

## DAFTAR PUSTAKA

- 1 Gravholt CH, Andersen NH, Conway GS, Dekkers OM, Geffner ME, Klein KO *et al.* Clinical practice guidelines for the care of girls and women with Turner syndrome: Proceedings from the 2016 Cincinnati International Turner Syndrome Meeting. *Eur J Endocrinol* 2017; **177**: G1–G70.
- 2 Sybert VP, McCauley E. Turner's Syndrome. *N Engl J Med* 2004; **351**: 1227–1238.
- 3 Turner HH. A syndrome of infantilism, congenital webbed neck, and cubitus valgus. *Endocrinology* 1938; **23**: 566–574.
- 4 Ullrich O. Über typische Kombinationsbilder multipler Abartungen. *Z Kinderheilkd* 1930; **49**: 271–276.
- 5 Bondy CA. Care of Girls and Women with Turner Syndrome: A Guideline of the Turner Syndrome Study Group. *J Clin Endocrinol Metab* 2007; **92**: 10–25.
- 6 Gunther DF. Ascertainment Bias in Turner Syndrome: New Insights From Girls Who Were Diagnosed Incidentally in Prenatal Life. *Pediatrics* 2004; **114**: 640–644.
- 7 Romero ST, Geiersbach KB, Paxton CN, Rose NC, Schisterman EF, Branch DW *et al.* Differentiation of genetic abnormalities in early pregnancy loss. *Ultrasound Obstet Gynecol* 2015; **45**: 89–94.
- 8 Ross JL, Kowal K, Quigley CA, Blum WF, Cutler GB, Crowe B *et al.* The phenotype of short stature homeobox gene (SHOX) deficiency in childhood: Contrasting children with Leri-Weill dyschondrosteosis and turner syndrome. *J Pediatr* 2005; **147**: 499–507.
- 9 Ross JL, Scott C. J, Marttila P, Kowal K, Nass A, Papenhausen P *et al.* Phenotypes associated with SHOX deficiency. *J Clin Endocrinol Metab* 2001; **86**: 5674–5680.
- 10 Toniolo D, Ph D, Rizzolio F. X chromosome and ovarian failure. *Semin Reprod Med* 2007; **1**: 264–271.
- 11 Bondy C, Bakalov VK, Cheng C, Olivieri L, Rosing DR, Arai AE. Bicuspid aortic valve and aortic coarctation are linked to deletion of the X chromosome short arm in turner syndrome. *J Med Genet* 2013; **50**: 662–665.
- 12 Säwendahl L, Davenport ML. Delayed diagnoses of Turner's syndrome: Proposed guidelines for change. *J Pediatr* 2000; **137**: 455–459.
- 13 Gotzsche CO, Krag-Olsen B, Nielsen J, Sorensen KE, Kristensen BO. Prevalence of cardiovascular malformations and association with karyotypes in Turner's syndrome. *Arch Dis Child* 1994; **71**: 433–436.
- 14 Gravholt CH, Juul S, Naeraa RW, Hansen J. Morbidity in Turner syndrome. *J Clin Epidemiol* 1998; **51**: 147–158.
- 15 Waters P, Bae D. *Pediatric Hand and Upper Limb Surgery*. Lippincott Williams & Wilkins: Philadelphia, 2012 [www.lww.com](http://www.lww.com).
- 16 Brook CGD, Morset G, Zachmann M, Prader A. Growth in children with 45 , XO Turner ' s syndrome. 1974; : 789–795.
- 17 Lyon AJ, Preece MA, Grant DB. Growth curve for girls with Turner syndrome. *Arch Dis Child* 1985; **60**: 932–935.
- 18 Singh RP, Carr DH. The anatomy and histology of XO human embryos and fetuses. *Obstet Gynecol Surv* 1967; **22**: 525–528.
- 19 Massarano AA, Adams JA, Preece MA, Brook CGD. Ovarian ultrasound appearances in Turner syndrome. *J Pediatr* 1989; **114**: 568–573.
- 20 Reyes FI, Koh KS, Faiman C. Fertility in women with gonadal dysgenesis. *Am J*

- Obstet Gynecol* 1976; **126**: 668–670.
