



# ISNS Luxembourg Going Virtual Programme

## 10-12 November 2021

**Time:** *Central European Time (CET), UTC/GMT +1*

### WEDNESDAY 10 NOVEMBER

12:15-12:45	<b>Welcoming Addresses</b>	
12:15	Introduction from the President of ISNS 2021 Luxembourg Going Virtual	Patricia Borde
12:20	Vice-President of the European Parliament Welcome	Fabio Massimo Castaldo
12:25	Deputy Head of the Health Directorate, Ministry of Health	Françoise Berthet
12:30	Director of the Laboratoire National de Santé	Friedrich Mühschlegel
12:35	ISNS President Welcome	Jim Bonham
12:40	An appreciation of Zoltan Lukacs and Louis Woolf	Peter Schielen

12:45-13:00	<b>Break</b>	
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13:00-14:00	<b>Session 1: Genetic Testing in Newborn Screening</b> <b>Chairs: Peter Schielen &amp; Dominique Bourgeois</b>	
13:00	Whole Genome Sequencing for Precision Diagnostics in Rare Diseases - Implications for Newborn Screening	Anna Wedell (Stockholm, Sweden)
13:15	The Impact of the Second-tier Tests and Role of Next Generation Sequencing on Newborn Screening Performance - Could They Become First Line Tests?	Belén Pérez González (Madrid, Spain)
13:30	Second-tier Genetic Testing in the Norwegian National Screening Programme	Janne Strand (Oslo, Norway)
13:45	Q&A	All

14:00-14:15	<b>Break</b>	
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14:15-15:15	<b>Session 2: Impact of Maternal Health on Newborn Screening</b> <b>Chairs: Dianne Webster &amp; Viktor Kožich</b>	
14:15	Maternal Vitamin B12 Deficiency: Definition in Relation to Newborn Screening, Are These True Positives?	Anne-Lise Björke Monsen (Bergen, Norway)
14:30	Maternal Vitamin B12 Deficiency - Impact on Screening for MMA/PA	Raquel Yahyaoui Mácias (Malaga, Spain)
14:45	Maternal Iodine - Too Little and Too Much	Natasha Heather (Auckland, New Zealand)
15:00	Q&A	All

15:15-15:30	Break
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15:30:16:30	Session 3: Ethical, Legal and Social Implications of Newborn Screening Chair: Jim Bonham & Peter Schielen	
15:30	If You Can Screen for 300 Disorders Why Stop at 30?	Martina Cornel (Amsterdam, Netherlands)
15:45	Just Because You Can Doesn't Mean You Should - Screen Newborns for Any Disorder	Wybo Dondorp (Amsterdam, Netherlands)
16:00	Ethical Dilemmas with Identification of Untreatable Conditions and Findings of Unclear Significance	Trine Tangeraas (Oslo, Norway)
16:15	Moderator for 15-minute Debate	Jim Bonham (Sheffield, UK)

16:30-16:45	Break
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16:45-17:45	Commercial Workshop: LabSystems Diagnostics Oy
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17:50-18:00	Summary of Day 1 Eugenie Dekkers & Rolf Zetterström
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## THURSDAY 11 NOVEMBER

12:45-13:05	Session 4: Country Reports Chair: Giancarlo la Marca	
12:45	European Overview	Dimitris Platis (Athens, Greece)
13:00	Q&A	All

13:05-13:10	Break
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13:10-14:20	Session 5: Orals from Delegates' Abstracts Chairs: Giancarlo la Marca & Kate Hall	
13:10	Application of a Protocol for Structured Follow-up and Texting of Inadequate or Borderline Positive Screen Results - <i>Natasha Heather</i>	
13:20	An Introduction to the Screen4Rare Stakeholder Platform for Equity in Neonatal Screening in Europe - <i>Gerard Loeber &amp; Johan Prevot</i>	
13:30	Enhancing Data-driven Disease Detection in Newborns: A National Data Platform for Modernizing Newborn Screening Data Analytics and Interpretation - <i>Carla Cuthbert &amp; Amy Gaviglio</i>	
13:40	Global Impact of COVID-19 on Newborn Screening Programmes - <i>Urh Groselj</i>	
13:50	Advances in Mass Spectrometry Based Newborn Screening: Quadrupling the Throughput of First-tier Screening and Same Day, Same Instrument, Second-tier Screening Using Intelligent Reflex - <i>Konstantinos Petritis</i>	
14:00	Q&A	All

