

8. Bibliografia

8. Bibliografia

Abuelo, D.N.; Tint, G.S.; Kelley, R.; Batta, A.K.; Shefer, S.; Salen, G. (1995). "Prenatal detection of the cholesterol biosynthetic defect in the Smith-Lemli-Opitz syndrome by the analysis of amniotic fluid sterols." *Am. J. Med- Genet.*; 56 (3): 281-285.

Ahmida, H.S.M; Bertucci, P.; Franzò, L.; Massoud, R.; Cortese, C.; Lala, A.; Federici, G. (2006). "Simultaneous determination of plasmatic phytosterols and cholesterol precursors using gas chromatography/mass spectrometry (GC-MS) with selective ion monitoring (SIM)." *Journal of Chromatography B*; 842: 43 - 47.

Amâncio, F.A.M.; Scalco, F.B.; Coelho, C.A.R. (2007). "Investigação diagnóstica de erros inatos do metabolismo em um hospital universitário." *J. Bras. Patol. Med. Lab.*; 43 (3): 169 - 174.

Araújo, A.P.Q.C. (2004). "Doenças metabólicas com manifestações psiquiátricas." *Rev. Psiq. Clin.*; 31: 285 - 289.

Batta, K.A.; Tint, G.S.; Shefer, S.; Abuelo, D.; Salen, G. (1995). "Identification of 8-dehydrocholesterol (cholesta-5,8-dien-3 β -ol) in patients with Smith-Lemli-Opitz syndrome." *J. of Lipid Research*; 36: 705 - 713.

Baric, I.; Fumic, K.; Hoffmann, G.F. (2001). "Inborn Errors of Metabolism at the turn of the Millennium." *Croat. Med. J.*; 42: 379 - 383.

Berg, J.M., *et al.* *Biochemistry*, 5.º ed. 2002; New York, W.H. Freeman.

Cardoso, M. L.; Balreira, A.; Martins, M.; Nunes, L.; Cabral, A.; Marques, M; Lima, M.R.; Marques, J.S.; Medeira, A.; Cordeiro, I.; Pedro, S.; Mota, M.C.; Dionisi-Vici, C.; Santorelli, F.M.; Jakobs, C.; Clayton, P.T. e Vilarinho, L. (2005). "Molecular studies in Portuguese patients with Smith-Lemli-Opitz syndrome and report of three new mutations in *DHCR7*." *Mol. Genet. Metab.*

Cardoso, M. L.; Fortuna, A. M.; Castedo, S.; Martins, M.; Montenegro, N.; Jakobs, C.; Clayton, P.; Vilarinho, L. (2005). "Diagnóstico Pré-Natal de Síndrome de Smith-Lemli-Opitz." *ArquiMed.*

Chevy, F.; Humbert, L.; Wolf, C. (2005). "Sterol profiling of amniotic fluid: a routine method for the detection of distal cholesterol synthesis deficit." *Prenatal Diagnosis*; 25: 1000-1006.

Clayton, P.T. (1998). "Disorders of cholesterol biosynthesis." *Arch. Dis. Child*; **78**: 185 – 189.

Correa-Cerro, L.S.; Porter, F.D. (2005). “ 3β -Hydroxysterol Δ^7 -reductase and the Smith–Lemli–Opitz syndrome.” *Mol. Genet. Metab.*

Dallaire, L.; Mitchell, G.; Giguère, R.; Lefebvre, F.; Melançon S.B.; Lambert, M. (1995). “Prenatal diagnosis of Smith-Lemli-Opitz syndrome is possible by measurement of 7-dehydrocholesterol in amniotic fluid.” *Prenatal Diagnosis*; 15 (9): 855 - 858.

Danzer, E.; Schier, F.; Giggel, S.; Bondartschuk, M. (2000). “Smith-Lemli-Opitz Syndrome: Case Report and Literature Review.” *Journal of Pediatric Surgery*; 35: 1840 - 1842

Dehart, D.B.; Lanoue, L.; Tint, G.S.; Sulik, K.K. (1997). “Pathogenesis of malformations in a rodent model for Smith-Lemli-Opitz syndrome.” *Am. J. Med. Genet.*; **68**: 328 - 337.

Fernades, J.O. (1993). “Cromatografia gasosa/ espectrometria de massa – aplicação da técnica na análise de vinhos.” *Relatório para efeito de realização de Provas de Aptidão Pedagógica e Capacidade científica*, Faculdade de Farmácia da Universidade do Porto.

Fitzky, *et al.* (2001). “7-Dehydrocholesterol-dependent proteolysis of HMG-CoA reductase suppresses sterol biosynthesis in a mouse model of Smith-Lemli-Opitz/RSH syndrome”. *J. Clin. Invest.* **108**: 905 - 915.