- 21 King C, Magenis E, Bennet S. Pregnancy and the Turner syndrome. *Obstet Gynecol* 1978; **52**: 617–624.
  - 22 Ho VB, Bakalov VK, Cooley M, Van PL, Hood MN, Burklow TR *et al.* Major vascular anomalies in Turner syndrome: Prevalence and magnetic resonance angiographic features. *Circulation* 2004; **110**: 1694–1700.
  - 23 Loscalzo ML. Association Between Fetal Lymphedema and Congenital Cardiovascular Defects in Turner Syndrome. *Pediatrics* 2005; **115**: 732–735.
  - 24 Mortensen KH, Andersen NH, Gravholt CH. Cardiovascular phenotype in turner syndrome-integrating cardiology, genetics, and endocrinology. *Endocr Rev* 2012; **33**: 677–714.
  - 25 Sachdev V, Matura LA, Sidenko S, Ho VB, Arai AE, Rosing DR *et al.* Aortic Valve Disease in Turner Syndrome. *J Am Coll Cardiol* 2008; **51**: 1904–1909.
  - 26 Versacci P, Silvestri LM, Maiolo S, Marino B. Surgical outcomes for patients with Turner syndrome. *Pediatr Cardiol* 2014; **35**: 1080–1081.
  - 27 Matura LA, Ho VB, Rosing DR, Bondy CA. Aortic dilatation and dissection in Turner syndrome. *Circulation* 2007; **116**: 1663–1670.
  - 28 Carlson M, Airhart N, Lopez L, Silberbach M. Moderate aortic enlargement and bicuspid aortic valve are associated with aortic dissection in turner syndrome: Report of the international turner syndrome aortic dissection registry. *Circulation* 2012; **126**: 2220–2226.
  - 29 Committee P, Society A. Increased maternal cardiovascular mortality associated with pregnancy in women with Turner syndrome. *Fertil Steril* 2012; **97**: 282–284.
  - 30 Bondy CA, Ceniceris I, Van PL, Bakalov VK, Rosing DR. Prolonged Rate-Corrected QT Interval and Other Electrocardiogram Abnormalities in Girls With Turner Syndrome. *Pediatrics* 2006; **118**: e1220–e1225.
  - 31 Bakalov VK, Chen ML, Baron J, Hanton LB, Reynolds JC, Stratakis CA *et al.* Bone mineral density and fractures in Turner syndrome. *Am J Med* 2003; **115**: 259–264.
  - 32 Hansen S, Brixen K, Gravholt CH. Compromised trabecular microarchitecture and lower finite element estimates of radius and tibia bone strength in adults with turner syndrome: A cross-sectional study using high-resolution-pQCT. *J Bone Miner Res* 2012; **27**: 1794–1803.
  - 33 Gravholt CH, Fedder J, Naeraa RW, Müller J. Occurrence of gonadoblastoma in females with turner syndrome and Y chromosome material: A population study. *J Clin Endocrinol Metab* 2000; **85**: 3199–3202.
  - 34 Hasle H, Olsen JH, Nielsen J, Hansen J, Friedrich U, Tommerup N. Occurrence of cancer in women with Turner syndrome. *Br J Cancer* 1996; **73**: 1156–1159.
  - 35 Bösze P, Tóth A, Török M. Hormone replacement and the risk of breast cancer in Turner’s syndrome. *N Engl J Med* 2006; **355**: 2599–600.
  - 36 Davenport ML, Roush J, Liu C, Zagar AJ, Eugster E, Travers S *et al.* Growth hormone treatment does not affect incidences of middle ear disease or hearing loss in infants and toddlers with turner syndrome. *Horm Res Paediatr* 2010; **74**: 23–32.
  - 37 Hultcrantz M, Sylven L, Borg E. Ear and hearing problems in 44 middle-aged women with Turner’s syndrome. *Hear Res* 1994; **76**: 127–132.
  - 38 Verver EJJ, Freriks K, Thomeer HGXM, Huygen PLM, Pennings RJE, Alfen-van der Velden AAEM *et al.* Ear and hearing problems in relation to karyotype in children with Turner syndrome. *Hear Res* 2011; **275**: 81–88.
