



Program

15th–16th of June 2018

25th Annual Meeting of the German Society of Newborn Screening

Newborn Screening – Review – Insights – Outlook

Van Swieten Hall, Medical University of Vienna

Van-Swieten-Gasse 1a, 1090 Vienna

Conference committee:

Dr. Vassiliki Konstantopoulou

PD Dr. Maximilian Zeyda



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Vorwort

Sehr geehrte Damen und Herren, liebe Kolleginnen und Kollegen,

Wir freuen uns, Sie zur 25. Jahrestagung der Deutschen Gesellschaft für Neugeborenencreening (DGNS) 2018 einladen zu dürfen. Als Besonderheit findet diese Tagung erstmalig außerhalb Deutschlands, nämlich in Wien statt. Somit hoffen wir, neben einem wissenschaftlich interessanten Programm auch ein kulturell interessantes Ambiente bieten zu können. Darüber hinaus wollen wir den Ort Wien nutzen, um Kontakt und Austausch mit unseren Nachbarländern herzustellen bzw. zu erweitern.

Unter dem Motto „Rückblick – Einblick – Ausblick“ wollen wir im Programm auf Errungenschaften eingehen, um uns danach vertieft der aktuellen Problematik von Recalls und falsch positiven Ergebnissen zu widmen und schließlich mit dem Schwerpunkt „SCID und andere noch nicht gescreente Konditionen“ in die Zukunft zu blicken.

Wir hoffen auf Ihre Teilnahme und eine rege Beteiligung am wissenschaftlichen Programm durch Einsendung von Abstracts zu diesen oder verwandten Themen.

Wir freuen uns, Sie begrüßen zu dürfen und auf einen fruchtbaren Meinungs- und Erfahrungsaustausch.

Mit freundlichen Grüßen



Vassiliki Konstantopoulou



Maximilian Zeyda



Vassiliki Konstantopoulou



Maximilian Zeyda

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Foreword

**Dear Guests,
dear Colleagues,**

We are pleased to invite you to the 25th Annual Meeting of the German Society of Newborn Screening (DGNS). We are pleased to announce that for the first time, for the first time this meeting will take place outside of Germany – in Vienna. Thus, we hope that beyond an interesting scientific program, we can offer a nice cultural ambience. Moreover, we would like to take advantage of the location Vienna to foster contact and exchange to our neighboring countries.

Following the theme, "Review – Insights – Outlook", the program will highlight achievements, focus on the present problem of recalls and false positive results, and look into the future with the emphasis on SCID and other not yet screened conditions.

We hope you will join us and take part in the scientific program by submitting abstracts on these or related topics.

We are looking forward to meeting you and a fruitful exchange of opinions and experience.

With best regards



Vassiliki Konstantopoulou



Maximilian Zeyda



Vassiliki Konstantopoulou



Maximilian Zeyda

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Program, Friday 15th of June 2018

- 08:50 **Welcome**
Vassiliki Konstantopoulou & Maximilian Zeyda
- 09:00 **Opening**
Markus Müller, Rector Medical University of Vienna, Austria

Session 1 Development of newborn screening and specifics in Austria

- Chairs:** **Susanne Greber-Platzer**, Head of Department of Pediatrics, Medical University of Vienna, Austria
Vassiliki Konstantopoulou, Austrian Newborn Screening, Department of Pediatrics, Medical University of Vienna, Austria
- 09:15 L4 **Beginnings, developments, future of Austrian newborn screening – a Canadian point of view**
Sylvia Stöckler-Ipsiroglu
Department of Pediatrics, University of British Columbia, Vancouver, Canada
- 10:00 L5 **Austrian nation-wide mandatory prenatal toxoplasmosis screening**
Michael Hayde
Department of Pediatrics, Medical University of Vienna, Austria
- 10:45 Coffee, industrial exhibition & poster viewing

Session 2 Recalls & psychological aspects

- Chairs:** **Sylvia Stöckler-Ipsiroglu**
Maximilian Zeyda, Austrian Newborn Screening, Department of Pediatrics, Medical University of Vienna, Austria
- 11:30 L1 **First results from a study for the evaluation of 26 additional target disorders for the German newborn screening panel**
Gwendolyn Gramer
Center for Pediatric and Adolescent Medicine, University Hospital Heidelberg, Germany
- 12:15 01 **Parents' opinions on the communication of positive newborn screening results for cystic fibrosis in Bavaria**
Inken Brockow
Bavarian State Office for Health and Food Safety, Munich, Germany
- 12:30 02 **Psychological follow-up of patients with inborn errors of metabolism diagnosed in newborn screening at the Department of Pediatrics and Adolescent Medicine, Medical University of Vienna**
Marion Herle
Department of Pediatrics, Medical University of Vienna, Austria
- 12:45 Lunch

