Ass. Prof. Dr.Dorothea Möslinger
Medical University Vienna, AKH, Department of Pediatrics and Adolescent Medicine
Währinger Gürtel 18 – 20
1090 Vienna, Austria
Tel +43 1 40400 19278
dorothea.moeslinger@meduniwien.ac.at

CURRICULUM VITAE

UNIVERSITY TRAINING :

Studies of human medicine at the University of Innsbruck and University of Vienna Austria
1986 - Graduation : Medical Doctor at the University of Vienna

POSTGRADUATE EDUCATION AND MEDICAL PROFESSIONAL TRAINING :

1986 Start basic Medical Training at University Hospital of Vienna , Dept. of Surgery and Pediatrics

1992 : Resident : Pediatric Department Tygerberg Hospital, Capetown, University of Stellenbosh, South Africa

1995 Board Certification as a general Pediatrician

Since 1998 : Austrian Program for Neonatal Screening of Inborn Errors of Metabolism and special training in Metabolic diseases under Guidance of Prof S. Stöckler - Ipsiroglu

2001 : Ass Prof , Medical University Vienna, Department of Paediatrics
Consultant for Inborn Errors of Metabolism

Current Function :

Lead of the Clinical Center for metabolic disorders at the Department of Paediatrics and Adolescent Medicine at the Medical University Hospital Vienna

National Austrian Newborn Screening Program for inborn metabolic and endocrine disorders

Austrian B Expertise center rare metabolic disorders (Lead)

Local representative of MetabERN (European Reference network) as affiliated member

Memberships :

Austrian Society for Paediatrics (ÖGKJ)

Working group on congenital metabolic disorders of the ÖGKJ
Leader : 2011 – 2017

Austrian Forum for Rare Diseases

APS : Working society for paediatric metabolic diseases of the German Society for Paediatrics and Adolescent medicine

SSIEM : Society for the studies of Inborn Errors of Metabolism

Activities as a reviewer and in advisory boards in the metabolic field

Selected Publications :

Budd – Chiari syndrome associated with coagulation abnormalities in a child with carbohydrate deficient glycoproteine syndrome type Ix
Huemer M, Huber WD, Schima W, Moeslinger D, Holzbach U, Wevers R, Wank H, Stöckler – Ipsiroglu S
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Tyrosinämie Typ I: Klinische und biochemische Symptome bei drei Säuglingen
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Mutation analysis in Glycogen storage disease Type I non A
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Oral betahydroxybutyrate supplementation in two patients with hyperinsulinemic hypoglycemia : monitoring of beta – hydroxybutyrate levels in blood, cerebrospinal fluid, and in the brain by in vivo magnetic resonance spectroscopy.
Plecko B, Stoeckler – Ipsiroglu S, Schober E, Harrer G, Mylnarik V, Gruber S, Moser E, Moeslinger **D**, Silgoner H, Ipsiroglu O
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Molecular characterisation and neuropsychological outcome of 21 patients with profound biotinidase deficiency detected by newborn screening and family studies.
Möslinger D, Mühl A, Suormala T, Baumgartner R , Stöckler –Ipsiroglu S
Eur J Pediatr 2003, 162 , Suppl 1, S 46 – 49

Estimation of the false negative rate in newborn screening for congenital adrenal hyperplasia.
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European Journal of Endocrinology (2005) , 152 ; 869 - 874

Transcultural pediatrics: Compliance and outcome of PKU patients from families with an immigration background
Osman Ipsiroglu, Marion Herle, Elisabeth Spoula, Dorothea Möslinger, Banu Wimmer, Peter Burghard, Harald Bode, Sylvia Stöckler – Ipsiroglu
Wien Klin Wochenschr (2005), 117/15 – 16: 541 – 547

Growth and body composition in children with classical Phenylketonuria : Results in 34 patients and rewiew of literature
M Huemer, C Huemer, D. Möslinger, D Huter, S Stöckler – Ipsiroglu
J Inherit Metab Dis (2007) 30 : 694 - 699

