

## References

- Ahmed SF, Hughes IA. (2002). The genetics of male undermasulinization. Clin Endocrinol .56:1-18.
- Akane A., Shiono H., Matsubara K., Nakahori Y., Seki S., Nagafuchi S., Yamada M., Nakagome Y. (1991) Sex identification of forensic specimens by polymerase chain reaction (PCR): two alternative methods, Forensic Sci. Int. 49 : 81-88.
- Allen TD. (1976). Disorders of sexual differentiation. Urology. 7(4 Suppl): 1-32.
- Berta P, Hawkins JR, Sinclair AH, Taylor A, Griffiths BL, Goodfellow PN, Fellous M. (1990) Genetic evidence equating SRY and the testis-determining factor. Nature. 348:448-451.
- Danon M., Friedman SC. (1996) Ambiguous genitalia, micropenis, hypospadias, and cryptorchidism. In: Lifshitz F (ed) Pediatric Endocrinology. Marcel Dekker, New York. 281-304.
- De Peretti E., Forest MG. (1982) Pitfalls in the etiological diagnosis of congenital adrenal hyperplasia in the early neonatal period. Horm Res. 16:10-22.

- Donahoe PK. (1987) The diagnosis and treatment of infants with intersex abnormalities. *Pediatr cli North AM* 34: 1333- 1348.
- Federmann DD. (1967) *Abnormal Sexual Development: A Genetic and Endocrine Approach to Differential Diagnosis*. Philadelphia. WB Saunders Co.
- Forest MG. (1981) Inborn errors of testosterone biosynthesis. In: Josso N, ed. *The Intersex Child, Pediatric and Adolescent Endocrinology*. S Karger AG. 133-55.
- Frimberger D., Gearhart JP. (2005a) Ambiguous genetalia and intersex. *Urol Int*. 75: 291- 297.
- Frimberger D., Gearhart JP. (2005b) Ambiguous genetalia and intersex. *Urol Int*. 75: 291- 297.
- Grumbach MM, Ducharme JR. (1960) The effects of androgens on fetal sexual development. Androgen-induced female pseudohermaphroditism. *Fertil Steril*. 11:157.
- Jager et al. (1990) A human XY female with frame shift mutation in the candidate testis-determining gene SRY. *Nature* .348:452-453.
- Josso N., Cate R., Picard, J-Y., Vigier B., Di Clemente N., Wilson C., Imbeaud S., Pepinsky R.B., Guerrier D., Boussin L., Legeai L., Carré-Eusèbe D.(1993) Anti-Müllerian Hormone: The Jost Factor. *Recent Progress in Hormone Research*.Vol. 48: 1-59.

- Jost A. (1985) Initial stages of gonadal development. Theories and methods.  
Arch Ana Microsc Morphol Exp. 74(1): 39-41.
- Jost A. (1985) Initial stages of gonadal development. Theories and methods  
Arch Ana Microsc Morphol. Exp. 18: 41-49.
- Jost A. (1970) Hormonal factors in the sex differentiation of the mammalian  
foetus. Philos Trans R Soc Lond B Biol Sci. 259:119-130.
- Jost A. (1954) Hormonal factors in development of the fetus. Cold Spring  
Harbor Symp Quant Biol. 19: 167.
- Koopman, P. et al. (2001) Regulation of male sexual development by SRY  
and Sox9. J. Exp. Zool. 290: 463-474.
- Kula K., Slowikowska \_Hilczer J. (2000). Sexual differentiation of the human  
brain. 124: 601-609.
- Lau E.C., T.K. Mohandas, Shapiro L.J., Slavkin H.C., Snead M.L. (1989)  
Human and mouse amelogenin gene loci are on the sex chromosomes,  
Genomics 4: 162-168.
- Lee MM, Donahoe PK. (1993) Müllerian inhibiting substance: a gonadal  
hormone with multiple functions. Endocr Rev. 14:152-164.
- Mizunok ,Kojima Y.,Tozowa K, Sasaki S, Hayashi Y , Kohri K . (2005)  
Molecular evaluation of the SRY gene for gonads of patients with mixed  
gonadal dysgenesis. Int J Urol. 12: 673-676.

- Nakahori Y., Hamano K., Iwaya M., Nakagome Y. (1991) Sex identification by the polymerase chain reaction using X–Y homologous primers, *Am. J. Med. Genet.* 39: 472-473.
- Nakahori Y., Takenaka O., Nakagome Y. (1991) A human X–Y homologous region encodes amelogenin, *Genomics.* 9: 264-269.
- New MI. (2004) an update of congenital adrenal hyperplasia. *Ann NY A cad Sci* 1038:14-43.
- Ng KL, Ahmed SF, Hughes IA. (2000) Pituitary-gonadal axis in male undermasculinisation. *Arch Dis Child* . 82:54–8.
- Ostrer H. (2000) Sexual differentiation. *Semin Reported Med.* 18: 41-49.
- Painter Ts. (1921) The Y chromosome in mammals science. 53: 503-504.
- Parker KL, Schedl A, Schimmer BP. (1999) Gene interactions in gonadal development. *Annu Rev Physiol* 61:417-433.
- Parker. KL., Schimmer BP.,Schedl A. (2001) Genes essential for early events in gonadal development. *Exs.* 11-24.
- Penning TM. (1997) Molecular endocrinology of hydroxysteroid dehydrogenases. *Endocr Rev.* 18:281-305.
- Rosenfield RL, Lucky AW, Allen TD. (1980) The diagnosis and management of intersex. *Curr Prob pediatri* 10: 1-66.
- Salido E.C., Yen P.H., Koprivnikar K., Yu L., Shapiro L.J. (1992) The human enamel protein gene amelogenin is expressed from both the X and

- the Y chromosomes, *Am. J. Hum. Genet.* 50: 303-316.
- Schafer AJ, Goodfellow PN. (1996) Sex determination in humans. *Bioassays* 18: 955-963.
- Speiser PW, DuPont J, Zhu D, Serrat J, Buegeleisen M, Tusie-Luna MT, Lesser M, New MI, White PC. (1992) Disease expression and molecular genotype in congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *J Clin Invest.* 90:584-595.
- Sullivan K.M., Mannucci A., Kimpton C.P., Gill P. (1993) A rapid and quantitative DNA sex test: fluorescence-based PCR analysis of X–Y homologous gene amelogenin, *Biotechniques* .15: 636-641.
- Vainio S, Heikkila M, Kispert A. (1999) Female development in mammals is regulated by Wnt-4 signalling. *Nature.* 379:707-710.
- Villee DB.(1972) The development of steroidogenesis. *Am J Med.* 53:533-544.
- White PC, Curnow KM, Pascoe L. (1994) Disorders of steroid 11 $\beta$  hydroxylase isozymes. *Endocr Rev* .15:421-438.
- Wilson JD, Foster DW, Kronenberg HM, Larsen PR. (1998) Disorders of sex differentiation. In: (eds) *Williams Textbook of Endocrinology*. W.B. Saunders, Philadelphia. 1303-1426.
- Wilson JD, Griffin JE, Russell DW. (1993) Steroid 5 $\alpha$ -reductase 2 deficiency. *Endocr Rev* 14:577-593.