



DAFTAR PUSTAKA

- Al-Baradie R, Yamada K, St.Hilaire C, Chan W-M, Andrews C, McIntosh N, Nakano M, Martonyi EJ, Raymond WR, Okumura S, Okihiro MM, Engle EC. Duane radial ray ayndrome (Okihiro syndrome) maps to 20q13 and results from mutations in *SALL4*, a new member of the SAL family. *Am J Hum Genet*, 2002;71:1195-1199.
- Arias S, Penchaszadeh VB, Pinto-Cisternas J, Larrauri S. The IVIC syndrome: a new autosomal dominant complex pleiotropic syndrome with radial ray hypoplasia, hearing impairment, external ophthalmoplegia, and thrombocytopenia. *Am J Med Genet*, 1980;6:25-59.
- Auerbach AD. Fanconi anemia and its diagnosis. *Mutat Res*. 2009;668(1-2):4-10.
- Barham G, Clarke NMP. Genetic regulation of embryological limb development with relation to congenital limb deformity in humans. *J Child Orthop*, 2008;2:1-9.
- Czeizel A, Goblyos P, Kodaj I. IVIC syndrome: report of a third family. *Am J Med Genet*, 1989;32:282-283.
- Dufour C. How I manage patients with Fanconi anaemia. *Br J Haematol*. 2017;178(1):32-47.
- Giri N, Batista DL, Alter BP, Stratakis CA. Endocrine abnormalities in patients with Fanconi anemia. *J Clin Endocrinol Metab*. 2007;92(7):2624-2631.
- Glanz A, Fraser C. Spectrum anomalies in Fanconi anemia. *J Med Genet*, 1982;19:412-416.
- Hays L, Frohnmayer D, Guinan E, Kennedy T, Larsen K. *Fanconi Anemia: Guidelines for Diagnosis and Management 4th ed*. 2014, Fanconi Anemia Research Fund Inc.
- James MA, Green HD, McCarroll R, Manske PR. The association of radial deficiency with thumb hypoplasia. *J Bone Joint Surg*, 2004;86A(10):2196-2205.
- Johnson CP, Blasco PA. Infant growth and development. *Ped Rev*, 1997;18(7):224-242.
- Jones KJ (ed.) *Smith's Recognizable Pattern of Human Malformation 6th ed*. 2005, Saunders, New York.
- McDonald R, Goldschmidt B. Pancytopenia with Congenital Defects (Fanconi's Anaemia). *Arch Dis Child*. 1960;35(182):367-372.
- Pakkasjarvi N, Koskimies E, Ritvanen A, Nietosvaara Y, Makitie O. Characteristics and associated anomalies in radial ray deficiencies in Finland – a population-based study. *Am J Med Genet Part A*, 2013;161A:261-267.
- Paradisi I, Arias S. IVIC syndrome is caused by a c.2607delA mutation in the *SALL4* locus. *Am J Med Genet Part A*, 2007;143A:326-332.



- Sammito V, Motta G, Capodieci S, Sanfilippo S, Neri G. IVIC syndrome: report of a second family. *Am J Med Genet*, 1988;29:875-881.
- Stivaros SM, Alston R, Wright NB, et al. Central nervous system abnormalities in Fanconi anaemia: patterns and frequency on magnetic resonance imaging. *Br J Radiol*. 2015;88(1056):20150088.
- Stivaros SM, Punekar M, Chandler K, Rost I, Schindler D, Meyer S. Pollicization of the index finger in Fanconi anaemia: appearances and functionality 40 years after the intervention. *Br J Haematol*. 2014;166(6):807.
- Tischkowitz MD, Hodgson SV. Fanconi Anemia. *J Med Genet* 2003;40:1-10.