Gondré-Lewis, M.C.; Petrache, H.I.; Wassif, C.A.; Harries, D.; Parsegian, A.; Porter, F.D.; Loh, Y.P. (2006). “Abnormal sterols in cholesterol-deficiency diseases cause secretory granule malformation and decreased membrane curvature.” *Journal of Cell Science*; 119: 1876 - 1885

Hass, D.; Armbrust, S.; Hass, J.-P.; Zschocke, J.; Muhlmann, K.; Fusch, C.; Neumann, L.M. (2005). “Smith-Lemli-Opitz síndrome with a classical phenotype, oesophageal achalasia and borderline plasma sterol concentrations.” *J. Inherit. Metab. Dis*; 28: 1191 - 1196.

Herman, G.E. (2003). “Disorders of cholesterol biosynthesis: prototypic metabolic malformation Syndromes.” *Human Molecular Genetics*; 12: 75 - 88

Husny, A.S.E.; Fernandes-Caldato, M.C. (2006). “Erros Inatos do Metabolismo: revisão da literatura.” *Revista Paraense de Medicina*; 20: 41 - 45.

Irons, M.B.; Tint, G.S. (1998). “Prenatal diagnosis of Smith-Lemli-Opitz syndrome.” *Prenatal Diagnosis*; 18: 369 - 372.

Karasek, F.W.; Ray, E.C. *Basic Gas Chromatography-Mass Spectrometry: Principles & Techniques*, 1988; Amsterdam, Elsevier.

- Kelley**, R.I. (1995). "Diagnosis of Smith-Lemli-Opitz syndrome by gas chromatography/mass spectrometry of 7-dehydrocholesterol in plasma, amniotic fluid and cultured skin fibroblasts." *Clinica Chimica Acta*; 236: 45 - 58.
- Kelley**, R.I. and Hennekam, R.C.M. (2000). "The Smith-Lemli-Opitz syndrome"; *J. Med. Genet.*; 37: 321 - 335
- Koide**, T.; Hayata, T.; Cho, K.W.Y. (2006). "Negative regulation of Hedgehog signalling by the cholesterologenic enzyme 7-dehydrocholesterol reductase." *Development and Disease*; 133: 2395 - 2405
- Kratz**, E.L.; Kelley, R.I. (1999). "Prenatal diagnosis of the RSH/Smith-Lemli-Opitz syndrome." *American Journal of Medical Genetics*; 82: 376 - 381.
- Marroco**, J. *Análise estatística com utilização do SPSS*, 3.º ed. 2007; Lisboa, Edições Sílabo, LDA.
- Mills**, K.; Mandel, H.; Montemagno, R.; Soothill, P.; Gershoni-Baruch, R.; Clayton, P.T. (1996). "First trimester prenatal diagnosis of Smith-Lemli-Opitz syndrome (7-dehydrocholesterol reductase deficiency)." *Pediatric Research*; 39 (5): 816 - 819.
- Moebius**, F.F.; Fitzky, B.U.; Glossmann, H. (2000). "Genetic Defects in Postsqualene Cholesterol Biosynthesis." *TEM*; 11: 106 - 114.
- Murray**, K.R.; Granner, D.K.; Mayes, P.A.; Rodwell, V.W., *Harper's Illustrated Biochemistry*, 26.º ed., 2003, McGraw-Hill Companies.
- Natowicz**, M.R.; Evans, J.E. (1994). "Abnormal bile acids in the Smith-Lemli-Opitz syndrome." *Am J Med Genet*; 50: 364 - 7.
- Nelson**, D.L.; Cox, M.M. *Principles of Biochemistry*, 4.º ed. 2005; New York, W.H., Freeman and Company.
- Nissinen**, M.J.; Gylling, H.; Kaski, M.; Tammisto, P. (2000). "Smith-Lemli-Opitz syndrome and other sterol disorders among Finns with developmental disabilities." *J. Lab. Clin. Med.*; 136: 457 - 467.
- Nowaczyk**, M.J.; Heshka, T.; Kratz, L.E.; Kelley, R.E. (2000). "Difficult prenatal diagnosis in mild Smith-Lemli-Opitz syndrome." *Am. J. Med. Genet.*; 95 (4): 396 - 398.
- Nwokoro**, N.A.; Wassif, C.A.; Porter, F.D. (2001). "Genetic Disorders of Cholesterol Biosynthesis in Mice and Humans." *Mol. Genetics and Metabolism*; 74: 105 - 119.

