

## Celebrating the First International Neonatal Screening Day: 28th June 2021

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*Amsterdam, Netherlands; Donderry, UK; Delft, Netherlands.*

The International Society for Neonatal Screening (ISNS), in partnership with the International Patient Organisation for Primary Immunodeficiencies (IPOPI) and with the support of the European Society for Immunodeficiencies (ESID), are pleased to announce the launch of the first **International Neonatal<sup>1</sup> Screening Day**, which will be held on June 28<sup>th</sup>, 2021.

ISNS, IPOPI and ESID for some years now have been working together under the multi-stakeholder Screen4Rare initiative to promote the importance of neonatal (or newborn) screening. Screening newborns for rare but treatable diseases is possible and many screening programmes have been successfully implemented over the past decades to screen for metabolic and other inborn defects in a systematic manner. These programmes enable faster diagnosis, allowing for lifesaving and often curative early intervention and care for babies with life-threatening. Neonatal screening plays a critical role in improving healthcare outcomes for rare diseases patients. The International Neonatal Screening Day will provide a yearly momentum to draw attention to neonatal screening and its benefits and encourage collaboration and best practices as a way of ever improving routine screening and incorporating of the newest scientific evidence.

Countries vary in their approach to newborn screening and their panels of conditions to screen for vary considerably. Some conditions, such as phenylketonuria (PKU) or congenital hypothyroidism (CH) are implemented in most national programmes. Others, like severe combined immunodeficiencies (SCID) or spinal muscular atrophy (SMA) have gained a growing attention in the past decade but with some countries now screening all neonates for these conditions or supporting pilot screening projects, implementation is much more limited.

On this first International Neonatal Screening day, newborn screening experts (ISNS), patients (IPOPI) and healthcare professionals (ESID) jointly call for an increased collaboration and discussions on this key preventive measure for the wellbeing of newborns and their families.

28<sup>th</sup> June has been chosen to commemorate the birth of Dr Robert Guthrie. Dr Guthrie introduced the paper blood spot card and a new assay to screen newborns for PKU in the United States in the 1960s. Dr Guthrie's work and activities, and especially the paper blood spot card, today applied universally in all screening programmes, revolutionized the detection of children with inborn conditions, enabling the improvement of children's health.

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<sup>1</sup> ISNS, IPOPI, ESID and Screen4Rare consider 'neonatal' and 'newborn' synonyms. In most languages outside English "neonatal" is more consistent with the local language (e.g. Dutch, German, French, Scandinavian languages). Hence it was decided to use "neonatal" when ISNS was created in 1987. Still, newborn and neonatal are synonyms.

Quote from Prof Jim Bonham, ISNS President

*“For more than 50 years children around the world have been able to benefit from the life changing potential offered by Newborn Screening. Much has been achieved and much remains still to be done. The International Society for Neonatal Screening (ISNS) believes that ‘International Neonatal Screening Day’ will provide a unique chance to celebrate the vision of those who have helped make this possible and will, in turn, encourage a new generation of doctors, scientists and policy makers to extend these benefits to more children suffering from a wider range of disorders.”*

Quote from Martine Pergent, IPOPI President

*“Thanks to advances in science and medicine, babies affected with rare and devastating diseases such as SCID can be cared for and cured. The International Neonatal Screening Day is a fantastic opportunity to raise awareness on the paradigm change offered by these advances and to create a momentum where all the stakeholders can consider the best way to implement, improve or extend newborn screening practices and policies for the benefit of families and the society”.*

Quote from Prof Isabelle Meyts, ESID President:

*“The International Neonatal Screening Day is a day to be grateful for what has been achieved and a day to look forward towards implementing screening programs for additional rare diseases, which are both life-threatening and devastating if not picked up timely, such as SCID. The INSD is the day for reflexion and action on this important step in improving health at a global level”.*

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