Call to Action – Newborn Screening for Rare Diseases

The European Union's (EU) added value in the field of rare diseases is well known. From the European Reference Networks (ERNs) to the EU research funding programmes, positive strides have been taken on health-related matters. Extrapolating best practices from Member States has also played a crucial role and it is undoubtedly clear that this extrapolation has improved the lives of rare disease patients.

It is evident, however, that the EU cooperation on Newborn Screening for Rare Diseases (NBS for RD) is still an underdeveloped area. NBS for RD plays a critical role in improving healthcare outcomes and mortality for rare diseases. In the specific case of primary immunodeficiencies[1], the results from recently established programmes at national level demonstrate that newborn screening can dramatically improve the outcomes for infants with Severe Combined Immunodeficiencies (SCID).[2]

In the field of rare diseases, gathering data on prevalence through regional or national pilot projects over short periods of time can be problematic, especially in those countries with low population or birth rates. For this reason, there is an urgent need to improve the mechanisms by which NBS is assessed and adopted by different EU Member States, for all newborn EU citizens to have the same access to life-saving preventative programmes. Currently, there is no EU centralized body or resource that can act to recommend, guide or share data on newborn screening practices. This leads to the slow uptake of newborn screening in Member States, a live saving tool for babies.

In light of the above, we, the undersigned Members of the European Parliament, together with the supporting organisations, including Screen4Rare, a multi-stakeholder initiative launched by the International Patient Organisation for Primary Immunodeficiencies (IPOPI), the International Society for Neonatal Screening (ISNS), the European Society for Immunodeficiencies (ESID), are calling on the European Commission and Member States to:

1. Develop and implement **overarching guidelines in the field of newborn screening for rare diseases.** This will ultimately:
    a. Assist Member States in having a much better-informed assessment of a new NBS technique.
    b. Help Member States in preparing their healthcare systems for the implementation of a NBS for a specific rare condition.

2. Work together within the European Commission Steering Group on Health Promotion and Prevention to **develop an EU-wide platform on newborn screening for rare diseases and pool from the expertise of recognised societies working in the field**. This can be achieved by following-up on the 2012 report on practices in the field of neonatal screening for rare disorders where several crucial recommendations were identified **including the creation of a European newborn screening standing committee** and a recommendation stating that EU collaboration could have significant added value in encouraging uptake of the newest scientific evidence in newborn screening among Member States.

3. Cooperate to position the EU as the central point for data collection and information on rare diseases newborn screening practices and encourage the exchange of best practices that would support Member States' efforts in the development of newborn screening practices according to the existing science, knowledge, and know-how.

[1] A large group of more than 380 rare and congenital rare disorders in which the immune system does not work properly or at all, leading to an increased susceptibility to infections, virus and bacteria, that can lead to organ damage or become even fatal.
[2] Severed combined immunodeficiency (SCID) is a rare, fatal syndrome of diverse genetic cause in which there is a combined absence of T-lymphocyte and B-lymphocyte function. These defects lead to extreme susceptibility to serious infections that can result in permanent organ damage or failure or even death unless definitive treatment can be used to correct the underlying immune defect. Early diagnosis, before the infant has had a chance to develop any infections, is extremely valuable since bone marrow transplants or blood or gene therapy given in the first 3 months of life have a 96% success rate.

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IPOPI INTERNATIONAL PATIENT ORGANISATION FOR PRIMARY IMMUNODEFICIENCIES

ISNS International Society for Neonatal Screening

ESID

Supporting Organisations:

AEDIP

APDIP 17ANOS

ARPID

dsai Defektes Immunsystem? Starke Patientenorganisation!

SAS

irfs ASSOCIATION DE PATIENTS DÉFICITS IMMUNITAIRES PRIMITIFS

European Reference Network MetabERN European Reference Network for Hereditary Metabolic Disorders

PIO Primär immunbrist organisationen

Partner Organisations: