IPOPI Response
Eurordis email consultation 6 May 2013 on EURORDIS/CNA/CEF- OPINION ON EU COLLABORATION IN THE AREA OF NEW BORN SCREENING

The following document outlines IPOPI's response to the email consultation sent by Eurordis on 6 may 2013 to rare diseases patient representatives in preparation for the next EUCERD meeting on 5-6 June during which EUCERD members are scheduled to adopt an official position on areas of collaboration on New Born Screening at the EU level.

With regards to Eurordis question "*We would like to receive your opinion as RD patients' representatives on what you deem important and feasible to implement in the field of NBS without interfering on Member States' national policy*", IPOPI believes that:

→ Action and Collaboration on Newborn Screening is necessary at EU level

- There is probably no other area in public health in which 27 national approaches could be considered to be as inefficient and ineffective as with rare diseases. The reduced number of patients for these diseases and the need to mobilise resources could be only efficient if done in a coordinated European way.
- In 2009 the Council of the EU recommended "the development of European guidelines on diagnostic tests or population screening" (Council Recommendation of 8 June 2009 on an action in the field of rare diseases, point 17.d). No guidelines have yet been produced.
- In 2011, the European Commission still was identifying "newborn screening for rare diseases" as a priority. As such, the preparation of a proposal of Council Recommendation it was envisaged by end 2012, beginning 2013. No proposal has been made by the Commission yet.
- In its work programme for 2013, the European Commission does not envisage to work on the proposal. It appears that the envisaged proposal for Council Conclusions on Newborn screening for rare diseases seems to have been postponed.
- Newborn screening is the missing piece in rare disease policy and could significantly contribute to paediatric care.

- Taking action on newborn screening for rare diseases now would ensure coherence with the European policy in the field of rare diseases. Most importantly, it is a clear sign to European patients with rare diseases that Europe is not the addition of its Member States, but a greater entity that protects citizens' lives and ensures they all have equal rights to adequate diagnosis and subsequent care, no matter how rare their disease is.

IPOPI agrees with Eurordis proposed key points including:

Production of good practice guidelines or development of "points to consider" for the management and follow-up of patients, for each screened disease.

- *IPOPI would favour "good practice guidelines" or "recommendations", rather than "points to consider" (Priority 2).*

Production of recommendations for the communication with parents with an emphasis on how to announce the diagnosis to parents whose child is screened positive but whose diagnosis is not yet confirmed. These recommendations could gather best practices sharing amongst Member States.

- *IPOPI fully agrees this should be a priority*

Increase networking between laboratories to ease collaboration and resource sharing in order to improve the quality and cost-efficiency of national operations and to prepare the laboratories to their future participation in European Reference Networks (priority 7).

- *IPOPI fully agrees this should be a priority and this is in line with other EU policies on rare diseases. An example from the PID community is the European expert committee on SCID Newborn Screening which groups together leading clinicians, researchers and patient representatives (IPOPI) to share information, expertise, data and knowledge between countries.*

IPOPI furthermore believes that point 8 "Establishment of shared databases between NBS laboratories and centres of expertise in charge of the follow-up of patients to gain better knowledge of the screened diseases and to assess the benefit of the screening strategy" is important and agrees with the United Kingdom comment made (see page 6 of the "Opinion of the EUCERD on Potential Areas for Collaboration" circulated in Eurordis' email) which stated that a more explicit emphasis on the need for a shared

approach to research for these rare conditions would be welcome. SCID Newborn screening, as highlighted in the same comment by the UK on page 6, provides a striking example of a rare disease, which can be treated and even cured through timely screening, diagnosis and treatment. It is therefore needed, that the EU takes action in order to ensure that those patients with rare diseases have access to the care and treatment they need to live a normal life. The European Parliament Environment, Public Health and Food Safety (ENVI) committee recently supported the need for an EU action on newborn screening and tabled an Oral Question on Newborn Screening for Rare Diseases using SCID NBS as a case study example, see here

On the basis of the above, IPOPI would strongly support a set of EU recommendations on newborn screening and do not believe NBS for rare diseases should only be seen as a national matter but one where Europe can bring a real added-value by providing guidance and recommendations to Member States in a growing area where most conditions eligible for newborn screening are rare diseases and by promoting increased collaboration in terms of research, data sharing, networks of laboratories and patient information.