An accompanying event of

the Slovenian Presidency of the Council of the European Union 2021 on

Achieving Equity and Innovation in Newborn Screening and in Familial Hypercholesterolemia Paediatric Screening across Europe

11th October 2021 (09:00 – 16:00 CET)

You are kindly invited to **save the date** to attend the **virtual conference**.

The link for registration will be provided with the final programme.

Rationale

For some rare disorders, early detection offered by newborn screening (NBS) can be life changing and is able to prevent long term disability or even death.

Following the first expansion of the NBS program in 2018, Slovenia is currently planning a new expansion of NBS to detect spinal muscular atrophy, severe congenital immune disorders (18 in all), cystic fibrosis and congenital adrenal hyperplasia; in total, over 40 congenital diseases. Due to **significant technological developments in the last few years**, **expanded newborn screening** has also become **more accessible** and has been introduced in several EU countries. It is likely, in the future with a growing range of therapeutic possibilities, the need for further expansion of the NBS programme will increase markedly. The increasing use of genomics in this area is also likely with a broad range of societal, professional, and ethical implications.

Despite this progress, **the current situation regarding the NBS in the EU is rather inequitable with significant differences between individual member states**. To help achieve equity and ensure that good practice becomes common practice, a dedicated expert forum needs to be established in the EU to bring together policy makers, patient group representatives and professionals able to share experience, explore the options and offer impartial advice. The European Reference Networks for Rare Diseases (ERNs) provide a natural framework from which to assemble and support a constructive and expert group.

Familial Hypercholesterolemia (FH) is the most common inherited life-threatening disorder and a non-modifiable CVD risk factor. This most common genetic condition in the world is severely underdiagnosed and undertreated, leading to premature morbidity and mortality due to atherosclerotic cardiovascular disease. It affects 1 in 250 to 300 people around the world of every race and ethnicity.

The lack of general awareness of FH among the public and medical community has resulted in **only 10% of the FH population being diagnosed and adequately treated**. In 1998 during a consultation meeting in Geneva, Switzerland**, the World Health Organization recognized FH as a public health priority**. Little has however changed over the past 20 years to improve the situation of families affected by FH. **Majority of children identified today are non-index cases, meaning their identification is the result of their family member being previously diagnosed, often as a consequence of a heart attack, stroke or death of one of the parents.**

Slovenia has been a model country for familial hypercholesterolemia (FH) paediatric screening, alongside NBS, with an effective approach to detect this globally severely underdiagnosed inherited disorder. Recently, the Slovenian programme was identified as one of the "Best Practices" by the European Commission, and the WHF White Paper on Cholesterol recognized it as a possible model for FH-screening and potentially a model for NBS in general.

Under its EU presidency, Slovenia is spearheading an **initiative designed to enhance cooperation and equity in provision of newborn and FH paediatric screening within the EU. Several differing models of care can be recognized within the individual EU countries**. As part of this, there will be some focus on the Western Balkans region where NBS programs are less developed.

We would therefore like to invite **relevant stakeholders to engage in this technical meeting to share experience and explore potential future strategy. Key participants include policy makers at EU and national levels; patient organizations; representatives of the main professional organizations; regulators and HTA agencies.**

Target audience

- Representatives of the Institutions of **European Commission** (Commissioner, DG SANTE, DG RESEARCH, DG CONNECT, SGPP) and of the **Slovenian Government** (Ministry of Health, Ministry for Digital Transformation);
- Representatives of **WHO Europe** and **OECD;**
- Representatives of the **French, Czech and Swedish Governments (**the next presidencies);
- Representatives of **EU Member States** (EU Council) and neighbouring countries and other European countries;
- Targeted Members of the **European Parliament** (Screen4Rare, Networks of Parliamentary Advocates for Rare Diseases, MEP Heart Group) and national MPs;
- Representatives of the **Patient Organisations** (EURORDIS, IPOPI, FH Europe and its Member organizations, European Patients Forum, and other disease specific patient organisations);
- Representatives of **European Reference Networks** for Rare Diseases (ERN) - Coordinators;
- Representatives of **Board of Member States f**or the European Reference Network;
- Representatives of the **International Society for Neonatal Screening** (ISNS);
- Representatives of the **Paediatrics Societies, European Society for Immunodeficiencies, European Public Health Alliance, Health Technology Assessment International (HTAi);**
- Representatives of important **professional stakeholders** (Screen4Rare, SSIEM);
- Representatives of the **Scientific organizations connected to the FH and CVD**
- Representatives of the **European Alliance for Cardiovascular Health;**
- Representatives of the **Global FH Advocacy Group under the World Heart Federation;**
- Representatives of the Pharma and MedTech industry (EFPIA, EUCOPE, MedTech).

Objectives

- **To identify good practice in the field of NBS** in the EU;
- To **define the main gaps** and to discuss **outlines to improve the equity of provision in the field of NBS** in the EU;
- **To identify best practices** in the field of **FH paediatric screening** and how they could be replicated and taken to scale;
- To assess the fundamental challenges, barriers, and opportunities **to achieve equity of access to FH screening** across Europe;
- To explore and agree on next steps to ensure a systematic and comprehensive approach to screening in Europe that **leaves no-one behind**.

11th October 2021 (09:00 – 16:00 CET)

INTRODUCTORY PART (09:00-09:15)
Welcome: Janez Poklukar, Minister for Health, Slovenia
Opening Statement: Senior representative of a European Union Institution or Senior representative of the Slovenian Ministry of Health
Setting the scene: Tadej Battelino, University Medical Centre Ljubljana
PART I (09:15-13.00): NEWBORN SCREENING - TOWARDS EQUITY OF PROVISION IN THE EUROPEAN UNION
An overview of the current state and future of NBS in the EU on behalf of the ISNS: James Bonham
Focus on national experiences/best practices:
– **Italy -** Giancarlo la Marca
– **Netherlands**
– **Slovenia -** Urh Grošelj
– **Germany -** Stefan Koelker
– **Sweden -** Rolf Zetterstrom
– **Czech Republic -** Viktor Kožich
– **Southeastern Europe/Western Balkans -** Urh Grošelj
Stakeholders' contribution:
– **How can the European Commission support EU-wide collaboration?**
– **Screen4rare: an EU joint stakeholder initiative on Neonatal Screening (ISNS-IPOPI-ESID joint initiative):** Johan Prevot

– **The views of people living with rare diseases and their families: 11 key principles:** Yann Le Cam (EURORDIS)
– **The role of ERNs in NBS:** Maurizio Scarpa (MetabERN)
– **NBS, an EU matter:** Member of the European Parliament
Coffee break
Panel discussion - Speakers & Member States Representatives: How to improve coordination and uptake of newborn screening across EU as part of enhanced screening policy.
Debate with audience
WRAPPING-UP PART ONE - Towards equity of provision of NBS across Europe
Closing remarks
Lunch break (12:00-12:40)
SPOTLIGHT: (12:40 – 13:00) ENGAGEMENT OF EU INSTITUTIONS FOR EQUITY OF PROVISION OF NBS AND FH SCREENING ACROSS EUROPE.
PART TWO (13:00-16:00): FAMILIAL HYPERCHOLESTEROLEMIA SCREENING - CURRENT GAPS AND THE NEED FOR ACTION PLAN IN EUROPE
Patient story: My life with FH and why paediatric screening matters - Patient
Welcome: FH Europe Magdalena Daccord, Chief Executive, FH Europe
Welcome: Time for digital transformation and implications for FH paediatric screening Mark Boris Andrijanič, Minister for Digital Transformation, Slovenia
Setting the scene: Marius Geanta, Trustee, FH Europe and President, Centre for Innovation in Medicine
The wider context: CVD landscape in EU and the need for innovation MEP Heart Group Representative European Alliance for Cardiovascular Health Representative DG RESEARCH Representative
FH Europe: The unmet needs of patients with FH in Europe - Magdalena Daccord
FH screening - the Global Call to Action Recommendations Samuel S. Gidding, Senior Author of the Global Call to Action on FH, WHF Scientific Committee Member, and FH Europe Trustee
Coffee break
Global FH Registry - the paediatric data as evidence Kanika Dharmayat, EAS FH Studies Collaboration

Brief overview: Current FH screening programs in the EU and the EC Public Health Best Practice Portal Urh Grošelj, Scientific Advisor, FH Europe
Focus on national experiences/best practices:
– **Slovenia** - Urh Grošelj
– **Netherlands** - Albert Wiegman
– **Czech Republic** - Tomas Freiberger
– **Germany**
– **France**
Action Plan on paediatric FH screening in EU Albert Wiegman
Towards a Consensus on FH screening in Europe - A dialogue with key stakeholders Member States Representatives and Kausik Ray, President, European Atherosclerosis Society Raul Santos, President, International Atherosclerosis Society Fausto Pinto, President World Heart Federation Iñaki Gutiérrez-Ibarluzea, Member of Technical Advisory Group, WHO, Past President, HTAi Representatives of ESC and FH Europe
Next steps: How to move forward the Action Plan on FH screening in Europe? - a policy response President ITRE Committee European Parliament Representative of French Presidency of the Council Co-chair MEP Heart Group DG SANTE DG RESEARCH
Closing remarks - FH Europe Magdalena Daccord
Closing remarks of the whole meeting – Senior representative of the Slovenian Ministry of Health

This event is endorsed by:

- University Medical Centre Ljubljana, www.kclj.si
- Faculty of Medicine, University of Ljubljana, www.mf.uni-lj.si
- International Society for Neonatal Screening (ISNS), www.isns-neoscreening.org
- European Reference Network for Hereditary Metabolic Disorders (MetabERN), https://metab.ern-net.eu/
- International Patient Organisation for Primary Immunodeficiencies (IPOPI), www.ipopi.org
- Screen4Rare
- European Organization for Rare Diseases (EURORDIS), www.eurordis.org
- European Society for Immunodeficiencies (ESID), www.esid.org
- FH Europe, The European FH Patient Network, www.fheurope.org
- World Heart Federation (WHF), www.world-heart-federation.org
- International Atherosclerosis Society (IAS), www.athero.org
- European Atherosclerosis Society (EAS), www.eas-society.org
- European Atherosclerosis Society - FH Studies Collaboration (EAS - FHSC), www.eas-society.org/page/fhsc
- Centre for Innovation in Medicine, www.ino-med.ro
- Amsterdam UMC, www.amc.nl
- Emma kinderziekenhuis Amsterdam UMC, www.amc.nl
- Slovenian Heart Foundation, www.zasrce.si

Local Organizing Committee: Urh Grošelj, MD, PhD[1], chair; Jaka Šikonja[1]; Tadej Battelino, MD, PhD[1]; Matej Trpin[2]. [1] University Children's Hospital, UMC Ljubljana, Slovenia; [2] Republic of Slovenia, Ministry of Health

International Organizing Committee: James Bonham (ISNS), Magdalena Daccord (FH Europe), Yordan Aleksandrov (RPP Group), Nicola Bedlington (FH Europe), Tomas Freiberger (FH Europe), Marius Geanta (FH Europe), Samuel S. Gidding (FH Europe), Gulcin Gumus (EURORDIS), Corine van Lingen (MetabERN), Antoni Montserrat Moliner (FH Europe), Elliot Tricot O'Farrell (RPP Group), Martine Pergent (IPOPI), Johan Prevot (IPOPI), Maurizio Scarpa (MetabERN), Peter Schielen (ISNS), Albert Wiegman (FH Europe).