  - 39 Wikiera B, Mulak M, Koltowska-Haggstrom M, Noczynska A. The presence of eye defects in patients with Turner syndrome is irrespective of their karyotype.

- Clin Endocrinol (Oxf)* 2015; **83**: 842–848.
- 40 Chrousos GA, Ross JL, Chrousos G, Chu FC, Kenigsberg D, Cutler G *et al*. Ocular Findings in Turner Syndrome: A Prospective Study. *Ophthalmology* 1984; **91**: 926–928.
  - 41 Vladimir K. Bakalov<sup>1</sup>, Liat Gutin<sup>1</sup>, Clara M Cheng<sup>1</sup>, Jian Zhou<sup>1</sup>, Puja Sheth<sup>1</sup> KS, Sruthi Arepalli<sup>1</sup>, Vien Vanderhoof<sup>2</sup>, Lawrence M. Nelson<sup>2</sup> and CAB. Autoimmune Disorders in Women With Turner Syndrome and Women With Karyotypically Normal. 2013; **38**: 315–321.
  - 42 Caprio S, Boulware S, Diamond M, Sherwin RS, Carpenter TO, Rubin K *et al*. Insulin resistance: An early metabolic defect of turner's syndrome. *J Clin Endocrinol Metab* 1991; **72**: 832–836.
  - 43 Freriks K, Timmermans J, Beerendonk CCM, Verhaak CM, Netea-Maier RT, Otten BJ *et al*. Standardized multidisciplinary evaluation yields significant previously undiagnosed morbidity in adult women with turner syndrome. *J Clin Endocrinol Metab* 2011; **96**: 1517–1526.
  - 44 Bakalov VK, Cheng C, Zhou J, Bondy CA. X-chromosome gene dosage and the risk of diabetes in Turner syndrome. *J Clin Endocrinol Metab* 2009; **94**: 3289–3296.
  - 45 Wooten N, Bakalov VK, Hill S, Bondy CA. Reduced abdominal adiposity and improved glucose tolerance in growth hormone-treated girls with Turner syndrome. *J Clin Endocrinol Metab* 2008; **93**: 2109–2114.
  - 46 Elsheikh M, Hodgson HJF, Wass JAH, Conway GS. Hormone replacement therapy may improve hepatic function in women with Turner's syndrome. *Clin Endocrinol (Oxf)* 2001; **55**: 227–231.
  - 47 Handler MZ, Derrick KM, Lutz RE, Morrell DS, Davenport ML, Armstrong AW. Prevalence of Pilomatricoma in Turner Syndrome. *JAMA Dermatology* 2013; **149**: 559.
  - 48 Skuse DH, James RS, Bishop DVM, Coppin B, Dalton P, Aamodt-Leeper G *et al*. Evidence from Turner's syndrome of an imprinted X-linked locus affecting cognitive function. *Nature* 1997; **387**: 705–708.
  - 49 Hanton L, Axelrod L, Bakalov V, Bondy CA. The Importance of Estrogen Replacement in Young Women with Turner Syndrome. *J Women's Heal* 2003; **12**: 971–977.
  - 50 Azcona C, Bareille P, Stanhope R. Lesson of the week: Turner's syndrome mosaicism in patients with a normal blood lymphocyte karyotype. *BMJ* 1999; **318**: 856–7.
  - 51 Gemmill RM, Pearce-birge L, Bixenman H, Hecht BK, Allanson JE. Y Chromosome-specific DNA Sequences in Turner-Syndrome Mosaicism. 1987; : 157–167.
  - 52 Schoemaker MJ, Swerdlow AJ, Higgins CD, Wright AF, Jacobs PA. Cancer incidence in women with Turner syndrome in Great Britain: a national cohort study. *Lancet Oncol* 2008; **9**: 239–246.
  - 53 Ranke MB, Pflüger H, Rosendahl W, Stubbe P, Enders H, Bierich JR *et al*. Turner syndrome: Spontaneous growth in 150 cases and review of the literature. *Eur J Pediatr* 1983; **141**: 81–88.
