Curriculum vitae

PD Dr. med. Nikolas Boy

Metabolic Paediatrician
Centre for Child and Adolescent Medicine
Division of Child Neurology and Metabolic Medicine
Heidelberg University Hospital
Im Neuenheimer Feld 669, 69120 Heidelberg
Tel. +49 6221 56-36973 | Fax. +49 6221 56-6391 |
E-Mail: nikolas.boy@med.uni-heidelberg.de
http://www.klinikum.uni-heidelberg.de

Clinical and scientific background

2001-2008	**University Medical School** in Magdeburg and Heidelberg, Germany
2007-2008	**Final year elective** at University Hopsital Heidelberg, Germany and Mount Sinai Hospital, Kravis Children's Hospital, New York, USA
2008	**Medical exam and final approbation**
2008	**Dissertation** *Dr. med* (Assessment: *magna cum laude*, *„Influence of newborn screening and early start of treatment on outcome and dynamic of neuroradiologic abnormalities in patients with glutaric aciduria type I)*
2009-2014	**Specialization in Paediatrics**
Since 2015	**Guideline coordinator** for the national AWMF-S3-guideline (AMWF-Regist.nr. 027-018) "Diagnosis, treament and management of glutaric aciduria type 1"
2012-2016	**Specialization in Metabolic Medicine** European Certificate (UEMS) of Paediatric Metabolic Medicine
Since 2018	**Specialization in Child Neurology**
2019	**Venia legendi and habilitation in Pediatrics** *Cumulative postdctoral thesis: Clinical long-term outcome and success of therapy in glutaric aciduria type 1*
2019	**Metabolic consultant**

Since 2010

Clinical research activities

(1) Clinical and cognitive longterm outcome in glutaric aciduria type 1
(2) Clinical phenotypes and disease variants in glutaric aciduria type 1
(3) Dynamics of neuroradiologcal abnormalities in glutaric aciduria type 1
(4) International guideline development coordinator for glutaric aciduria type 1
(5) Evaluation of longterm outcome of patients with organic acidurias and urea cycle defects („European registry and network for Intoxication type Metabolic Diseases“, E-IMD)
(6) Evaluation of longterm outcome of patients with IMD diagnosed by newborn screening in Germany *„Langzeitoutcome von Patienten aus dem Neugeborenenscreening (LZO)“)*
(7) Study site coordinator for the s*tandardized patient registry and natural history study within the German network for mitochondrial disorders (mitoNET)*

Heidelberg, July7th, 2020

PD Dr. med. Nikolas Boy

Reference list

First author

Boy N, Garbade SF, Heringer J, Seitz A, Kölker S, Harting I (2019) "Patterns, evolution, and severity of striatal injury in insidious- versus acute-onset glutaric aciduria type 1".
Journal of Inherited Metabolic Disease; 42: 117-127. **Peer-reviewed, IF 4,092**

Boy N, Heringer J, Brackmann R, Bodamer O, Seitz A, Kölker S, Harting I (2017) "Extrastriatal changes in patients with late-onset glutaric aciduria type I highlight the risk of long-term neurotoxicity".
Orphanet Journal of Rare Diseases; 12: 77. **Peer-reviewed, IF 3.607**

Boy N, Heringer J, Haege G, Glahn EM, Hoffmann GF, Garbade SF, Kölker S, Burgard P (2015) "A cross-sectional controlled developmental study of neuropsychological functions in patients with glutaric aciduria type I".
Orphanet Journal of Rare Diseases; 10: 163. **Peer-reviewed, IF 3.607**

Heringer J*, **Boy NPS***, Ensenauer R, Assmann B, Zschocke J, Harting I, Lücke T, Maier EM, Mühlhausen C, Haege G, Hoffmann GF, Burgard P, Kölker S (2010) "Use of guidelines improves the neurological outcome in glutaric aciduria type I".
Annals of Neurology; 68: 743-752. **Peer-reviewed, IF 10,244**
*both authors contributed equally