Session 3 Free topics

- Chairs:** **Gwendolyn Gramer**, Center for Pediatric and Adolescent Medicine, University Hospital Heidelberg, Germany
Dorothea Möslinger, Department of Pediatrics, Medical University of Vienna, Austria
- 13:45 03 **Newborn screening and vitamin disorders**
Petr Chrastina
Department of Pediatrics and Adolescent Medicine, General University Hospital in Prague, Czech Republic



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Bei einer Vielzahl schwerwiegender Erkrankungen, wie beispielsweise der Phenylketonurie, liegt der Fokus von BioMarin auf der Forschung, inspiriert durch die Patienten und die möglichen Auswirkungen einer Therapie auf ihr Leben.

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Program, Friday 15th of June 2018

- 14:00 04 **Combined tandem mass spectrometry (TMS) screening method for biotinidase deficiency and sickle cell disease (SCD)**
Jeannette Klein
Newborn Screening Laboratory, Charité Medical University Berlin, Germany
- 14:15 05 **High-throughput identification of hemoglobinopathies & thalassemias by HRAM/MS**
Zoltan Lukacs
Hamburg University Medical Center, Germany
- 14:30 06 **Simultaneous detection of amino acids, acylcarnitines and succinylacetone by tandem mass spectrometry without butylation: Influence of succinylacetone extraction on an established non derivatised newborn screening procedure**
Sabine Rönicke
Newborn Screening, Institute for Clinical Chemistry and Pathobiochemistry, Otto-von-Guericke University Magdeburg, Germany
- 14:45 Coffee & industrial exhibition

15:00 Poster sessions I & II

Poster session I – Screening programs; diseases

- P1 **Screening for galactosemia: A study from Hungarian NBS system**
Ildikó Szatmári
Center of Newborn Screening and Inherited Metabolic Disorders, 1st Department of Pediatrics, Semmelweis University, Budapest, Hungary
- P2 **Severe anemia in a newly diagnosed infant with LCHAD deficiency**
Andraz Šmon
University Children's Hospital, UMC Ljubljana, Slovenia
- P3 **Newborn screening in Slovakia – overview**
Maria Knapkova
Newborn Screening Centre Slovak Republic, Children's University Hospital Banská Bystrica, Slovakia
- P4 **Newborn Screening and Childbirth outside Health Care Facility/Center in Slovakia**
Andrea Revtakova
Newborn Screening Centre Slovak Republic, Children's University Hospital Banská Bystrica, Slovakia
- P5 **Marked reduction of false-positives in Austrian newborn screening for cystic fibrosis after implementation of pancreatitis-associated protein (PAP) as second-tier test**
Andrea Schanzer
Austrian Newborn Screening, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria
- P6 **Decreased concentrations of isovaleryl-carnitine in patients with maple syrup urine disease (MSUD)**
Ralph Fingerhut
University Children's Hospital, Swiss Newborn Screening Laboratory and Children's Research Center, Zurich, Switzerland



SCREEN-ID Neonatal TREC-KREC kit

Newborn screening for SCID and XLA
based on quantification of
TREC and KREC

Program, Friday 15th of June 2018

Poster session II – Vitamin B12; parameter evaluation

- P7 **Vitamin B12 deficiency detected in newborn screening – an important secondary finding**
Vassiliki Konstantopoulou
Austrian Newborn Screening, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria
- P8 **Maternal vitamin B12 deficiency leads to elevated methylmalonate and total homocysteine with normal propionyl carnitine in dried blood spots from newborn screening**
Alexandra Tsouka
Center des maladies moléculaires – CHUV, Lausanne, Switzerland
- P9 **Screening for Vitamin B12 deficiency in Austrian newborns by measurement of homocysteine as second-tier test**
Tomaž Rozmarič
Austrian Newborn Screening, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria
- P10 **Evaluation of newborn screening for congenital adrenal hyperplasia in Austria since its implementation in 2001**
Melanie Friedrich
Department of Pediatrics and Adolescent Medicine, Clinical Division of Pulmology, Allergology and Endocrinology, Medical University of Vienna, Austria
- P11 **Stability of IRT in dried blood samples**
Ralph Fingerhut
University Children's Hospital, Swiss Newborn Screening Laboratory and Children's Research Center, Zurich, Switzerland
- P12 **Seasonal variation of IRT values and sex differences**
Thomas Metz
Austrian Newborn Screening, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria
-

- 15:30 **DGNS report and discussion**
(in German)
- 16:30 **DGNS general meeting**
(for members only, in German)
- 18:00 **Guided walk and educational dinner**

Abstracts published in the meeting report in the International Journal of Neonatal Screening 2018, 4(2), 17.

Available online at:
<http://www.mdpi.com/journal/neonatalscreening>



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Program, Saturday 16th of June 2018

09:00 **Opening remarks**
Vassiliki Konstantopoulou & Maximilian Zeyda

Session 4 Screening for primary immunodeficiencies

Chairs: **Kaan Boztug**, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases; CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences; St. Anna Children's Hospital and Children's Cancer Research Institute; Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria
Elisabeth Förster-Waldl, Jeffrey Modell Diagnostic & Research Center Vienna and Division of Pediatric Immunology, Department of Pediatrics, Medical University of Vienna, Austria

09:05 L2 **Experiences from Israeli nation-wide screening for primary immunodeficiencies**
Raz Somech

Jeffrey Modell Foundation Center, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Sackler Faculty of Medicine, Tel Aviv University; Israeli national lab for confirming PID in NBS, Tel-Hashomer, Israel

09:50 L3 **Screening for primary immunodeficiencies in Andalusia**

Beatriz de Felipe, Unidad de Pediatría, Hospital Virgen del Rocío, Sevilla; Instituto de Biomedicina de Sevilla, Spain

10:35 Coffee, industrial exhibition & poster viewing

Session 5 Conditions considered for NBS

Chairs: **Beatriz de Felipe**
Raz Somech

11:15 07 **First PID screening experience with a qPCR-based Trec/Krec-assay**
Ralph Fingerhut

University Children's Hospital, Swiss Newborn Screening Laboratory and Children's Research Center, Zurich, Switzerland

11:30 08 **RareScreen – a cross-border newborn screening project detecting SCID, hemoglobinopathies and familial hypercholesterolemia**

Theresa Winter, University of Medicine Greifswald, Germany

11:45 09 **Early Check: Generating evidence to inform newborn screening for rare disorders**

Don Bailey, Center for Newborn Screening, Ethics, and Disability Studies, Research Triangle Park, North Carolina, USA

12:00 **General discussion – new conditions**

Chair: **Ralph Fingerhut** & **Uta Nennstiel**, President DGNS

12:20 **Best poster award**

Uta Nennstiel, President DGNS

12:25 **Introduction of IJNS, best abstract award**

Ralph Fingerhut, Editor-in-Chief IJNS

12:35 **Introduction of next meeting**

12:45 End of meeting

13:00 Snack

Program modifications reserved.

Abstracts published in the meeting report in the International Journal of Neonatal Screening 2018, 4(2), 17.

Available online at:

<http://www.mdpi.com/journal/neonatalscreening>



CF NEWBORN SCREENING: THE IRT/PAP STRATEGY

RATIONALE

PAP is a pancreatic secretory protein

PAP is - ABSENT from normal pancreas

- OVEREXPRESSED in the diseased pancreas (e.g. in CF) and leaks to blood

In newborns, elevated IRT AND PAP strongly suggests CF

THE IRT/PAP STRATEGY

Assay IRT in all newborns, then assay PAP in newborns with elevated IRT

Newborns with high IRT and PAP are recalled for a sweat test

COMPARED TO IRT/DNA

- same specificity and sensitivity
- number of detected mild forms of CF is decreased
- no need for extending the panel of CFTR mutations in multi-ethnic populations
- cheaper
- may be followed by a DNA tier to increase specificity

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- 3 - van der Ploeg CP, van den Akker-van Marle ME, Vernooij-van Langen AM, Elvers LH, Gille JJ, Verkerk PH, Dankert-Roelse JE; CHOPIN study group.
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- 4 - Sarles J, Giorgi R, Berthézène P, Munck A, Cheillan D, Dagorn JC, Roussey M.
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