

## DAFTAR PUSTAKA

1. Hughes I.A. 2002. *Intersex. BJU International* 1990: 769-776
2. Faradz SMH. Sexual Ambiguity in Semarang, *a Cytogenetic Approach. In : Proceedings of the National seminar and Workshop on Ambiguous Genitalia*. Semarang, Indonesia, 2004 : 4
3. Stuart ALO and Brain C E. *Early assessment of ambiguous genitalia. Arch Dis Child* 2004 ; 89: 401-407
4. Susanto R. Profil Hormonal pada Anak dengan Ambigus Genitalia. *In: Proceedings of the National seminar and Workshop on Ambiguous Genitalia*, Semarang, Indonesia, 2004 : 46-76
5. Maharaj NR, Dhai A, Wiersma R, and Moodley. *Intersex condition in children and adolescents; surgical, ethical and legal considerations. J Pediatr Adolesc Gynecol* 2005;18: 399-402
6. Göllü G, Yildiz RV, Bingol KM, Yagmurlu A, Senyu" cel, Tanju A, Go"kcora H, Dindar H. *Ambiguous Genitalia: An Overview of 17 years Experience. jped surg* 2007; 42: 840-844
7. MacLaughlin DT and Donahoe PK. *Sex Determination and Differentiation. Review article in N Engl J Med* 2004; 350:367-378
8. Ferlin A, Moro E, Rossi A, Dallapiccola and Foresta. *The human Y chromosome's azoospermia factor b (AZFb) region: sequence, structure, and deletion analysis in infertile men. J Med Genet* 2003; 40: 18-24
9. Quilter CR, Nathwani N, Conway GS, Stanhope R, Ralph D, Bahadur, et al. *A comparative study between infertile males and patients with Turner syndrome to determine the influence of sex chromosome mosaicism and the breakpoints of structurally abnormal Y chromosomes on phenotypic sex. J Med Genet* 2002; 39: 80-86
10. Aktas D, Alikasifoglu M, Gonc N, Senocak ME, Tuncbilek E. *Isodicentric Y (p11.32) chromosome in an infant with mixed gonadal dysgenesis. J.ejmg.* 2006; 49 : 141-149
11. Liow SL, Yong EL, and Ng SC. *Prognostic value of Y Deletion Analysis. Hum. Reprod.* 2001; 16 (1): 9-12.

12. Siffroi JP, Bourhis CL, Krausz C, Barbaux S, Murci LQ, Kanafani S, et al. *Sex Chromosome Mosaicism in Males Carrying Y Chromosome Long Arm Deletion. Hum. Reprod.* 2000; 15 (12): 2559-2562
13. Sankar VH, Phadke R Shuba. *Ring Chromosome 13 in an Infant with Ambiguous Genitalia. Indian Ped* 2006; 43.
14. Thangaraj K, Subramanian S, Reddy AG, Singh L. *Unique Case of Deletion and Duplication in the Long Arm of the Y Chromosome in an Individual With Ambiguous Genitalia. Research Letter in Am J Med Gen* 2003;116A: 205–207
15. Liou JD, Ma YY, Gibson LH, Hua Su, Charest N, Lau Yun-Fai C, and Yang-Feng TL. *Cytogenetic and molecular studies of a familial paracentric inversion of Y chromosome present in a patient with ambiguous genitalia. Am J Med Gen* 1998; 70 (2): 134-137
16. DesGroseilliers M, Bergeron MB, Brochu P, Lemyre E, Lemieux N. *Phenotypic variability in isodicentric Y patients: study of nine cases. Short Report on Clin Gen* 2006; 70 (2): 145-150
17. Dundar M, Gordon L, Hasan A, Kurtoglu S, Demiryilmaz F, and Kucukaydin M. *A case of ambiguous genitalia presenting with a 45,X/46,Xr(Y)(p11.2;q11.23)/ 47,X, idic(Y)(p11.2), idic(Y)(p11.2) karyotype. Ann Gén* 2001; 44 :5–8
18. Smith YR, Stetten G, Charity L, Isacson C, Gearhart, And Namnoum AB. *Ambiguous Genitalia in an Elderly Woman With A Mosaic 45,X/46,X,DICM(Q 11.2) Karyotype. A Case Report in Urology* 1996; 47: 259-262.
19. Mundhofir FEP, Winarni TI, Juniarto AZ, Faradz SMH. *Ring Chromosome Y in a boy with genital anomaly, a case report Publish in PAAI, Semarang* 2006
20. Bruyere, Speevak, Winsor, Fr'eminville, Farrell, Gowan-Jordan, et al. *Isodicentric Yp: prenatal diagnosis and outcome in 12 cases. Prenat Diagn* 2006; 26: 324-329
21. Winarni TI, Juniarto AZ, Faradz SMH, Hamel B. *Chromosomal Finding Among Female with Amenorhe Primer. Presented in Joint 7<sup>th</sup> Human Genome Organization Asian pacific Meeting and The 8<sup>th</sup> Asian pacific Conference on Human Genetic, Philipines April 2-5<sup>th</sup> 2008*