Boy N, Mengler K, Thimm E, Schiergens KA, Marquardt T, Weinhold N, Marquardt I, Das AM, Freisinger P, Grünert SC, Vossbeck J, Steinfeld R, Baumgartner MR, Beblo S, Dieckmann A, Näke A, Lindner M, Heringer J, Hoffmann GF, Mühlhausen C, Maier EM, Ensenauer R, Garbade SF, and Kölker S (2018) "Newborn screening, a disease-changing intervention for glutaric aciduria type 1".
Annals of Neurology; 83: 970-979. **Peer-reviewed, IF 10,244**

Boy N, Haege G, Heringer J, Assmann B, Mühlhausen C, Ensenauer R, Maier EM, Lücke T, Hoffmann GF, Müller, E, Burgard P, Kölker S (2013) "Low lysine diet in glutaric aciduria type I – effect on anthropometric and biochemical follow-up parameters".
Journal of Inherited Metabolic Diseases; 36: 525-533. **Peer-reviewed, IF 4,092**

Boy N, Mühlhausen C, Maier EM, Heringer J, Assmann B, Burgard P, Dixon M, Fleissner S, Greenberg CR, Harting I, Hoffmann GF, Karall D, Koeller DM, Krawinkel M, Okun JG, Opladen T, Posset R, Sahm K, Zschocke J, Kölker S, additional individual contributors (2017) "Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision".
Journal of Inherited Metabolic Disease; 40: 75-101. **Peer-reviewed, IF 4,092**

Co-author

Kölker S, Garbade SF, **Boy N**, Maier EM, Meissner T, Mühlhausen C, Hennermann JB, Lücke T, Häberle J, Baumkötter J, Haller W, Muller E, Zschocke J, Burgard P, Hoffmann GF (2007) "Decline of acute encephalopathic crises in children with glutaryl-CoA dehydrogenase deficiency identified by newborn screening in Germany".
Pediatric Research; 62:357-363. **Peer-reviewed, IF 2.761**

Harting I, Neumaier-Probst E, Seitz A, Maier EM, Assmann B, Baric I, Troncoso M, Mühlhausen C, Zschocke J, Mühlhausen C, Zschocke J, **Boy NPS**, Hoffmann GF, Garbade SF, Kölker S (2009) "Dynamic changes of striatal and extrastriatal MRI abnormalities in glutaric aciduria type I". *Brain*; 132:1764-82. **Peer-reviewed, IF 10.103**

Kölker S, **Boy NPS**, Heringer J, Müller E; Maier EM, Ensenauer R, Mühlhausen C, Schlune A, Greenberg CR, Koeller DM, Hoffmann GF, Haege G, Burgard P (2012) "Complementary dietary treatment using lysine-free, arginine-fortified amino acids supplements in glutaric aciduria type I - a decade of experience". *Molecular Genetics and Metabolism*; 107:72-80. **Peer-reviewed, IF 3.093**

Harting I, **Boy N**, Heringer J, Seitz A, Bendszus M, Pouwels PJW, Kölker S (2015) "(1)H-MRS in glutaric aciduria type 1. Impact of biochemical phenotype and age on the cerebral accumulation of neurotoxic metabolites". *Journal of Inherited Metabolic Disease*; 38:829-38. **Peer-reviewed, IF 3.541**

