TACKLING DIAGNOSTIC ODYSSEY IN RARE DISEASES THROUGH ERNS: THE CASE OF PIDs

Recommendations of the International Patient Organisation for Primary Immunodeficiencies

December 2017

Introduction

IPOPI is the International Patient Organisation for Primary Immunodeficiencies. As the association of national patient organisations, it is dedicated to improving awareness, access to early diagnosis and optimal treatments for primary immunodeficiency (PID) patients worldwide.

Similarly to other rare diseases, PID diagnosis is still not optimised across the European Union and many patients do not receive an accurate and timely diagnosis which is crucial for successful PID management and care. Poor diagnosis in the rare disease field is often characterised by both low awareness of the condition and limited access to reference centres specialised in rare disease diagnosis, treatment, and care.

One of the major advances of the 24 ERNs launched in March 2017 is the potential to improve the speed and accuracy of diagnosis, improve and harmonise the management of patients in the EU, and support health professionals in sharing information and data.

IPOPI's recommendations on ERNs and diagnosis are aimed to provide the EU and the Member States with guidance on the steps to take in order to enhance the role of ERNs with regards to rare diseases diagnosis, with a focus on PIDs.

ERNs and PID diagnosis

ERNs have a great potential to improve the speed and accuracy of diagnosis, harmonise the management of patients in the EU, and support health professionals in sharing information and data. ERNs have the potential of being beneficial especially for member states with less diagnostic means and expertise. However, insufficient knowledge on how to use ERNs, potential issues with reimbursement and lack of continuous funding might significantly impair ERNs' benefits for rare disease patients.

Many PIDs are complex and require genetic testing to ensure the best diagnosis possible. Genetic diagnosis for PIDs is not available in some EU countries. In other cases, the specific PID is not so complex and can be diagnosed by an immunologist. The problems arise when the general practitioners and other healthcare professionals do not suspect a PID and patients are confronted with many tests and visits to specialists before getting the right diagnosis.

Severe Combined Immunodeficiency (SCID) is the only screenable PID condition that can be cured with treatments such as Hematopoietic Stem Cell Transplantation (HSCT) or gene therapy. SCID newborn screening is a cost-effective method of diagnosing patients and saving lives, fulfilling all the all the internationally recognised criteria for a clinical condition to be screened at birth.

RECOMMENDATIONS

EU

- The EU should develop a common approach on harmonized access to a newborn screening in all Member States, which is crucial for all rare diseases but particularly for some PIDs such as Severe Combined Immunodeficiency (SCID) where a curative treatment exists if detected early.

- A standardized and uniform European-wide approach for genetic molecular diagnosis of PIDs is needed to ensure access for genetic diagnosis for all patients.

- Based on the ERNs' performance and identified need, the EU should continue funding ERNs to improve diagnosis and outcomes of rare disease patients', including those of PID patients.

Member States

- ERN Coordinators should consider, where relevant, organising a working group on newborn screening within relevant ERNs, to provide the Commission with scientific proof from the Member States.

- Member States should ensure the use of advanced cross-border information technologies and raise further awareness on PID for better patient referral.