

1: Huemer M, Diodato D, Martinelli D, Olivieri G, Blom H, Gleich F, Kölker S, Kožich V, Morris AA, Seifert B, Froese DS, Baumgartner MR, Dionisi-Vici C; EHOD consortium, Martin CA, Baethmann M, Ballhausen D, Blasco-Alonso J, Boy N, Bueno M, Burgos Peláez R, Cerone R, Chabrol B, Chapman KA, Couce ML, Crushell E, Dalmau Serra J, Diogo L, Ficicioglu C, García Jimenez MC, García Silva MT, Gaspar AM, Gautschi M, González-Lamuño D, Gouveia S, Grünewald S, Hendriksz C, Janssen MCH, Jesina P, Koch J, Konstantopoulou V, Lavigne C, Lund AM, Martins EG, Meavilla Olivas S, Mention K, Mochel F, Mundy H, Murphy E, Paquay S, Pedrón-Giner C, Ruiz Gómez MA, Santra S, Schiff M, Schwartz IV, Scholl-Bürgi S, Servais A, Skouma A, Tran C, Vives Piñera I, Walter J, Weisfeld-Adams J. **Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry.** *J Inher Metab Dis.* 2019 Mar;42(2):333-352. doi: 10.1002/jimd.12041. Epub 2019 Feb 17. PubMed PMID: 30773687.

2: Molema F, Gleich F, Burgard P, van der Ploeg AT, Summar ML, Chapman KA, Lund AM, Rizopoulos D, Kölker S, Williams M; Additional individual contributors from E-IMD. **Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment.** *Mol Genet Metab.* 2019 Feb 25. pii: S1096-7192(18)30658-9. doi: 10.1016/j.ymgme.2019.02.003. [Epub ahead of print] PubMed PMID: 30827756.

3: Molema F, Gleich F, Burgard P, van der Ploeg AT, Summar ML, Chapman KA, Barić I, Lund AM, Kölker S, Williams M; Additional individual contributors from E-IMD. **Evaluation of dietary treatment and amino acid supplementation in organic acidurias and urea-cycle disorders: On the basis of information from a European multicenter registry.** *J Inher Metab Dis.* 2019 Feb 8. doi: 10.1002/jimd.12066. [Epub ahead of print] PubMed PMID: 30734935.

4: Keller R, Chrastina P, Pavlíková M, Gouveia S, Ribes A, Kölker S, Blom HJ, Baumgartner MR, Bártl J, Dionisi-Vici C, Gleich F, Morris AA, Kožich V, Huemer M; and individual contributors of the European Network and Registry for Homocystinurias and Methylation Defects (E-HOD), Barić I, Ben-Omran T, Blasco-Alonso J, Bueno Delgado MA, Carducci C, Cassanello M, Cerone R, Couce ML, Crushell E, Delgado Pecellin C, Dulin E, Espada M, Ferino G, Fingerhut R, Garcia Jimenez I, Gonzalez Gallego I, González-Irazabal Y, Gramer G, Juan Fita MJ, Karg E, Klein J, Konstantopoulou V, la Marca G, Leão Teles E, Leuzzi V, Lilliu F, Lopez RM, Lund AM, Mayne P, Meavilla S, Moat SJ, Okun JG, Pasquini E, Pedron-Giner CC, Racz GZ, Ruiz Gomez MA, Vilarinho L, Yahyaoui R, Zerjav Tansek M, Zetterström RH, Zeyda M. **Newborn screening for homocystinurias: Recent recommendations versus current practice.** *J Inher Metab Dis.* 2019 Jan;42(1):128-139. doi: 10.1002/jimd.12034. PubMed PMID: 30740731.

5: Diodato D, Olivieri G, Pro S, Maiorani D, Martinelli D, Deodato F, Taurisano R, Di Capua M, Dionisi-Vici C. **Axonal peripheral neuropathy in propionic acidemia: A severe side effect of long-term metronidazole therapy.** *Neurology.* 2018 Sep 18;91(12):565-567. doi: 10.1212/WNL.0000000000006209. Epub 2018 Aug 17. PubMed PMID: 30120134.

6: Semeraro M, Boenzi S, Carrozzo R, Diodato D, Martinelli D, Olivieri G, Antonetti G, Sacchetti E, Catesini G, Rizzo C, Dionisi-Vici C. **The urinary organic acids profile in single large-scale mitochondrial DNA deletion disorders.** *Clin Chim Acta.* 2018 Jun;481:156-160. doi: 10.1016/j.cca.2018.03.002. Epub 2018 Mar 10. PubMed PMID: 29534959.