  - 54 Davenport ML, Crowe BJ, Travers SH, Rubin K, Ross JL, Fechner PY *et al*. Growth Hormone Treatment of Early Growth Failure in Toddlers with Turner Syndrome: A Randomized, Controlled, Multicenter Trial. *J Clin Endocrinol Metab* 2007; **92**: 3406–3416.
  - 55 Klein KO, Rosenfield RL, Santen RJ, Gawlik AM, Backeljauw PF, Gravholt CH

- et al.* Estrogen Replacement in Turner Syndrome: Literature Review and Practical Considerations. *J Clin Endocrinol Metab* 2018; **103**: 1790–1803.
- 56 Lucaccioni L, Wong SC, Smyth A, Lyall H, Dominiczak A, Faisal Ahmed S *et al.* Turner syndrome-issues to consider for transition to adulthood. *Br Med Bull* 2015; **113**: 45–58.
- 57 van Pareren YK, de Muinck Keizer-Schrama SMPF, Stijnen T, Sas TCJ, Jansen M, Otten BJ *et al.* Final Height in Girls with Turner Syndrome after Long-Term Growth Hormone Treatment in Three Dosages and Low Dose Estrogens. *J Clin Endocrinol Metab* 2003; **88**: 1119–1125.
- 58 Neely EK, Bachrach LK. Turner Syndrome Adolescents Hormone Are Not Osteopenic\*. 1993.
- 59 Elsheikh M, Bird R, Casadei B, Conway GS, Wass JAH. The Effect of Hormone Replacement Therapy on Cardiovascular Hemodynamics in Women with Turner's Syndrome. *J Clin Endocrinol Metab* 2000; **85**: 614–618.
- 60 Gawlik A, Malecka-Tendera E. Treatment of Turner's syndrome during transition. *Eur J Endocrinol* 2014; **170**. doi:10.1530/EJE-13-0900.
- 61 Webb G, Gatzoulis MA. Atrial septal defects in the adult: Recent progress and overview. *Circulation* 2006; **114**: 1645–1653.
- 62 Park MK (Myung K. Park's pediatric cardiology for practitioners. 2008, p 676.
- 63 Flynn JT, Kaelber DC, Baker-Smith CM. Clinical Practice Guideline for Screening and Management of High Blood Pressure in Children and Adolescents. *Pediatrics* 2017; **140**. doi:10.1542/peds.2017-1904.
- 64 Kubota T, Wakui K, Nakamura T, Ohashi H, Watanabe Y, Yoshino M *et al.* The proportion of cells with functional X disomy is associated with the severity of mental retardation in mosaic ring X Turner syndrome females. *Cytogenet Genome Res* 2002; **99**: 276–284.
- 65 Carvalho AB, Guerra Júnior G, Baptista MTM, de Faria APM, Marini SHV de L, Guerra ATM. Cardiovascular and renal anomalies in Turner syndrome. *Rev Assoc Med Bras* 1992; **56**: 655–9.
- 66 Elsheikh M, Wass JAH, Conway GS. Autoimmune Thyroid Syndrome in Women With Turner's Syndrome—The Association With Karyotype. *Obstet Gynecol Surv* 2002; **57**: 31–32.
- 67 Binder G, Koch A, Wajs E, Ranke MB. Nested polymerase chain reaction study of 53 cases with Turner's syndrome: Is cytogenetically undetected Y mosaicism common? *J Clin Endocrinol Metab* 1995; **80**: 3532–3536.
- 68 Navot D, Laufer N, Kopolovic J, Rabinowitz R, Birkenfeld A, Lewin A *et al.* Artificially Induced Endometrial Cycles and Establishment of Pregnancies in the Absence of Ovaries. *N Engl J Med* 1986; **314**: 806–811.
- 69 Pulungan AB. Pubertas Normal dan Gangguannya. In: Batubara JR, Tridjaja B, Pulungan AB (eds). *Buku Ajar Endokrinologi Anak Edisi kedua*. Badan Penerbit Ikatan Dokter Anak Indonesia: Jakarta, 2018, pp 92–101.
