

Ernährungstherapie bei angeborenen Stoffwechselerkrankungen

Teil 3: Störungen des Fettstoffwechsels

Ulrike Och, Tobias Fischer, Thorsten Marquardt

Literatur

- Iqbal J, Hussain MM: Intestinal lipid absorption. *Am J Physiol Endocrinol Metab* 2009; 296(6): E1183–94.
- Alves-Bezerra M, Cohen DE: Triglyceride Metabolism in the Liver. *Compr Physiol* 2017; 8(1): 1–8.
- Ramasamy I: Recent advances in physiological lipoprotein metabolism. *Clin Chem Lab Med* 2014; 52(12): 1695–727.
- Adeva-Andany MM, Carneiro-Freire N, Seco-Filgueira M, Fernández-Fernández C, Mourinho-Bayolo D: Mitochondrial β -oxidation of saturated fatty acids in humans. *Mitochondrion* 2019; 46: 73–90.
- Kumari A: Beta Oxidation of Fatty Acids. In: *Sweet Biochemistry*: Elsevier 2018; 17–19.
- Wanders RJ, Vreken P, den Boer ME, Wijburg FA, van Gennip AH, IJlst L: Disorders of mitochondrial fatty acyl-CoA beta-oxidation. *J Inher Metab Dis* 1999; 22(4): 442–87.
- Kim J-JP, Miura R: Acyl-CoA dehydrogenases and acyl-CoA oxidases. Structural basis for mechanistic similarities and differences. *Eur J Biochem* 2004; 271(3): 483–93.
- Institute of Medicine (IOM): Dietary Reference Intakes for Energy, Carbohydrate, Fiber, Fat, Fatty Acids, Cholesterol, Protein, and Amino Acids. Washington, D.C.: National Academies Press 2005.
- Deutsche Gesellschaft für Ernährung, Österreichische Gesellschaft für Ernährung, Schweizerische Gesellschaft für Ernährungsforschung, Schweizerische Vereinigung für Ernährung (ed.): Referenzwerte für die Nährstoffzufuhr. 2nd ed. Bonn: Deutsche Gesellschaft für Ernährung 2019.
- Shaw V (ed.): *Clinical Paediatric Dietetics*: Hoboken, New Jersey: Wiley 2020.
- Gillingham MB, Scott B, Elliott D, Harding CO: Metabolic control during exercise with and without medium-chain triglycerides (MCT) in children with long-chain 3-hydroxy acyl-CoA dehydrogenase (LCHAD) or trifunctional protein (TFP) deficiency. *Mol Genet Metab* 2006; 89(1–2): 58–63.
- Marten B, Pfeuffer M, Schrezenmeir J: Medium-chain triglycerides. *International Dairy Journal* 2006; 16(11): 1374–82.
- Thevenet J, Marchi U de, Domingo JS, et al.: Medium-chain fatty acids inhibit mitochondrial metabolism in astrocytes promoting astrocyte-neuron lactate and ketone body shuttle systems. *FASEB J* 2016; 30(5): 1913–26.
- Romano A, Koczwara JB, Gallelli CA, et al.: Fats for thoughts: An update on brain fatty acid metabolism. *Int J Biochem Cell Biol* 2017; 84: 40–5.
- Vockley J, Burton B, Berry G, et al.: Effects of triheptanoin (UX007) in patients with long-chain fatty acid oxidation disorders: Results from an open-label, long-term extension study. *J Inher Metab Dis* 2020.
- Deutsches Institut für Medizinische Dokumentation und Information (DIMDI): Internationale statistische Klassifikation der Krankheiten und verwandter Gesundheitsprobleme ICD-10-GM: 10. Revision, German Modification 2020.
- Nennstiel U, Lüders A, Blankenstein O, et al.: Nationaler Screeningreport Deutschland 2017. Deutsche Gesellschaft für Neugeborenen-Screening e. V. (DGNS) 2019: 1–47.
- Merritt JL 2, Chang IJ: Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency. In: Merritt JL 2, Chang IJ (eds.): *GeneReviews®* [Internet]: University of Washington, Seattle 2019.
- Derks TGJ, van Spronsen FJ, Rake JP, van der Hilst CS, Span MM, Smit GPA: Safe and unsafe duration of fasting for children with MCAD deficiency. *Eur J Pediatr* 2007; 166(1): 5–11.
- El-Gharbawy A, Vockley J: Inborn errors of metabolism with myopathy: defects of fatty acid oxidation and the carnitine shuttle system. *Pediatr Clin North Am* 2018; 65(2): 317–35.
- Schaub J, Schulte F-J, Hoffmann GF, Lentze MJ, Spranger J, Zepp F: *Pädiatrie: Grundlagen und Praxis*. 4th ed. Berlin, Heidelberg: Springer 2014.
- Zschocke J, Hoffmann GF: *Vademecum metabolicum: diagnosis and treatment of inborn errors of metabolism*. Foreword by William L. Nyhan, San Diego, USA. 4th ed. s.l.: Schattauer GmbH Verlag für Medizin und Naturwissenschaften 2011.
- Primassin S, Ter Veld F, Mayatepek E, Spiekeroetter U: Carnitine supplementation induces acylcarnitine production in tissues of very long-chain acyl-CoA dehydrogenase-deficient mice, without replenishing low free carnitine. *Pediatr Res* 2008; 63(6): 632–7.
- Gregersen N, Andresen BS, Corydon MJ, et al.: Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship. *Hum Mutat* 2001; 18(3): 169–89.
- Almannai M, Alfadhel M, El-Hattab AW: Carnitine Inborn Errors of Metabolism. *Molecules* 2019; 24(18).
- Bennett MJ, Santani AB (eds.): *GeneReviews®* [Internet]: University of Washington, Seattle 2016.
- Magoulas PL, El-Hattab AW: Systemic primary carnitine deficiency: an overview of clinical manifestations, diagnosis, and management. *Orphanet J Rare Dis* 2012; 7: 68.
- Scaglia F, Wang Y, Singh RH, et al.: Defective urinary carnitine transport in heterozygotes for primary carnitine deficiency. *Genet Med* 1998; 1(1): 34–9.
- Schulze A, Lindner M, Kohlmüller D, Olgemöller K, Mayatepek E, Hoffmann GF: Expanded newborn screening for inborn errors of metabolism by electrospray ionization-tandem mass spectrometry: results, outcome, and implications. *Pediatrics* 2003; 111(6 Pt 1): 1399–406.
- Przyrembel H, Wendel U, Becker K, et al.: Glutaric aciduria type II: Report on a previously undescribed metabolic disorder. *Clinica Chimica Acta* 1976; 66(2): 227–39.

