Wissenschaftliche Arbeiten

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Originalarbeiten als Erstautorin oder korrespondierende Autorin (*equal contribution)

1. Keller R, Chrastina P, Pavlíková M, Gouveia S, Ribes A, Kölker S, Blom HJ, Baumgartner MR, Bartl J, Dionisi Vici C, Gleich F, Morris AA, *Kožich V, *Huemer M on behalf of the European network and registry for homocystinurias and methylation defects (E-HOD). Newborn screening for homocystinurias and methylation defects: current practice and future prospects. Under Review J Inherit Metab Dis 2017

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Fallbeschreibungen

1. Fleger M, Willomitzer J, Meinsma R, Alders M, Meijer J, Hennekam RCM, Huemer M*, van Kuilenburg ABP*. Dihydropyrimidine dehydrogenase deficiency - metabolic disease or biochemical phenotype? Accepted for publication JIMD Rep

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Auswahl von Abstracts und Postern

1. Huemer M, Baumgartner M, Kasper D, Merinero B, Pasquini E, Ribes A, Vilarinho L, Blom H, Kozich V. Recommended guidelines for newborn screening in the homocystinurias. J Inherit Metab Dis
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