



13th
European
ISNS
2025

23 - 26 March 2025
Luxembourg



Sunday, 23rd March 2025

17:00 – 20:00 Welcome and Opening

17:00	Conference President	Patricia Borde, Luxembourg
17:05	Chief Medical Officer at the Directorate of Health	Jean-Claude Schmit, Luxembourg
17:10	Deputy Chief Executive Officer, Luxembourg National Health Laboratory	Thomas Dentzer, Luxembourg
17:15	ISNS President	Jim Bonham, England

Monday, 24th March 2025

08:30 – 09:00 Session 1 : Keynote Lecture

Moderators: Jim Bonham and Clement Kebbabi

	Reactomics and its future in newborn screening	Raquel Yahyaoui, Spain
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09:00 – 10:30 Session 2 : World view and Country reports

World view

09:00	The world of neonatal screening in numbers and Europe compared	Peter Schielen, Netherlands
09:15	The spectrum of approaches to neonatal screening around the world, Europe and unmet needs	Urh Groselij, Slovenia

Country reports

09:30	1.	
09:40	2. Czech Republic	Felix Votava
09:50	3. Switzerland	Susanna Sluka
10:00	4. Netherlands	Eugenie Dekkers
10:10	5. Finland	Riikka Kurkijärvi
10:20	6. Austria	Maximilian Zeyda

10:30 – 11:00 Coffee break & Poster viewing



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11:00 – 12:45 Session 3 : Molecular and biochemical markers in newborn screening

Moderators: Stuart Moat and Viktor Kozich

11:00	A global view of genomic sequencing in newborn screening: Where we are now and where we are going?	David Bick, England
11:15	Population based first tier genomic newborn screening – the BabyDetect project: what will it tell us?	Francois Boemer, Belgium
11:30	Decoding uncertainty: variants of unknown significance and the limitations of genomics without biochemistry	Asbjörg Stray-Pederson, Norway Jernej Kovac, Slovenia
12:00	Untargeted metabolomics: great potential for the discovery of new biomarkers for inclusion in newborn screening	Leo Kluijtmans, Netherlands
12:30	Practical lessons in integrating genetics into the daily workflow in a national neonatal screening unit setting.	Janne Strand, Norway

12:45 – 13:15 Lunch break

13:15 – 14:15 REVVITY Workshop – Advancements in MSMS (*see website-page [Workshops](#)*)

14:15 – 15:15 Session 4 : Neuromuscular disorders

Moderators: Rose Maase and Lesley Tetlow

14:15	Management of Duchenne muscular dystrophy: How newborn screening could be game-changing.	Laurent Servais, England
14:45	Bloodspot screening for Duchenne muscular dystrophy, the past and the future.	Stuart Moat, Wales
15:00	Two years of spinal muscular atrophy, SMA, screening in the Netherlands and the treatment options	Els Voorhoeve, Netherlands Ludo van der Pol, Netherlands

15:15 – 15:45 Coffee break & Poster viewing

15:45 – 16:45 Session 5a : Long term outcomes

Moderators: Dianne Webster and Rolf Zetterström

15:45	Registries are key to precision medicine and improvement of screening programmes	Stefan Kölker, Germany
16:15	National registry studies of long-term outcome for CAH and CH in Sweden – examples of using unique, existing registers.	Anna Nordenstrom, Sweden



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16:45 – 17:20 Session 5b : Missed babies, children previously unscreened, specimen storage

16:45	Dried bloodspot storage, biobanking - European inventory and legal issues	Mette Nyegaard, Denmark
17:00	Experiences of screening older, previously untested children in the Swedish DBS Newborn Screening Program	Lene Sörensen, Sweden

17:20 – 18:00 Session 6 : Medley

17:20	Mutation analysis in congenital hypothyroidism – clear links and cautionary tales	Rolf Zetterström, Sweden Alexander Rowe, Norway
17:45	Use of AI and automated bloodspot punch machines to evaluate blood spot quality	Nick Flynn, England

18:00 – 18:10 End of day summary

Tuesday, 25th March 2025

08:30 – 09:20 Session 7a: Orals Selected from Poster Abstracts

Moderators: Marizela Kulisic and Peter Schielen

08:30	Routine application of a high-throughput targeted metabolomics 2 nd tier test in Newborn Screening	Michela Perrone Donnorso, Italy
08:40	CLIR Status Update: Utilization, Regulation and Opportunities	Patricia Hall, USA
08:50	A novel neonatal screening modality: In-depth non-targeted proteome analysis of dried blood spots	Osamu Ohara, Japan
09:00	Pilot newborn screening for vitamin B12 deficiency in Czechia: benefit for newborns and their mothers	Tomas Honzik, Czech Republic
09:10	Evaluation of Neonatal Screening Programs for Tyrosinemia Type 1 Worldwide	Allysa Kuypers, Netherlands

