ISNS 2018 programme

Sunday October 14	
17.00 - 17.10	Welcoming addresses
17.10 – 17.25 **17.25 - 18.10**	Opening session (moderator: Rodney Howell) **Svetozár Dluholucký** – Some Notes on the History of newborn Screening in Slovakia **Jim Bonham** - Newborn screening is Pandora's box; new techniques make anything screenable
18.10 - 20.00	Welcome Reception
Monday October 15	
08.30 - 10.10	Session 1 (moderators: Gerard Loeber, Peter Schielen) *Newborn screening current status in Europe – country reports part 1* **Iceland** - Leifur Franszon **United Kingdom** – Lesley Tetlow **Portugal** – Hugo Rocha **Spain** – Raquel Yahyaoui **Netherlands** – Eugenie Dekkers **Luxembourg** – Patricia Borde **Norway** – Rolf Pettersen **Denmark** – David Hougaard **Germany** – Zoltan Lukacs **Switzerland** – Ralph Fingerhut **Italy** – Giancarlo La Marca **Malta** – Ian Brincat
10.10 - 10.40	Coffee/tea break and Poster viewing
10.40 - 12.15	Session 1 (continued, moderators: Gerard Loeber, Peter Schielen) *Newborn screening current status in Europe – country reports part 2* **Czech Republic** – Felix Votava **Slovenia** – Urh Groselj **Poland** – Mariusz Oltarzewski **Slovakia** – Maria Knapkova **Hungary** – Ildiko Szatmari **Lithuania** – Jurgita Songailiené **Romania** – Florentina Moldovanu **Greece** – Panagiotis Girginoudis **Russia** – Natalia Pechatnikova **Kazakhstan** – Gulnara Syvatova Summary – Gerard Loeber
12.15 - 13.30	Lunch break and Industrial workshop

12.30 - 13.30
Industrial Workshop (sponsored by Sanofi Genzyme)
(moderator Rodney Howell)
Successful addition of lysosomal storage disorders to a newborn screening program
Speakers: David Kasper, Alberto Burlina

13.30 – 14.10
Session 2 (moderators: Maria Knapkova, Piero Rinaldo)
Definitions for newborn screening conditions
13.30 – 13.50 Veronica Wiley – Does it count?
13.50 – 14.10 Stefan Koelker - Long-term outcome studies, the (often) neglected part of newborn screening programmes

14.10-15.30
Session 3 (moderators: Kate Hall, Marelle Bouva)
Pitfalls and solutions in newborn screening part 1
14.10 - 14.30 Toni Torresani - Neonatal screening for congenital hypothyroidism: 50 years of changing definitions
14.30 – 14.50 Patrice Held - Detection of congenital adrenal hyperplasia: Is there a need to screen all babies twice?
14.50 – 15.30 Piero Rinaldo - Precision newborn screening driven by results adjustments for multiple covariates

15.30 - 16.00 Coffee/tea break and Poster viewing

16.00 - 17.30
Session 3 (continued, moderators: Kate Hall, Toni Torresani))
16.00 – 16.12 Can Ficicioglu – Newborn screening for X-linked adrenoleukodystrophy, the CHOP experience
16.12 – 16.24 Rachel Carling - Increased use of Pivmecillinam highlights the need for a 2nd tier screening test to improve analytical reliability of isovaleric acidaemia screening
16.24 – 16.36 Kate John - Sickle screening by MSMS: the practicalities of introducing into the routine newborn screening service
16.36- 16.48 Marelle Bouva – Simultaneous transition to a new tandem mass spectrometry system in five (Dutch) laboratories: experimental evaluation to monitor analytical changes
16.48 - 17.00 Rendelien Verschoof – Improving dried blood spot quality in preparation of the expansion of the neonatal screening program in the Netherlands
17.00 – 17.12 Rachel Carling - Using a common internal standard to reduce interlaboratory variation and improve analytical reliability of expanded newborn screening in England
17.12 – 17.30 Joel Ehrenkranz - Smartphone point-of-care newborn screening for endocrine and hematologic disorders: indications, regulations, experience, and future directions

Tuesday October 16

08.30 – 09.50
Session 4 (moderators: Jim Bonham, Stephan Borte)
Newborn screening for severe combined immunodeficiency
08.30 – 08.50 Bobby Gaspar- Newborn screening for severe combined immunodeficiency (SCID)
08.50 – 09.10 Leire Solis - SCID NBS - IPOPI's perspective
09.10 – 09.30 Dianne Webster – Introduction of SCID screening to New Zealand
09.30 - 09.50 Peter Schielen - Newborn screening for SCID-The Dutch approach