7: Pinto A, Adams S, Ahring K, Allen H, Almeida MF, Garcia-Arenas D, Arslan N, Assoun M, Atik Altınok Y, Barrio-Carreras D, Belanger Quintana A, Bernabei SM, Bontemps C, Boyle F, Bruni G, Bueno-Delgado M, Caine G, Carvalho R, Chrobot A, Chyž K, Cochrane B, Correia C, Corthouts K, Daly A, De Leo S, Desloovere A, De Meyer A, De Theux A, Didycz B, Dijsselhof ME, Dokoupil K, Drabik J, Dunlop C, Eberle-Pelloth W, Efring

K, Ekengren J, Errekalde I, Evans S, Foucart A, Fokkema L, François L, French M, Forssell E, Gingell C, Gonçalves C, Gökmen Özel H, Grimsley A, Gugelmo G, Gyüre E, Heller C, Hensler R, Jardim I, Joost C, Jörg-Streller M, Jouault C, Jung A, Kanthe M, Koç N, Kok IL, Kozanoğlu T, Kumru B, Lang F, Lang K, Liegeois I, Liguori A, Lilje R, Lubina O, Manta-Vogli P, Mayr D, Meneses C, Newby C, Meyer U, Mexia S, Nicol C, Och U, Olivas SM, Pedrón-Giner C, Pereira R, Plutowska-Hoffmann K, Purves J, Re Dionigi A, Reinson K, Robert M, Robertson L, Rocha JC, Rohde C, Rosenbaum-Fabian S, Rossi A, Ruiz M, Saligova J, Gutiérrez-Sánchez A, Schlune A, Schulpis K, Serrano-Nieto J, Skarpalezou A, Skeath R, Slabbert A, Straczek K, Gizewska M, Terry A, Thom R, Tooke A, Tuokkola J, van Dam E, van den Hurk TAM, van der Ploeg EMC, Vande Kerckhove K, Van Driessche M, van Wegberg AMJ, van Wyk K, Vasconcelos C, Velez García V, Wildgoose J, Winkler T, Żółkowska J, Zuvadelli J, MacDonald A. **Early feeding practices in infants with phenylketonuria across Europe.** *Mol Genet Metab Rep.* 2018 Aug 8;16:82-89. doi: 10.1016/j.ymgmr.2018.07.008. eCollection 2018 Sep. PubMed PMID: 30101073; PubMed Central PMCID: PMC6082991.

8: Pinto A, Alfadhel M, Akroyd R, Atik Altınok Y, Bernabei SM, Bernstein L, Bruni G, Caine G, Cameron E, Carruthers R, Cochrane B, Daly A, de Boer F, Delaunay S, Dianin A, Dixon M, Drogari E, Dubois S, Evans S, Gribben J, Gugelmo G, Heidenborg C, Hunjan I, Kok IL, Kumru B, Liguori A, Mayr D, Megdad E, Meyer U, Oliveira RB, Pal A, Pozzoli A, Pretese R, Rocha JC, Rosenbaum-Fabian S, Serrano-Nieto J, Sjoqvist E, Timmer C, White L, van den Hurk T, van Rijn M, Zweers H, Ziadlou M, MacDonald A. **International practices in the dietary management of fructose 1-6 biphosphatase deficiency.** *Orphanet J Rare Dis.* 2018 Jan 25;13(1):21. doi: 10.1186/s13023-018-0760-3. PubMed PMID: 29370874; PubMed Central PMCID: PMC5785792.

9: Ponzi E, Maiorana A, Lepri FR, Mucciolo M, Semeraro M, Taurisano R, Olivieri G, Novelli A, Dionisi-Vici C. **Persistent Hypoglycemia in Children: Targeted Gene Panel Improves the Diagnosis of Hypoglycemia Due to Inborn Errors of Metabolism.** *J Pediatr.* 2018 Nov;202:272-278.e4. doi: 10.1016/j.jpeds.2018.06.050. Epub 2018 Sep 5. PubMed PMID: 30193751.

10: Porta F, Chiesa N, Martinelli D, Spada M. **Clinical, biochemical, and molecular spectrum of short/branched-chain acyl-CoA dehydrogenase deficiency: two new cases and review of literature.** *J Pediatr Endocrinol Metab.* 2019 Feb 25;32(2):101-108. doi: 10.1515/jpem-2018-0311. Review. PubMed PMID: 30730842.

11: Martinelli D, Goffredo BM, Stefania Falvella F, Marano M. **Acute hyperammonemia in children under deferasirox treatment: cutting the Gordian knot.** *Clin Toxicol (Phila).* 2018 Nov 16:1-2. doi: 10.1080/15563650.2018.1523425. [Epub ahead of print] PubMed PMID: 30442064.

12: Repp BM, Mastantuono E, Alston CL, Schiff M, Haack TB, Rötig A, Ardisson A, Lombès A, Catarino CB, Diodato D, Schottmann G, Poulton J, Burlina A, Jonckheere A, Munnich A, Rolinski B, Ghezzi D, Rokicki D, Wellesley D, Martinelli D, Wenhong D, Lamantea E, Ostergaard E, Pronicka E, Pierre G, Smeets HJM, Wittig I, Scurr I, de Coo IFM, Moroni I, Smet J, Mayr JA, Dai L, de Meirleir L, Schuelke M, Zeviani M, Morscher RJ, McFarland R, Seneca S, Klopstock T, Meitinger T, Wieland T, Strom TM, Herberg U, Ahting U, Sperl W, Nassogne MC, Ling H, Fang F, Freisinger P, Van Coster R, Strecker V, Taylor RW, Häberle J, Vockley J, Prokisch H, Wortmann S. **Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?** *Orphanet J Rare Dis.* 2018 Jul 19;13(1):120. doi: 10.1186/s13023-018-0784-8. PubMed PMID: 30025539; PubMed Central PMCID: PMC6053715.