09:20 – 09:35 Session 7b : Special Presentation

	Egypt's Comprehensive Achievements and Overview: 25 Years of Newborn Screening.	Hon. Prof Mohamad Hassany, Egypt
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09:35 – 10:15 Session 8: ISNS Members Meeting

09:35	Information, updates, benefits, Q&A	Jim Bonham, England
09:55	The ISNS - 10 th anniversary celebration	Neil Ding

10:15 – 10:45 Coffee break and poster viewing



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10:45 – 12:15 Session 9 - European countries and their different phases of development of newborn screening

Moderators: Patricia Borde and Dimitris Platis

10:45	IFCC/ISNS Global Task Force	Jim Bonham, England
10:55	Our Journey from Individual Efforts to Nationwide Support: Implementing Newborn Screening for SMA and other disorders in Serbia	Milos Brkusanin, Serbia
11:05	Newborn screening in Georgia, problems and future perspectives.	Nazi Tabatadze, Georgia
11:15	Newborn screening in Romania – a video	Dorica Dan, Romania
11:25	Neonatal Screening in Bulgaria - an ongoing challenge.	Iva Stoeva, Bulgaria
11:35	Two Years of Expanded Neonatal Screening in Ukraine: Success Despite the Challenges	Natalia Olkovych, Ukraine
11:45	Challenges of newborn screening expansion in Lithuania	Jurgita Songailiene, Lithuania
11:55	National and pilot screening programs in Estonia	Katrin Õunap, Estonia
12:05	Neonatal screening in the Republic of Kazakhstan: current state and prospects for further development	Damilya Salimbayeva, Kazakhstan

12:15 – 12:45 Lunch break

12:45 – 13:45 Sponsored workshop

13:45– 14:25 Session 10: Ethical, Legal and Social Implications (ELSI) of newborn screening

Moderators: Dianne Webster and Rolf Zetterström

13:45	ELSI implications of genomic NBS for each step of the programme	Věra Franková, Czech Republic
14:05	Psychosocial communication - doctor-patient - dietitian-patient/parent	Eleana Petropoulou, Greece

14:25 – 15:15 Session 11: The newborn screening laboratory and cyber attacks, resilience

Moderators: Dimitris Platis and Urh Groselj

	Dublin Children's Hospital	Loretta O'Grady, Ireland
	Viapath Laboratory, London	Rachel Carling, England
	Waters	Ian Hutton, England
	Revvity	Tommi Elo, Finland

15:15 – 15:45 Coffee break and poster viewing



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15:45 – 17:00 Session 12a: New diseases in Europe

Moderators: Jim Bonham and Raquel Yahyaoui

15:45	Newborn screening for remethylation disorders: the state of the art.	Giancarlo Lamarca, Italy
16:05	The Dutch laboratory experience of recent NBS programme expansions - sharing key insights and knowledge gained	Rose Maase, Netherlands
16:20	Metachromatic leukodystrophy, a treatable disease: the case for newborn screening - video	Lucia Laugwitz, Germany
16:35	Screening baby boys for adrenoleukodystrophy in the Netherlands. The impact of variants of unknown significance.	Stephan Kemp, Netherlands

17.00 -17.20 Session 12b: The patient experience

17:00	Adrenoleukodystrophy - a family story behind the need for screening.	Karen Harrison, England
17:10	Screening for MPS1: what is the real life result?	Hanka Dekker, Netherlands

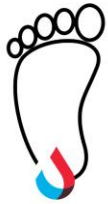
Wednesday, 26th March 2025

08:30 – 10:00 Session 13: Newborn screening in Europe

08:30	Screen4Rare, the involvement of ISNS	Jim Bonham, England
08:40	Workstream 1 a blueprint for screening in Europe – identifying good and best screening practices	Peter Schielen, Netherlands
08:57	Workstream 2 Case Definitions - Who is the patient?	Annet Bosch, Netherlands
09:14	Workstream 3 Data-driven guidance of a screening strategy for rare diseases	Ulrike Mütze, Germany
09:31	The EU advisory committee on newborn screening under the umbrella of the ERNs	Andrea Bordugo, Italy
09:48	Future perspective, next steps, what needs to be done next, forward looking to 2028.	Leire Solis, Portugal

10:00-10:30

Coffee break and poster viewing



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10:30 – 12:10 Session 14 : Cystic fibrosis

10:30	Life changing drugs for some people with CF; implications for screening	Kevin Southern, England
11:00	Neonatal Screening for CF in Europe: Different Challenges Require Different Algorithms - Is PAP the Solution for All?	Olaf Sommerburg, Germany
11:30	Impact of increasing ethnic diversity on the UK algorithm	Toby Greenfield, England
11:40	Challenges of providing genetic testing for CF in a small country	Marizela Kulisic, Luxembourg
11:50	Swiss CF register: improving performance through careful evaluation of outcomes	Jürg Barben, Switzerland

12:10 – 12:45

Award of poster prizes and closing ceremony