09.50 - 10.20 Coffee/tea break & Poster viewing

10.20 – 12.10
Session 4 (continued, moderators: Jim Bonham, Stephan Borte)
10.20 – 10.40 Simon Hailstone – SCID Screening: A public health perspective
10.40 – 11.00 Rolf Zetterström - Neonatal screening for severe primary immunodeficiencies in Sweden
11.00 – 11.20 Stephan Borte - Needs and perspectives of future neonatal screenings for severe immunodeficiencies
11.20 - 11.30 Peter Čižnár - Severe combined immunodeficiency – single centre experience
11.30 – 11.40 Asbjørg Stray-Pedersen – Second-tier next generation sequencing in newborn SCID screening provides rapid molecular confirmation of disease with implications for follow-up and treatment
11.40 – 11.50 James Chilcott - Screening for severe combined immunodeficiency: Using probabilistic uncertainty analysis of economic models to assess the reliability of screening policy decisions

11.50 – 12.10 General discussion

12.10 - 13.30	Lunch break

12:30 – 13:30
A live demo of Collaborative Laboratory Integrated Reports (CLIR), the R4S replacement website

13.30 – 15.15
Session 5 (moderators: Dianne Webster, Rodney Howell)
Pitfalls and solutions in NBS part 2
13.30 - 13.50 Natasha Heather - How does TSH cutoff impact the reliability of congenital hypothyroid screening?
13.50 – 14.10 Anita Boelen - Critical evaluation of the Dutch Neonatal screening on Central Congenital Hypothyroidism
14.10 – 14.30 Joanne Mei – Cutoff determinations and risk assessment methods in dried blood spot newborn screening
14.30 – 14.50 Christine McRoberts - Optimizing preanalytical processes to ensure the reliability of newborn screening results

14.50 - 15.20	Coffee/tea break & Poster viewing

15.20 – 18.00
Session 6 (moderators: Joanne Mei, Veronica Wiley)
NBS for SMA
15.20 – 15.45 David Kasper - Screening for neuromuscular disorders. Long history, novel opportunities
15.45 - 16.10 Katarina Okalova - Clinical presentation of spinal muscular atrophy
16.10 – 16.35 Laurent Servais - Spinal muscular atrophy: a challenging disease for newborn screening ?
16.35 – 17.00 Martina Cornel -When is a treatment "accepted"? Balancing pros and cons for expensive and invasive treatment after neonatal screening

17.00 – 17.12 Jennifer Taylor - Screening for spinal muscular atrophy in North Carolina State
17.12 – 17.24 Henk Engel - Validation of a fast, robust, inexpensive, first-tier molecular neonatal screening test on dried blood spots for spinal muscular atrophy without carrier detection
17.24 – 17.30 Francois Boemer - (S)un (M)ay (A)rise on SMA: the newborn screening Liege's experience

17.30 – 18.00 General discussion

Wednesday October 17

08.30 – 09.30
Session 7 (moderators: Martina Cornel, Felix Votava)
Information to parents
08.30 – 08.50 Marleen Jansen - Information for parents and professionals, status in Europe

08.50 – 09.02 Vera Frankova – Factors influencing parental awareness about newborn screening
09.02 – 09.14 Sarah Viall - "So your baby had an abnormal neonatal screen result. Now what?": Keys to effective communication
09.14 – 09.26 Scott Shone - Early Check: a voluntary public health research program to inform newborn screening

09.30 – 10.15	**ISNS membership meeting**
10.15 – 10.45	Coffee/tea break & Poster viewing

10.45 - 12.15
Session 8 (moderators: Gerard Loeber, Viktor Kožich)
Inborn errors of metabolism
10.45 – 11.10 Viktor Kožich - Guidelines and practice of newborn screening for homocystinurias

11.10 – 11.22 David Millington - Lysosomal storage disorder newborn screening: comparing performance of tandem mass spectrometry and digital microfluidics platforms
11.22 – 11.34 Guisou Zarbalian - Quality improvement facilitation of newborn screening systems via site visits offered by the U.S. Association of Public Health Laboratories
11.34 – 11.45 Dianne Webster - Reliability of newborn screening in preterm, low birthweight and sick infants

11.50 – 12.15 General Discussion

12.15 - 12.30	**Announcement of poster prize winner** **Closure**