- 70 Yang MC, Wu JR. Recent review of transcatheter closure of atrial septal defect. *Kaohsiung J. Med. Sci.* 2018; **34**: 363–369.
- 71 Corrado C. Blood pressure measurement in children. *Ital J Pediatr* 2015; **41**: A19.
- 72 Faienza MF, Ventura A, Colucci S, Cavallo L, Grano M, Brunetti G. Bone fragility in turner syndrome: Mechanisms and prevention strategies. *Front Endocrinol (Lausanne)* 2016; **7**: 1–8.
- 73 Muntaj S, Ganaie FA, Purva S V, Radhika S, Tilak P. Karyotypic Variables in Turner Syndrome : A Case Series. *Int J Sci Study* 2015; **3**: 171–175.
- 74 Zhong Q, Layman LC. Genetic considerations in the patient with Turner

- syndrome—45,X with or without mosaicism. *Fertil Steril* 2012; **98**: 775–779.
- 75 Al Alwan I, M K, Amir, G N, A O, L B *et al.* Turner Syndrome Genotype and phenotype and their effect on presenting features and timing of Diagnosis. *Int J Health Sci (Qassim)* 2014; **8**: 195–202.
  - 76 Ostberg JE, Thomas EL, Hamilton G, Attar MJH, Bell JD, Conway GS. Excess visceral and hepatic adipose tissue in Turner syndrome determined by magnetic resonance imaging: Estrogen deficiency associated with hepatic adipose content. *J Clin Endocrinol Metab* 2005; **90**: 2631–2635.
  - 77 Reubinoff BE, Grubstein A, Meirow D, Berry E, Schenker JG, Brzezinski A. Effects of low-dose estrogen oral contraceptives on weight, body composition, and fat distribution in young women. *Fertil Steril* 1995; **63**: 516–521.
  - 78 Ismail N, Metwaly NE, El-Moguy F, Hafez M, Abd El Dayem S, Farid T. Bone age is the best predictor of growth response to recombinant human growth hormone in Turner's syndrome. *Indian J Hum Genet* 2010; **16**: 119.
  - 79 Kavoussi SK, Christman GM, Smith YR. Healthcare for Adolescents with Turner Syndrome. *J. Pediatr. Adolesc. Gynecol.* 2006; **19**: 257–265.
  - 80 Saenger P. Transition in Turner's syndrome. *Growth Horm IGF Res* 2004; **14 Suppl A**: S72–S76.
  - 81 Paterson WF, Hollman AS, Donaldson MDC. Poor uterine development in Turner syndrome with oral oestrogen therapy. *Clin Endocrinol (Oxf)* 2002; **56**: 359–365.
  - 82 Frias JL, Davenport ML. Health Supervision for Children With Turner Syndrome. *Pediatrics* 2003; **111**: 692–702.
  - 83 Lee MC, Conway GS. Liver dysfunction in Turner syndrome and its relationship to exogenous oestrogen. *Eur J Gastroenterol Hepatol* 2013; **25**: 1141–1145.
  - 84 Hanew K, Tanaka T, Horikawa R, Hasegawa T, Fujita K, Yokoya S. Women with Turner syndrome are at high risk of lifestyle-related disease -From questionnaire surveys by the Foundation for Growth Science in Japan. *Endocr J* 2016; **63**: 449–456.
  - 85 S. Mannu G, J.S. Zaman M, Gupta A, U. Rehman H, K. Myint P. Evidence of Lifestyle Modification in the Management of Hypercholesterolemia. *Curr Cardiol Rev* 2013; **9**: 2–14.
  - 86 Alencia ED V, Erna LINAMAS, Etancur LINAMAB, Edina CAMAM, Rango ÁLA. Lipid profile in a group of patients with Turner ' s syndrome at Clínica Universitaria Bolivariana in the Medellín city between 2000 and 2009 \* Colombia Médica To date , there are no studies in the city of Medellín of 21 women with Turner Syndrome , who h. 2011; **42**: 54–60.
  - 87 Cooley M. Lipid Profiles in Women With 45,X vs 46,XX Primary Ovarian Failure. *JAMA J Am Med Assoc* 2003; **290**: 2127–2128.