31. Och U, Fischer T, Marquardt T: Ernährungstherapie bei angeborenen Stoffwechselerkrankungen: Teil 2: Störungen des Aminosäurestoffwechsels. *Ernährungs Umschau* 2020; 67(10): M606–19.
32. al-Essa MA, Rashed MS, Bakheet SM, Patay ZJ, Ozand PT: Glutaric aciduria type II: observations in seven patients with neonatal- and late-onset disease. *Journal of perinatology official journal of the California Perinatal Association* 2000; 20(2): 120–8.
33. Angle B, Burton BK: Risk of sudden death and acute life-threatening events in patients with glutaric acidemia type II. *Mol Genet Metab* 2008; 93(1): 36–9.
34. Schiff M, Froissart R, Olsen RKJ, Acquaviva C, Vianey-Saban C: Electron transfer flavoprotein deficiency: functional and molecular aspects. *Mol Genet Metab* 2006; 88(2): 153–8.
35. Frerman FE, Goodman SI: Deficiency of electron transfer flavoprotein or electron transfer flavoprotein: ubiquinone oxidoreductase in glutaric acidemia type II fibroblasts. *Proc Natl Acad Sci U S A* 1985; 82(13): 4517–20.
36. Fischer T, Och U, Marquardt T: Long-term ketone body therapy of severe multiple acyl-CoA dehydrogenase deficiency: A case report. *Nutrition* 2019; 60: 122–8.
37. Goodman SI, Frerman FE: Glutaric acidemia type II (multiple acyl-CoA dehydrogenation deficiency). *J Inher Metab Dis* 1984; 7 Suppl 1: 33–7.
38. Cornelius N, Frerman FE, Corydon TJ, et al.: Molecular mechanisms of riboflavin responsiveness in patients with ETF-QO variations and multiple acyl-CoA dehydrogenation deficiency. *Human molecular genetics* 2012; 21(15): 3435–48.
39. Ghisla S, Thorpe C: Acyl-CoA dehydrogenases. A mechanistic overview. *Eur J Biochem* 2004; 271(3): 494–508.
40. Bonham JR, Tanner MS, Pollit RJ, et al.: Oral sodium-3-hydroxybutyrate, a novel adjunct to treatment for multiple acyl-CoA dehydrogenase deficiency. *J Inher Metab Dis* 1999; 22(suppl): 101.
41. van Hove JLK, Grünewald S, Jaeken J, et al.: D,L-3-hydroxybutyrate treatment of multiple acyl-CoA dehydrogenase deficiency (MADD). *The Lancet* 2003; 361(9367): 1433–5.
42. Gautschi M, Weisstanner C, Slotboom J, Nava E, Zurcher T, Nuoffer J-M: Highly efficient ketone body treatment in multiple acyl-CoA dehydrogenase deficiency-related leukodystrophy. *Pediatr Res* 2015; 77(1–1): 91–8.
43. van Rijjt WJ, Heiner-Fokkema MR, du Marchie Sarvaas, Gideon J, et al.: Favorable outcome after physiologic dose of sodium-D,L-3-hydroxybutyrate in severe MADD. *Pediatrics* 2014; 134(4): e1224–8.
44. van Rijjt WJ, Jager EA, Allersma DP, et al.: Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. *Genet Med* 2020; 22(5): 908–16.
45. Kumari A: Cholesterol Synthesis. In: *Sweet Biochemistry: Elsevier* 2018; 27–31.
46. Walzer S, Travers K, Rieder S, Erazo-Fischer E, Matusiewicz D: Homozygous familial hypercholesterolemia (HoFH) in Germany: an epidemiological survey. *ClinicoEconomics and outcomes research CEOR* 2013; 5: 189–92.
47. Talmud PJ, Shah S, Whittall R, et al.: Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolemia: A case-control study. *The Lancet* 2013; 381(9874): 1293–301.
48. Klose G, Laufs U, März W, Windler E: Familial hypercholesterolemia: developments in diagnosis and treatment. *Deutsches Arzteblatt international* 2014; 111(31–32): 523–9.