Kölker S, Valayannopoulos V, Burlina AB, Sykut-Cegielska J, Wijburg FA, Teles EL, Zeman J, Dionisi-Vici C, Barić I, Karall D, Arnoux JB, Avram P, Baumgartner MR, Blasco-Alonso J, **Boy N**, Rasmussen MB, Burgard P, Chabrol B, Chakrapani A, Chapman K, Cortès I Saladelafont E, Couce ML, de Meirleir L, Dobbelaere D, Furlan F, Gleich F, González MJ, Gradowska W, Grünewald S, Honzik T, Hörster F, Ioannou H, Jalan A, Häberle J, Haege G, Langereis E, de Lonlay P, Martinelli D, Matsumoto S, Mühlhausen C, Murphy E, de Baulny HO, Ortez C, Pedrón CC, Pintos-Morell G, Pena-Quintana L, Ramadža DP, Rodrigues E, Scholl-Bürgi S, Sokal E, Summar ML, Thompson N, Vara R, Pinera IV, Walter JH, Williams M, Lund AM, Garcia Cazorla A (2015) "The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype". *Journal of Inherited Metabolic Disease*; 38:1059-1074. **Peer-reviewed, IF 3.541**

Garbade SF, **Boy N**, Heringer J, Kölker S, Harting I (2018) "Age-related changes and reference values of bicaudate ratio and sagittal brainstem diameters on MR"I. *Neuropaediatrics* 49:269-275. **Peer-reviewed, IF 1.549**

Posset R, Garbade SF, **Boy N**, Burlina AB, Dionisi-Vici C, Dobbelaere D, Garcia-Cazorla A, de Lonlay P, Teles EL, Vara R, Ah Mew N, Batshaw M, Baumgartner MR, McCandless S, Seminara J, Summar M, Hoffmann GF, Kölker S, Burgard P (2019) "Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders - a successful strategy for clinical research of rare diseases". *Journal of Inherited Metabolic Disease*; 42:93-106. **Peer-reviewed, IF 3.541**

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 (2019) "Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry". *Journal of Inherited Metabolic Disease*; 42:333-352. **Peer-reviewed, IF 3.541**

Further publications

Online-Publications

Boy N, Mühlhausen, C, Maier EM, Heringer J, Assmann B, Burgard P, Fleissner S, Harting I, Hoffmann GF, Karall D, Krawinkel M, Okun JG, Opladen T, Posset R, Sahm K, Zschocke J, Ballhausen D, Fingerhut R, Scholl-Bürgi S, Koletzko B, Lindner M, vom Dahl S, Kölker S (2017) „Diagnostik, Therapie und Management der Glutarazidurie Typ I (Synonym: Glutaryl-CoA-Dehydrogenase-Defizienz)“
AWMF-S3-Leitlinie, Reg. Nr. 027/018; 2. Revision Juni 2016; URL: http://www.awmf.org.

Case reports

Zielonka M, Braun K, Seitz A, Kölker S, **Boy N** (2015) “Severe acute subdural haemorrhage in a patient with glutaric aciduria type I after minor head trauma”.
Journal of Child Neurology; 30:1065-9. **Peer-reviewed, IF 1.434**

Reviews

Boy NPS, Opp S, Heringer J, Okun JG, Sauer SW, Kölker S (2011) “Glutaric aciduria type I. A translational approach to an enigmatic disease”.
Journal of Pediatric Sciences; 3:e67. **Peer-reviewed, IF 1.008**

Heringer J, **Boy N**, Burgard P, Okun JG, Kölker S (2015) “Newborn Screening for Glutaric Aciduria Type I: Benefits and limitations”.
International Journal of Neonatal Screening; 1:57-68. **Peer-reviewed, IF pending**

Tuncel AT, **Boy N**, Morath MA, Hörster F, Mütze U, Kölker S (2018). Organic acidurias in adults: late complications and management.
Journal of Inherited Metabolic Disease; 41:765-776 **Peer-reviewed, IF 3.541**

Book Chapters

Boy N and Kölker S (2017) “Arachnoid cysts in glutaric aciduria type I”. In: Wester – Arachnoid cysts. Editors: Guy Eslick, Knut Wester. Elsevier GmbH

Book Reviews

Boy N, Peters V (2015) 2Congenital Neurotransmitter Disorders: A Clinical Approach2. Editors: Georg F. Hoffmann and Nenad Blau; ISBN: 978-1-63463-071-9.
Journal of Inherited Metabolic Disease; 38:587 **Peer-reviewed, IF 3.54**