Pasquali, M; Monsen, G.; Richardson, L.; Alston, M.; Longo, N. (2006), "Biochemical findings in common Inborn Errors of Metabolism." *Am. J. Med. Genet. Part C., Semin. Med. Genet.*; 142C: 64 - 76.

Pereira, J.L.G.F.S.C. (2006). "Controlo de qualidade: diagnóstico estatístico de processos por estimativas directas." *Química*; 101: 41 - 43.

Porter, F.D. (2002). "Malformation syndromes due to inborn errors of cholesterol synthesis." *The Journal of Clinical Investigation*; 110: 715 - 724.

Porter, F. D. (2006). "Cholesterol precursors and facial clefting." *The Journal of Clinical Investigation*; 116: 2322 -2325.

Porter, F.D. (2008). "Smith-Lemli-Opitz syndrome: pathogenesis, diagnosis and management." *European Journal of Human Genetics*; 16: 535 - 541.

Raghuveer, T.S.; Garg, U.; Graf, W.D. (2006). "Inborn Errors of Metabolism in infancy and early childhood: an update." *American Family Physician*; 73: 1981 - 1990.

Salen, G.; Shefer, S.; Batta, A.K.; Tint, G. S.; Xu, G.; Honda, A.; Irons, M.; Elias, E. R. (1996). "Abnormal cholesterol biosynthesis in the Smith-Lemli-Opitz syndrome." *Journal of Lipid Research*; 37: 1169 - 1180.

Saudubray, J.M.; Nassogne, M.C.; Lonlay, P.; Touati, G. (2002). "Clinical approach to inherited metabolic disorders in neonates: an overview." *Semin. Neonatol.*; 7: 3 - 15.

Saudubray, J.M.; Sedel, F.; Walter, J.H. (2006). "Clinical approach to treatable inborn metabolic diseases: an introduction." *J. Inherit. Metab. Dis.*; 29: 261 - 274.

Shefer, S.; Salen, G.; Honda, A.; Batta, A.K.; Nguyen, L.B.; Tint, G.S.; Ioannou, Y.A. e Desnick, R. (1998) "Regulation of rat hepatic 3β -hydroxysterol Δ^7 -reductase: substrate specificity, competitive and non-competitive inhibition, and phosphorylation/dephosphorylation." *Journal of Lipid Research*; 39: 2471 - 2476.

Solca, C.; Pandit, B.; Yu, H.; Tint, G. S.; Patel, S. B. (2007). "Loss of apolipoprotein E exacerbates the neonatal lethality of the Smith-Lemli-Opitz syndrome mouse." *Mol. Genetics and Metabolism*; 91: 7 - 14.

Wassif et al. (2002). "Cholesterol storage defect in RSH/Smith-Lemli-Opitz syndrome fibroblasts." *Mol. Genet. Metab.* 75: 325 - 334.

Tanaka, K.; Hine, D.G.; West-Dull, A. (1980). "Gas-chromatographic method of analysis of urinary organic acids." *Clin. Chem.*; 26: 1839 - 46.

Tint, G.S., Abuelo, D.; Till, M.; Cordier, M.P.; Batta, A.K.; Shefer, S.; Honda, A.; Honda, M.; Xu, G.; Irons, M.; Elias, E.R.; Salen, G. (1998). "Fetal Smith-Lemli-Opitz syndrome can be detected accurately and reliably by measuring amniotic fluid dehydrocholesterols." *Prenatal Diagnosis*; 18: 651 - 658.

Waterham, H.R.; Wanders, R.J.A. (2000). "Biochemical and genetics aspects of 7-dehydrocholesterol reductase and Smith- Lemli-Opitz syndrome." *Biochimica et Biophysica Acta*; 1529: 340 - 356.

Waterham, H.R. (2006). "Defects of cholesterol biosynthesis." *FEBS Letters*; 580: 5442 - 5449.

Wilson, K.; Walker, J. M. *Principles and techniques of practical biochemistry*, 4.º ed. 1999; Cambridge University Press.

Witsch-Baumgartner, M.; Gruber, M.; Kraft, H. G.; Rossi, M.; Clayton, P.; Giros, M.; Haas, D.; Kelley, R. I.; Krajewska-Walasek, M.; Utermann, G. (2004). "Maternal apo E genotype is a modifier of the Smith-Lemli-Opitz syndrome." *J. Med. Genet.*; 41: 577 - 584.

Yu, H. and Patel, S.B. (2005). "Recent insights into the Smith-Lemli-Opitz syndrome." *Clin. Genet.*; 68: 383 - 391.

www.uma.pt/igc/iGC.html. Acesso no dia 16/5/08.

<http://www.forumsci.co.il:80/HPLC/html>. Acesso no dia 10/07/2007.