Joint Committee

on

Health

OPENING STATEMENT

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Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

Introduction

Good morning Chairman and members. Thank you for the invitation to meet with the Joint Committee on Health to discuss improving the lives of those affected by rare diseases and also the issue of genetic testing of newborn babies for rare diseases. I am joined by my colleague(s):

- Dr Ciara Martin, Consultant Paediatrician (Emergency Medicine) and National Clinical Advisor and Group Lead, Children and Young People.
- Mr Shaun Flanagan, Assistant National Director, Primary Care Reimbursement Service (PCRS).

With the support of the Department of Health, and patients and their families, the National Rare Diseases Office (NRDO) was established by the HSE in 2015. This was a key recommendation of first Irish National Plan for Rare Diseases (2014-2018). The HSE and Department of Health each have a role in this plan for patients with Rare Diseases.

Since establishment of the NRDO, Information Scientists have provided a free information helpline service for members of the public, patients and health care staff. Information provided includes current and reliable, evidence based information about rare diseases and informs patients where in Ireland the centres of expertise are located. They also give information about support groups for people living with rare diseases. The NRDO maintains and updates a database on rare diseases called Orphanet Ireland. The work of the NRDO in collaboration with multi-disciplinary clinical staff and RD patient representatives, in relation to care pathway development, reduces waiting times to see a rare disease specialist and shortens the time to a diagnosis which we know can be prolonged for people living with rare diseases.

The European Commission Cross Border Directive (Article 12 of EC Directive 2011/24/EU) supports strengthening co-operation between highly specialised healthcare providers across the European Union by the establishment of a system of European Reference Networks. The NRDO is the co-ordination hub for Ireland's European Reference Networks. A total of 18 collaborative groups from over 40 centres of expertise in Ireland, led from 5 major academic HSE teaching hospitals, have received full European Reference Network (ERN) membership to date.

ERN membership facilitates sharing of information and access to expertise across Europe. It provides Irish patients and healthcare professionals access to the latest research and education opportunities. ERNs are predicted to greatly improve the access and quality of care for Irish rare diseases patients, including access to participation in clinical trials, where Ireland may have limited capacity due to our small population. Funding will be sought in the Estimates process to support these 5 hospital sites to set up their rare disease registries and to support clinicians to participate in research.

The NRDO is currently in the process of designing care pathways, including paths to diagnosis and genetic testing, across the 18 ERNs. These pathways set out a multidisciplinary approach to care for the more common rare diseases which would address current service gaps, particularly in psychological and social care support and timely access to genetic testing and counselling.

The NRDO has led on and ensured that Ireland is a participant in a number of key EU grants related to rare diseases. There is now an opportunity to participate in an upcoming EC ERN 2023 Integration Grant (Joint Action) with further grant opportunities thereafter.

The HSE intends to work closely with the Department of Health on developing an updated plan for rare diseases. As with the National Rare Diseases Plan for Ireland 2014-2018 the involvement of patients and their families living with rare diseases will be fundamental to the new plan.

With regard to the issue of genetic testing of newborn babies for rare diseases, the purpose of population screening is to reduce death and illness and improve quality of life through early detection and treatment of disease.

In Ireland, the National Newborn Bloodspot Screening Programme is responsible for the 'heel prick' test for babies when they are 3 to 5 days old. This test identifies treatable metabolic disorders and other inherited or congenital disorders in infants and allows for timely intervention and treatment. The programme currently screens for nine disorders.

In 2019, a National Screening Advisory Committee (NSAC) was established. This is an independent advisory committee which advises the Minister and Department of Health on all new proposals for population-based screening programmes and revisions to existing programmes. The Committee plays a significant strategic role in the development and consideration of population-based screening programmes in Ireland.

In November 2021, the NSAC had a public call for submissions which was open to patients, public and health professionals and open to proposals for a new screening programme or changes to one of the existing programmes. All areas of national screening were considered including newborn screening.

In addition, in recent weeks the HSE has established an expert steering group, including patient and patient advocacy representation, to develop a National Genetics and Genomics Strategy for Ireland. This group will develop an agreed strategy and implementation plan for genomics in Ireland. This work will improve access for patients to genetic expertise and testing which can impact timely diagnosis and treatment.

This concludes my Opening Statement.

Thank you.