49. Mach F, Baigent C, Catapano AL, et al.: 2019 ESC/EAS Guidelines for the management of dyslipidaemias: lipid modification to reduce cardiovascular risk. *European heart journal* 2020; 41(1): 111–88.
50. Wolfram G, Bechthold A, Boeing H, et al.: Evidence-Based Guideline of the German Nutrition Society: Fat Intake and Prevention of Selected Nutrition-Related Diseases. *Ann Nutr Metab* 2015; 67(3): 141–204.
51. Bundesinstitut für Risikobewertung (BfR): Lebensmittel mit Pflanzensterol- und Pflanzenstanol-Zusatz: Bewertung einer neuen Studie aus den Niederlanden: Stellungnahme 006/2012 des BfR vom 1. Dezember 2011, ergänzt am 21. Januar 2013. 2011: 1–7.
52. Cabral CE, Klein MRST: Phytosterols in the treatment of hypercholesterolemia and prevention of cardiovascular diseases. *Arq Bras Cardiol* 2017; 109(5): 475–82.
53. Berger S, Raman G, Vishwanathan R, Jacques PF, Johnson EJ: Dietary cholesterol and cardiovascular disease: a systematic review and meta-analysis. *Am J Clin Nutr* 2015; 102(2): 276–94.
54. Carson JAS, Lichtenstein AH, Anderson CAM, et al.: Dietary Cholesterol and Cardiovascular Risk: A Science Advisory from the American Heart Association. *Circulation* 2020; 141(3): e39–53.
55. Vincent MJ, Allen B, Palacios OM, Haber LT, Maki KC: Meta-regression analysis of the effects of dietary cholesterol intake on LDL and HDL cholesterol. *Am J Clin Nutr* 2019; 109(1): 7–16.
56. Och U: Ernährungstherapie bei Hypercholesterinämie. *Ernährungs Umschau* 2018; 65(11): S73–6.
57. Porter FD, Herman GE: Malformation syndromes caused by disorders of cholesterol synthesis. *J Lipid Res* 2011; 52(1): 6–34.
58. Smith DW, Lemli L, Opitz JM: A newly recognized syndrome of multiple congenital anomalies. *J Pediatr* 1964; 64: 210–7.
59. Tint GS, Irons M, Elias ER, et al.: Defective cholesterol biosynthesis associated with the Smith-Lemli-Opitz syndrome. *N Engl J Med* 1994; 330(2): 107–13.
60. Movassaghi M, Bianconi S, Feinn R, Wassif CA, Porter FD: Vitamin D levels in Smith-Lemli-Opitz syndrome. *Am J Med Genet A* 2017; 173(10): 2577–83.
61. Porter FD: Smith-Lemli-Opitz syndrome: pathogenesis, diagnosis and management. *Eur J Hum Genet* 2008; 16(5): 535–41.
62. Sever N, Mann RK, Xu L, et al.: Endogenous B-ring oxysterols inhibit the Hedgehog component Smoothed in a manner distinct from cyclopamine or side-chain oxysterols. *Proc Natl Acad Sci U S A* 2016; 113(21).
63. Eroglu Y, Nguyen-Driver M, Steiner RD, et al.: Normal IQ is possible in Smith-Lemli-Opitz syndrome. *Am J Med Genet A* 2017; 173(8): 2097–100.
64. Kritchevsky D, Tepper SA: Solubility of cholesterol in various fats and oils. *Proc Soc Exp Biol Med* 1964; 116: 104–7.
65. Saad HY, Higuchi WI: Water solubility of cholesterol. *J Pharm Sci* 1965; 54(8): 1205–6.
66. Fischer T, Dütting S, Och U, Baumeister A, Marquardt T: Entwicklung einer Cholesterol-Emulsion zur ernährungstherapeutischen Anwendung bei Patienten mit Smith-Lemli-Opitz-Syndrom (SLO). In: *Ernährung 2018 – Ernährung ist Therapie und Prävention*. Stuttgart: Thieme 2018.
67. Fischer T, Dütting S, Och U, Baumeister A, Marquardt T: Sensorische und ernährungsphysiologische Adaption einer Cholesterol-Emulsion zum Einsatz bei Patienten mit Smith-Lemli-Opitz-Syndrom (SLO). In: *Ernährung 2018 – Ernährung ist Therapie und Prävention*. Stuttgart: Thieme 2018.

DOI: 10.4455/eu.2021.006