : Is there a Connection ? *Environmental Health Perspectives*[serial online]. 2001[cited 2010january 14] ; 109(11):1175-1183. Available from : <http://ehis.niehs.nih.gov/>
23. Chong JH, Wee CK, Ho SK, Chan DK. Factors associated with hypospadias in Asian newborn babies. *J Perinatal Med*. 2006; 34(6):497-500.
24. Hadidi AT. Hypospadias surgery. Paper presented at: The International Workshop on Hypospadias Surgery; Vienna, Austria. 2006.
25. Troncoso B, Lopez PJ. Hypospadias. *Pediatric Urology Book* [Internet]. [Cited 2013Dec15]. Available from: <http://www.pediatricurologybook.com/hypospadias.html>
26. Leung AKC, Robson WLM. Hypospadias:an update. *Asian Journal of Andrology*. 2007;9:16-22
27. Kalfa N, Philibert P, Sultan C. Is hypospadias a genetic, endocrine or environmental disease, or still an unexplained malformation? *International Journal of Andrology*. 2008;32:187-197
28. Willingham E, Baskin LS. Candidate genes and their response to environmental agent In the etiology of hypospadias. *Nature Clinical Practise Urology*. 2007 ; 4(5): 270-279
29. Thai hanh TT. Hypospadias : Gene mapping and candidate Gene Studies. Department of Women's and Children's Health.[PhD dissertation]. Karolinska Institutet. 2009 [Cited 2013 Dec 20].
30. Meireles AB, Kockum I, Lundberg F, Soderhall C, Nordenskjo A . Risk factors for hypospadias in the estrogen receptor 2 gene. *JCEM endojournals*. 2007; 92(9):3712-3718
31. Carbone P, Giordano F, Nori F, Mantovani A, Taruscio D, Laurias L, et al. The possible role of endocrine disrupting chemicals in the aetiology of

- cryptorchidism and hypospadias: a population-based case–control study in rural Sicily. *International Journal of Andrology*. 2007; (30):3–13
32. Dugas J, Nieuwenhuijsen MJ, Martinez D, Iszatt N, Elliott P. Use of biocides and insect repellent and risk of hypospadias. *Occup Environ Med* 2010;67:196-200
 33. Rocheleau CM, Romitti PA, Dennis LK . Pesticides and hypospadias: a meta-analysis. *Journal of Pediatric Urology*. 2009 ; 5 : 17-24
 34. Fisch H, Golden RJ, Libersen GL, Hyun GS, Madsen P, New MI, Hensle TW. Maternal Age as a Risk Factor For Hypospadias. *J Urol*. 2001. 165(3):934-936.
 35. Hussain N, Chaghtai A, Herndon A, Herson VC, Rosenkrantz TS, McKenna PH .Hypospadias and early gestation growth restriction in infants. *Official Journal of The American Academy of Pediatrics*. 2002; 109:473-478.
 36. Porter MP, Faizan MK, Grady RW, Mueller BA. Hypospadias in Washington State: Maternal Risk Factors and Prevalence Trends. *Pediatrics*. 2005. 115(4). 495-499.
 37. Charmichael SL, Shaw GM, Laurent C, Croughan MS, Olney RS, Lammer EJ: Maternal Progestin Intake and Risk of Hypospadias. *Arch Pediatr Adolesc Med*. 2005. 159(10):957-962.
 38. Pasqualotto FF, Lucon AM, Sobreiro BP, Pasqualotto EB, Arap S. Effects of medical therapy, alcohol, smoking, and endocrine disruptors on male infertility. *Rev.Hosp. Clín. Fac. Med. S. Paulo*. 2004;59(6):375-382.
 39. deCalais FL, Soardi FC, Petroli RJ, Gori Lusa AL, de Paiva e Silva RB, Maciel-Guerra AT, et al. Molecular Diagnosis of 5 α -Reductase type II deficiency in Brazilian sibling with 46,XY Disorder of Sex Development. *Int J.Mol.Sci*. 2011;12: 9471-9480
 40. Thai H, Kalbasi M, Lagerstedt K, Frisen L, Kockum I, Nordenskjold A. The Valine allele of the V89L polymorphism in the five-alpha reductase gene confers a reduced risk for hypospadias. *Journal of Clinical Endocrinology and Metabolism*. 2005;90:6695-6698.