14:20-14:30	Break	
14:30:15:15	Session 6: International Society for Neonatal Screening, ISNS Chair: Jim Bonham	
14:30	Information about the ISNS for Non-members and Members	Jim Bonham (Sheffield, UK)
14:35	Annual General Meeting - ISNS Members Only	Jim Bonham (Sheffield, UK)
15:05	Q&A	All
15:15-15:20	Break	
15:20-16:35	Session 7: Long-term Outcomes and Registries Chairs: Viktor Kožich & David Cheillan	
15:20	IT Infrastructure for Long-term Follow-up for Newborn Screening in Sweden	Rolf Zetterström (Stockholm, Sweden)
15:40	The Value of European Registries and Networks for Changing Practice and Informing Guidelines	Stefan Kölker (Heidelberg, Germany)
16:05	Film and Discussion - Value of Late Treatment of PKU Patients Born Before Newborn Screening	Maria Giżewska (Szczecin, Poland)
16:20	Q&A	All
16:35-16:45	Break	
16:45-17:45	Commercial Workshop: Perkin Elmer – <a href="#">PROGRAMME HERE</a>	
17:50-18:00	Summary of Day 2 Raquel Yahyaoui Mácias & Dianne Webster	
18:15-18:45	Social event: New Pirate Radio Luxembourg Interactive quiz	

## FRIDAY 12 NOVEMBER

12:50-14:00	Session 8: New Screening Disorders I Chairs: Raquel Yahyaoui Mácias & Dominique Bourgeois	
12:50	Newborn Screening for Spinal Muscular Atrophy, SMA: Achievements, Challenges, Controversies	Janbernd Kirschner (Bonn, Germany)
13:10	SMA - Survey of the European Screening Laboratory Experience	François Boemer (Liège, Belgium)
13:25	Oral from Abstract: Implementation of a Combined Screening Test for SCID and SMA in the Netherlands Based on Quantitative PCR - <i>Els Voorhoeve</i>	
13:35	Oral from Abstract: Statewide Newborn Screening for Spinal Muscular Atrophy: The Wisconsin Experience - <i>Mei Baker</i>	
13:45	Q&A	All

14:00-14:15	Break	
14:15-15:15	<b>Session 9: New Screening Disorders II</b> <b>Chairs: France Debaugnies &amp; Rose Maase</b>	
14:15	A European Overview of Newborn Screening for SCID: Experiences and Future Perspectives	Ana Argudo Ramírez (Barcelona, Spain)
14:30	The Dutch Newborn Screening Pilot Programme for X-linked Adrenoleukodystrophy (ALD)	Stephan Kemp (Amsterdam, Netherlands)
14:45	Newborn Screening for Lysosomal Storage Diseases, LSDs - The Italian Experience	Giancarlo la Marca (Florence, Italy)
15:00	Q&A	All
15:15-15:30	Break	
15:30-16:30	<b>Session 10: Intended and Unintended Consequences of Screening - The Patient Experience</b> <b>Chairs: Rolf Zetterström &amp; Rose Maase</b>	
15:30	What Happens After Recall, Who Should Give the Results to Families?	Trine Tangeraas Oslo, Norway)
15:45	The Maternal Stress of False Positive Newborn Screening Results	Vera Frankova (Prague, Czech Republic)
16:00	Dutch Parents' and Professionals' Perspectives on Expanding Neonatal Bloodspot Screening to Newer Disorders	Lidewij Henneman (Amsterdam, Netherlands)
16:15	Q&A	All
16:30-16:45	Break	
16:45-17:45	Commercial Workshop: Orchard Therapeutics – <a href="#">PROGRAMME HERE</a>	
17:45-18:15	 <b>Summary of Day 3 and Announcement of ISNS 2021 Luxembourg Poster Prize</b> Presented by Patricia Borde 12th ISNS European Regional Meeting President	