Joining the Screen4Rare Platform

Statement of Adherence to Screen4Rare General Principles, Mission & Goals

Screen4Rare is a multi-stakeholder platform launched by the **International Patient Organisation for Primary Immunodeficiencies** (IPOPI), the International Society for Neonatal Screening (ISNS), and the **European Society for Immunodeficiencies** (ESID) aiming to exchange knowledge and best practices on NBS for rare diseases. The group's ultimate objective is, through policy engagement, to work towards ensuring that all babies can have equitable access to newborn screening; a life-saving tool for conditions such as SCID.

The present document defines the guiding principles and objectives of Screen4Rare and the working relationship within the Screen4Rare initiative for prospective members of the group.

Screen4Rare's General Principles on Newborn Screening

Since its introduction in the mid-1960s in the US and the UK, newborn screening has touched the lives of many families and their babies around the world. There is general agreement that early asymptomatic detection enabled by whole population newborn screening permits a significant life-changing and sometimes life-saving benefit and can be a cost-effective means of identifying and treating babies with Rare Disorders as part of public health provision during infancy.

Despite this, it is widely recognised that "*All screening programmes do harm; some do good as well, and, of these, some do more good than harm at reasonable cost.*" (J A M Gray, J Patnick, R G Blanks, BMJ 2008; 336:480). The assessment of the balance between the benefits and harms of screening have led to a wide disparity of practice even when countries with similar levels of economic and health care development are compared. In Europe, for example, the number of conditions included within national newborn screening programmes varies from 2 to 35. Over the last decade, much attention has been placed at trying to understand how these differences in national policy have arisen.

It is assumed, without clear justification, that countries screening for a greater number of conditions within their national or regional programmes are providing 'better' healthcare as a result. This is not always the case and despite a number of international efforts to seek convergence, differences in national policy remain. It is, therefore, helpful to try and establish a common view of *'what good looks like'* whenconsidering newborn screening and not simply to assume that more screening or screening for more diseasesmeans better screening.

Analysis suggests that the Wilson and Jungner criteria, as well as the updated WHO adaptation of those criteria published and updated in years 1968,[1] 2008[2] and 2020[3] (Appendix 1-2), are widely adopted across Europe and the world. They are seen a basis for decision-making and few would disagree that having a common understanding of the factors to consider is helpful. If anything, the Wilson and Jungner criteria

[1] Wilson, James Maxwell Glover, Jungner, Gunnar & World Health Organization. (1968). Principles and practice of screening for disease. Available at: https://apps.who.int/iris/handle/10665/37650

[2] Andermann, Anne, Blancquaert, Ingeborg, Beauchamp, Sylvie & Déry, Véronique. (2008). Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years. Bulletin of the World Health Organization, 86 (4), 317 - 319. World Health Organization. Available at: http://dx.doi.org/10.2471/BLT.07.050112

[3] WHO Regional Office for Europe. (2020). Screening programmes: a short guide. Increase effectiveness, maximize benefits and minimize harm. Available at: https://apps.who.int/iris/bitstream/handle/10665/330829/9789289054782-eng.pdf

ensure that the entire system of screening, including diagnosis as well as facilities and means to treat, are in place before screening for a given disorder commences.

However, this shared understanding does not explain the degree of diversity in national practices – even when allowing for the availability of per capita spending on health – that arises from other factors. These factors can be chiefly summarised as follows:

- The make-up of the national decision-making bodies and, indeed, whether these exist and are able to make binding national policy recommendations.
- The level of evidence required to enable decision-making.
- The role of cost-effectiveness analysis in determining policy.
- The influence and input available to groups such as patients, specialist healthcare professionals, politicians, and commercial interests.

Screen4Rare adheres to the general principles of neonatal screening as laid out in the 1968, 2008 and 2020 publications to follow its mission and achieve its goals.

Screen4Rare's Mission and Goals

Screen4Rare is a multi-stakeholder platform who look to support international, regional policy makers, and those who have autonomous national policy-making responsibility with regards to newborn screening. We aim to do so through the provision of unbiased scientific information, evidence, and comparative data so as to help ensure the best decisions are made on behalf of the populations that they serve.

Our primary goal is to help promote *'The development of appropriate, well-organised and equitable newborn screening offered on a voluntary and informed basis to families to help identify well-defined treatable conditions where it is clear that their early asymptomatic detection and treatment during childhood results in significantly improved outcome.'*

Screen4Rare activities

At the regional level

Screen4Rare has launched an EU **Call to Action on Newborn Screening for Rare Diseases**. As of August 2021, Screen4Rare's proposals for the promotion of a European approach to newborn screening has received the support of 30 Members of the European Parliament (MEPs) and 15 stakeholder organisations

Screen4Rare's approach to the topic of newborn screening has led to its collaboration with a wide range of stakeholders, from MEPs to representatives of the ERNs. Together, we advocate for the realisation of the **Screen4Rare Call to Action on Newborn Screening for Rare Diseases** which places an emphasis on a science-based and rational approach to newborn screening that supports the efforts of EU Member States looking to optimise their newborn screening programmes.

Through legislative and awareness-raising efforts, Screen4Rare seeks to identify opportunities for discussing a supportive EU approach to newborn screening for rare diseases. One of the goals of the group would lead to the development of a **European Newborn Screening Standing Committee** as recommended in the EU Tender "**Evaluation of population newborn screening practices for rare disorders in Member States of the European Union**".

At the international level

On 28 June 2021, Screen4Rare through its founding organisations (IPOPI, ISNS, and ESID) launched the **International Neonatal Screening Day**. Over the coming years, we aim to continue, in collaboration with all interested stakeholders, organising this awareness-raising day and, where appropriate, to work alongside other international initiatives to ensure that appropriate newborn and good quality newborn screening is available to those who would benefit from it.

Screen4Rare's Structure

Founding Member Organisations

Screen4rare was founded by **IPOPI**, **ISNS**, and **ESID**. These organisations meet regularly to set Screen4Rare's overarching strategy and guide the development of various workstreams within the initiative.

Partner Organisations

Screen4Rare is committed to collaborating with all stakeholders, including relevant industrial partners, with an interest in newborn screening for rare diseases. In terms of partner organisation membership status, Screen4Rare welcomes requests by patient organisations, professional groups, screening experts, physician societies, and other relevant non-industrial stakeholder organisations, to join the initiative as **partner organisations**.

Organisations who seek to be involved Screen4Rare's activities are requested to abide by the general principles, mission and goals of the platform as outlined in this document. Partner organisations will have the ability to join specific Screen4Rare meetings and provide input on certain activities specified by the founding organisations and will be recognized as Screen4Rare partner organisations on relevant online and offline official communication channels. Screen4Rare's secretariat is run with the assistance of **Bridges Consulting**, a public affairs consultancy that provides support in establishing, running, and moderating the initiative. The Secretariat is responsible for the day-to-day support and coordination of activities as decided by Screen4Rare.

Interested organisations who request to join Screen4Rare will be asked to formally accede to the principles, mission and goals outlined here above by signing this statement of adherence and:

(1) Formally endorse the **Screen4Rare Call to Action** by providing written confirmation of their support;
(2) Provide a short summary outlining the reasons for wishing to join Screen4Rare as well as their activities in the field of newborn screening.