22. Jobling MA and Smith CT. *The Human Y Chromosome: An Evolution marker Comes of Age. Review Article in Nature* 2003; available on [www.nature.com](http://www.nature.com)
23. Barch MJ. *The AGT Cytogenetics Laboratory Manual*. Lippincot-Raven Publisher, Philadelphia, New York 1991; 10-11; 180-183
24. Passarge E. *Color Atlas of Genetics 3<sup>rd</sup> Edition*. Thieme Stuttgart, New York 2007: 200-204
25. Barbosa AAL, Cavalli IJ, Abel K, Santos M and Azevêdo ES. *Family names and the length of the Y chromosome in Brazilian blacks. Braz. J. Genet* 1997; vol. 20(1)
26. Simoni M, Bakker E, and Karausz C. *EAA/EMQN best practice guidelines for molecular diagnosis of y-chromosomal microdeletions State of the art 2004. Intl journal of androl* 2004; 27: 240-249
27. Hughes I.A. *Ambiguous Genitalia dalam Brook C.G.D, Clayton P.E, Brown R.S. (ed). Clinical Pediatric Endocrinology 5<sup>th</sup> ed. Blackwell Publihing, Victoria* 2005 : 171-182
28. Boehmer ALM. *Familial Disorder of sexual Differentiation; a clinical and molecular genetic evaluation* 2000: 3-30
29. Rittler Monica and Castilla E.E. *Endocrine Disruptor and Congenital Anomalies. Cad. Saude Publica, Rio de Janeiro* 2002; 18(2): 421-428
30. Sadler TW. *Langman's Medical Embriology. 9<sup>th</sup> ed. Pennsylvania: Lippincott Williams & Wilkins* 2000: 319-339
31. Mueller RF and Young ID. *Emery's Elements of Medical genetics. Churchill Livingstone, Edinburgh* 2001: 256
32. Berkovitz GD, Seeherunvong T. *Abnormalities of gonadal differentiation. Bailliere's clinical endocrinology and metabolism* 1998; 12 (1) : 133-142
33. Quigley CA, Bellis AD, Marschke KB, El-Awady MK, Wilson EM, French FS. *Androgen Receptor Defects: Historical, Clinical, and Molecular Perspectives. Endo Jnls* 1995; 16(3): 12
34. Kim S, Jung S, Kim H. *Chromosome Abnormalities in a referred Population for Suspected Chromosomal Aberrations: A Report of 4117 Cases. J Korean Med Sci* 1999; 14: 373-376

35. Telvi L, Lebbar A, Pino OD, Barbet JP and Louis J. *45,X/46,XY Mosaicism: Report of 27 Cases. Peds* 1999; 104: 304-308
36. Velissariou V, Antoniadi T, Patsalis P, Christopoulou S, Hatzipoulou A, Jackie Donoghue, et al. *Prenatal diagnosis of two rare de novo structural aberration of Y chromosome: cytogenetic and molecular analysis. Prenat Diagn* 2001; 21 :484-487
37. Faradz SMH. *Pengantar Sitogenetika, Genetika Molekuler, dan Alat bantu Konseling Genetika. Laboratorium Bioteknologi FK UNDIP. 2002*
38. Schinzel A. *Catalogue of Unbalanced Chromosome Aberrations in Man. 2<sup>nd</sup> Ed. de Gruyter New York, 2001.p. 924-941*
39. Gardner, McKinlay RJ, Sutherland, Grant R. *Chromosome Abnormalities and Genetic Counseling , 3rd Edition Oxford University Press, 2004. p. 265-273*
40. Rappold GA, Fukami M, Niesler B, et al. *Deletions of the Homeobox SHOX (Short Stature Homeobox) Are an Important Cause of Growth Failure in Children with Short Stature. J Clin Endocrinol Metab* 2002, 87(3) : 1402-14
41. Rappold GA, Blum WF, Shavrikova P, et al. *Genotypes and Phenotypes in Children with Short Stature: clinical indicators of SHOX haploinsufficiency. J Med Genet* 2007; 44: 306-313
42. Yusuf I. *Konseling Pra dan Pasca Tindakan Penyesuaian Kelamin. In: Proceedings of the National Seminar and Workshop on Ambiguous Genitalia, Semarang, Indonesia : 27-31*
43. Firth HV, Hurst JA. *Oxford desk Reference Clinical Genetics. In: Hall JG, editor. New York: Oxford University Press, 2005. p. 496-563*
44. Visoosak J, Aylstock M and Graham JM. *Kilnefelter Syndrome and Its Variants: An Update and Review for the Primary Pediatrician. Clin Pediatr (Phila)* 2001; 40; 639
45. Akkari Y, Lawee H, Kelson S, Smith C, Davis C, Boyd L, et al. *Y chromosome heterochromatin of differing lengths in two cell populations of same individual. Prenat Diagn* 2005; 25: 304-306.