Total Homocystein , B vitamins and genetic polymorphisms in patients with classical Phenylketonuria
Huemer M, Födinger M, Bodamer OA, Muehl A, Herle M, Weigmann C, Ulmer H, Stoeckler – Ipsiroglu S , Moeslinger D.
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Long term outcome of patients with Argininosuccinatlyase deficiency diagnosed by newborn screening in Austria
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Mol Genet Metab 2010 100 (1) : 24 – 8, EPub 2010 Feb 4

The National Austrian Newborn Screening Program – Eight years experience with mass spectrometry. Past Present, and future goals
David C. Kasper, Rene Ratschmann, Thomas F. Metz, Thomas P. Mechtler, Dorothea Möslinger, Vassiliki Konstantopoulou, Chike B. Item, Arnold Pollak, Kurt R. Herkner
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Bacterial expression of mutant argininosuccinate lyase reveals imperfect correlation of in vitro enzyme activity with clinical phenotype in argininosuccinic aciduria
Katharina Engel, Jean- Marc Vuissoz, Sandra Eggimann, Murielle Groux, Christoph Berning, Vera Klaus, Dorothea Möslinger**,** Saadet Mercimek – Mahmutoglu, Sylvia Stöckler, Bendicht Wermuth, Johannes Haeberle, Jean- Marc Nouffer
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Free asymmetric dimethylarginine (ADMA) is low in children and adolescents with classical phenylketonuria (PKU)
M. Huemer, B. Simma, D. Mayr, D Möslinger A Mühl, I Schmid, H Ulmer & O.A. Bodamer
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3 Methycrotonyl CoA deficiency: Clinical, biochemical, enzymatic and molecular studies in 88 individuals
Sarah C Grünert, Martin Stucki, Raphael J Morscher, Terttu Suormala,Celine Bürger,Patricie Burda,Ernst Christensen,Can Ficioglu, Jürgen Herwig,Stefan Kölker,Dorothea Möslinger,Elisabettta Pasquini,Rene Santer,K Ottfried Schwab,Briget Wilcken,Brian Fowler,Wyatt W Yue and Mathias Baumgartner
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Cultural aspects in the management of inborn errors of metabolism
Sylvia Stockler – Ipsiroglu, Dorothea Moeslinger, Marion Herle, Banu Wimmer, Osman S. Ipsiroglu
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Prevalence of tetrahydrobiopterin (BH4) – responsive allels among Austrian patients with PAH deficiency: comprehensive results from molecular analysis in 147 patients
E.Sterl, K. Paul, E. Paschke, J. Zschocke, M Brunner – Krainz, E Windisch,V.Konstantopoulou, **D**. Möslinger, D. Karall, S. Scholl – Bürgi, W. Sperl, F. Lagler, B Plecko
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Cross-sectional observational study of 208 patients with non-classical urea cycle Disorders
Corinne M. Rüegger1,2, Martin Lindner3, Diana Ballhausen4, Matthias R. Baumgartner1,2,Skadi Beblo5, Anibh Das6, Matthias Gautschi7, Esther M. Glahn3, Sarah C. Grünert8, Julia Hennermann9, Michel Hochuli10, Martina Huemer11, Daniela Karall12, Stefan Kölker3, RobinH. Lachmann13, Amelie Lotz-Havla14, Dorothea Möslinger15, Jean-Marc Nuoffer7,16, BarbaraPlecko2,17,18, Frank Rutsch19, René Santer20, Ute Spiekerkoetter8,21, Christian Staufner3, Tamar Stricker1,2, Frits A. Wijburg22, Monique Williams23, Peter Burgard3, Johannes Häberle1
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Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency

Rikke K.J. Olsen,1,28,* Eli_ska Kovnarı´kova´,2,3,28 Teresa A. Giancaspero,4,28 Signe Mosegaard,1,28Veronika Boczonadi,5,28 Lavinija Matakovi_c,6,28 Alice Veauville-Merllie´,7 Caterina Terrile,3Thomas Schwarzmayr,2,3 Tobias B. Haack,2,3 Mari Auranen,8 Piero Leone,4 Michele Galluccio,9Apolline Imbard,10,11 Purificacion Gutierrez-Rios,5,12 Johan Palmfeldt,1 Elisabeth Graf,3 Christine Vianey-Saban,7 Marcus Oppenheim,13 Manuel Schiff,14,15,16 Samia Pichard,15 Odile Rigal,10Angela Pyle,5 Patrick F. Chinnery,5,17 Vassiliki Konstantopoulou,18 Dorothea Möslinger,18,Rene´ G. Feichtinger,6 Beril Talim,19 Haluk Topaloglu,20 Turgay Coskun,21 Safak Gucer,19Annalisa Botta,22 Elena Pegoraro,23 Adriana Malena,23 Lodovica Vergani,23 Daniela Mazza,,24Marcella Zollino,24 Daniele Ghezzi,25 Cecile Acquaviva,7 Tiina Tyni,26 Avihu Boneh,27Thomas Meitinger,2,3 Tim M. Strom,2,3 Niels Gregersen,1 Johannes A. Mayr,6,29 Rita Horvath,5,29Maria Barile,4,29,* and Holger Prokisch2,3,29

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Demethylation of the promoter region of GPX3 in a newborn with classical phenylketonuria

Chike Bellarmine Item□, Sharmane Escueta, Andrea Schanzer, Somayeh Farhadi, Thomas Metz,Maximilian Zeyda, Dorothea Möslinger, Susanne Greber-Platzer, Vassiliki Konstantopoulou

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Manifestations of neurological symptoms and thromboembolism in adults with MTHFR-deficiency Paulus S. Rommer,□ , Johannes Zschocke , Brian Fowler , Manuela Födinger , Vassiliki Konstantopoulou **,** Dorothea Möslinger, Elisabeth Stögmann , Erhard Suess , Matthias Baumgartner , Eduard Auff , Gere Sunder-Plassmann

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Pompe disease in Austria: clinical, genetic and epidemiological aspects

W. N. Löscher,[1] M. Huemer,[2] T. M. Stulnig,[3] P. Simschitz,[4] S. Iglseder,[5] C. Eggers,[5] H. Moser,[6] D. Möslinger,[7] M. Freilinger,[7] F. Lagler,[8] S. Grinzinger,[9] M. Reichhardt,[10] R. E. Bittner,[11] W. M. Schmidt,[11] U. Lex,[12] M. Brunner-Krainz,[13] S. Quasthoff,[14] and J. V. Wanschitz[11]

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The natural history of classic galactosemia: lessons from the GalNet registry.

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Orphanet J Rare Dis. 2019 Apr 27;14(1):86. doi: 10.1186/s13023-019-1047-z.

Galactokinase deficiency: lessons from the GalNet registry

M. Estela Rubio-Gozalbo, MD, PhD 1 , Britt Derks, BSc1 , Anibh Martin Das, MD, PhD2 , Uta Meyer, RD2 , Dorothea Möslinger, MD3 , M. Luz Couce, MD, PhD4 , Aurélie Empain, MD5 , Can Ficicioglu, MD, PhD6 , Natalia Juliá Palacios, MD7 , Mariela M. De Los Santos De Pelegrin7 , Isabel A. Rivera, PhD8 , Sabine Scholl-Bürgi, MD, PhD9 , Annet M. Bosch, MD, PhD10, David Cassiman, MD, PhD11, Didem Demirbas, PhD12, Matthias Gautschi, MD, PhD13, Ina Knerr, MD, PhD14, Philippe Labrune, MD, PhD15,16, Anastasia Skouma, MD17, Patrick Verloo, MD18, Saskia B. Wortmann, MD, PhD19,20, Eileen P. Treacy, MD21, David J. Timson, PhD22 and Gerard T. Berry