41. Vineeth VS, Mallini SS. A Journey of Y chromosomal genes and male infertility. *Int J Hum Genet.* 2011;11(4):203-215.
42. Layman LC. Human gene mutations causing infertility. *J Med Genet.* 2002;39:153-161.
43. Foresta C, Moro E, Ferlin A. Prognostic value of Y deletion Analysis. *Hum Reprod.* 2001;16(8):1543-7.
44. C. Kamp, K. Huellen, and K. Huellen. High deletion frequency of the complete AZFa sequence in men with Sertoli-cell-only syndrome. *Molecular Human Reproduction.* 2001; 7(10): 987–994.
45. Costa PN, Plancha CE, Goncalves J. Genetic Dissection of the AZF region of the human Y chromosome: Thriller or filler for male (in)fertility?. *Journal of Biomedicine and Biotechnology.* 2010: 1-18. [Available from : <http://dx.doi.org/10.1155/2010/936569>]
46. Vogt PH. Azoospermia factor (AZF) in Yq11: towards a molecular understanding of its function for human male fertility and spermatogenesis. *Reprod Biomed Online* 2005;10:81–93.
47. Skaletsky H, Kuroda-Kawaguchi T, Minx PJ, Cordum HS, Hillier L, Brown LG, et al. The male-specific region of the human Y chromosome is a mosaic of discrete sequence classes. *Nature* 2003;423:825–37.
48. Tessari A, Salata E, Ferlin A, Bartolini L, Slongo ML, Foresta C. Characterization of HSFY, a novel AZFb gene on the Y chromosome with possible role in human spermatogenesis. *Mol Hum Reprod.* 2004;10(4):253-58.
49. Reynolds N, Cooke HJ. Role of the DAZ genes in male fertility. *Reprod Biomed Online.* 2005;10: 72–80.
50. Reijo R, Lee TY, Salo P, Alagappan R, Brown LG, Rosenberg M, et al. Diverse spermatogenic defects in humans caused by Y chromosome deletions encompassing a novel RNA-binding protein gene. *Nat Genet.* 1995;10:383–93.

51. Sadeghi-Nejad H, Farrokhi F. Genetics of azoospermia: current knowledge, clinical implications, and future directions. Part II. Y chromosome microdeletions. *Urol J*. 2007;4:192–206.
52. Jager RJ, Anvret M, Hall K, Scherer G. A human XY female with a frame shift mutation in the candidate testis-determining gene SRY. *Nature*. 1990;348:452-4.
53. Ostrer H. Sexual differentiation. *Sem Reprod Med*. 2000;18:41-9.
54. SRY. [Internet] 2008 November [Updated 2014 June 23; cited 2014 Juni 27]. Available from: <http://ghr.nlm.nih.gov/gene/SRY>
55. Markoulatos P, Siafakas N, Moncany M. Multiplex polymerase chain reaction: a practical approach. *Journal of Clinical Laboratory Analysis*. 2002;16:47-51.
56. Dogget NA. The polymerase chain reaction and sequence-tagged sites. Los Alamos Science. 1992;20:128-134
57. Simoni M, Bakker E, Krausz C. EAA/EMQN Best Practice Guidelines for Molecular Diagnosis of Y-chromosomal microdeletions. State of The Art. *International Journal of Andrology*. 2004;27:240-49.
58. Liow SL, Yong EL, Ng SC. Prognostic value of Y deletion analysis. *Hum Reprod*. 2001;16(1):9-12
59. Hoag CC, Gotto GT, Morrison KB, Coleman GU, MacNeily AE. Long-term functional outcome and satisfaction of patients with hypospadias repaired in childhood. *Canadian Urological Association*. 2008;2(1):23-31.
60. NCBI. dbSNP short genetic variation. refSNP rs523349. [cited 2014 April 21]. Available from: <http://www.ncbi.nlm.nih.gov/>
61. Vokwana CKJ. Mapping gene variation in Sub-Saharan African populations. [Dissertation of Faculty of Health Sciences]. University of Witwatersrand, Johannesburg. 2008. [Cited 2014 January 7]. Available from: <http://wiredspace.wits.ac.za/handle/10539/6892>
62. Bio-rad. A Guide to polyacrilamide gel electrophoresis and detection. <http://www.bio-rad.com/>

63. Faradz SMH. Pengantar sitogenetika, genetika molekuler dan alat bantu konseling genetika. Laboratorium Bioteknologi Kedokteran FK UNDIP. 2000
64. Sunarno JM. Distribusi gen *Azoospermia Factor* (AZF) pada pasien dengan hipospadia. Tesis. 2009. Available at : <http://eprint.undip.co.id>.
65. Roshdy OH, Mohammad NS, Kamha ES, Omar M. Genetic analysis of 5 α -reductase type II enzyme in relation to oxidative stress in cases of *Androgenetic alopecia* in a sample of Egyptian population. *Our Dermatol Online*.2013;4(4):468-474
66. Barril P, Nates S. Introduction to agarose and polyacrilamide gel electrophoresis matrices with respect to their detection sensitivities. *Institute de virologia "Dr. J.M.Vanella"*. Argentina. Available at : <http://www.intechopen.com>. [cited March 31th, 2015].
67. Anonimous. Safer Alternatives to Ethidium bromide. Risk Management Services environmental services. Available at: <http://www.riskmanagement.ubc.ca/environment>.
68. Van Rooij I.A.L.M, van der Zanden L.F.M, Brouwers M.M, Knoers N.V.A.M, Feitz W.F.J, Roelevend N. Risk factors for different phenotypes of hypospadias: results from a Dutch case-control study. *BJU International*. 2013;112:121-128.
69. Khan M, Majeed A, Hayat W, Ullah H, Naz S, Shah SA, et al. Hypospadia Repair : A single centre experience. *Plastic Surgery International*. 2014:1-7
70. Manzoni G, Bracka A, Palminteri E, Marriocco G. Hypospadia surgery: when, what, and by whom?. *BJU international*. 2004;94(8):1188-1195
71. Ferlin A, Arredi B, Speltra E, Cazzadore C, Selice R, Garolla A, et al. Molecular and clinical characterization of Y chromosome microdeletions in infertile men: a 10-year experience in Italy. 2007;92(3):762-770.