Genetics in Medicine (2021) 23:202–210; https://doi.org/10.1038/s41436-020-00942-9

Hepatorenal Tyrosinaemia: Impact of a Simplified Diet on Metabolic Control and Clinical Outcome

Friederike Bärhold 1, Uta Meyer 1, Anne-Kathrin Neugebauer 2, Eva Maria Thimm 2, Dinah Lier 3,Stefanie Rosenbaum-Fabian 4, Ulrike Och 5, Anna Fekete 6, Dorothea Möslinger 6, Carmen Rohde

7, Skadi Beblo 7,Michel Hochuli 8,9, Nina Bogovic 10, Vanessa Korpel 10, Stephan vom Dahl 10, Sebene Mayorandan 1,†,Aleksandra Fischer 3, Peter Freisinger 3, Katharina Dokoupil 11, Margret Heddrich-Ellerbrok 12,
Monika Jörg-Streller 13, Agnes van Teeffelen-Heithoff 5, Janina Lahl 14 and Anibh Martin Das 1,*
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A retrospective study on disease management in children and adolescents with phenylketonuria during the Covid-19 pandemic lockdown in Austria
Marion Herle1 , Michaela Brunner-Krainz2 , Daniela Karall3 , Bernadette Goeschl1 , Dorothea Möslinger1 , Joachim Zobel2 , Barbara Plecko2 , Sabine Scholl-Bürgi3 , Johannes Spenger4 , Saskia B. Wortmann4,5 and Martina Huemer6,7*
Orphanet J Rare Dis (2021) 16:367 https://doi.org/10.1186/s13023-021-01996-x

Project "Backtoclinic I": An overview on the state of care of adult PKU patients in Austria
Marianna Beghini [1], Felix J Resch [2], Dorothea Möslinger [3], Vassiliki Konstantopoulou [4], Daniela Karall [5], Sabine Scholl-Bürgi [6], Michaela Brunner-Krainz [7], Barbara Plecko [8], Johannes Spenger [9], Alexandra Kautzky-Willer [10], Thomas Scherer [11], Miriam Hufgard-Leitner [12]
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Risk factors for impaired health‐ related quality of life in a cohort of pediatric patients with inborn metabolic diseases
Sandy Siegert1 ,Anne Roscher1 ,Dorothea Moeslinger1 ,Vassiliki Konstantopoulou1 · Marion Herle1
European Journal of Pediatrics European Journal of Pediatrics (2022) 181:1063–1070https://doi.org/10.1007/s00431-021-04300-y

100 years of inherited metabolic disorders in Austria—A national registry of minimal birth prevalence, diagnosis,and clinical outcome of inborn errors of metabolism in Austria between 1921 and 2021
Gabriele Ramoser1| Federica Caferri2| Bernhard Radlinger3|Michaela Brunner-Krainz4| Sybille Herbst1| Martina Huemer5|Miriam Hufgard-Leitner6| Susanne G. Kircher7| Vassiliki Konstantopoulou8|Wolfgang Löscher9| Dorothea Möslinger8| Barbara Plecko4|Johannes Spenger10| Thomas Stulnig6| Gere Sunder-Plassmann11|Saskia Wortmann10| Sabine Scholl-Bürgi1| Daniela Karall1| Austrian IMDRegistry
J Inherit Metab Dis.2022;45:144–156.
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Galactose epimerase deficiency: lessons from the GalNet registry
Britt Derks1,2,3,4†, Didem Demirbas5, Rodrigo R. Arantes6, Samantha Banford7, Alberto B. Burlina3,8,Analia Cabrera9, Ana Chiesa10, M. Luz Couce3,11, Carlo Dionisi‐ Vici3,12, Matthias Gautschi13,Stephanie Grunewald14, Eva Morava15, Dorothea Moslinger16, Sabine Scholl‐ Burgi3,17, Anastasia Skouma18,Karolina M. Stepien19, David J. Timson20^, Gerard T. Berry5† and M. Estela Rubio‐ Gozalbo1,2,3,4*†
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