Rannán na nOspidéil Ghéarmhíochaine
Aonad 4A, Áras Dargan
An Ceantar Theas
An Bóthar Míleata
Cill Mhaighneann
Baile Átha Cliath 8

Acute Operations
Unit 4A, The Dargan Building
Heuston South Quarter
Military Road
Kilmainham
Dublin 8

18th April 2024

Deputy Colm Burke
Dáil Éireann,
Leinster House
Kildare Street
Dublin 2

PQ 14226/24 To ask the Minister for Health what steps he will take to address delays in diagnosis and treatment of rare diseases in light of the fact that more than a third of people with such conditions go for more than five years without a diagnosis and in recognition of the 300,000 people living with such diseases in Ireland; and if he will make a statement on the matter

Dear Deputy Burke,

The Health Service Executive has been requested to reply directly to you in the context of the above Parliamentary Question, which you submitted for response. I have examined the matter and the following outlines the position.

In 2022, the HSE published the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. The strategy outlines related key areas of focus that include ensuring equity of access to genomic testing, integrating advances in genomic testing and improving referral pathways. In 2023 the National Genetics and Genomics Office was tasked with implementation of this strategy. Over 72% of rare diseases are of genetic origin. Improving the patient experience in accessing clinical genomic services is recognised as a key contributor to the delivery of high quality services for a patient with a rare, undiagnosed disease. The development of these services aims to reduce time to diagnosis and generate improved health outcomes for patients with a rare disease in Ireland.

The National Rare Diseases Office (NRDO) assists in identifying the correct health care sites nationally for rare disease diagnosis and treatment. The NRDO has mapped out 113 centres of expertise for rare diseases in Ireland to Orphanet, the international rare disease database, which can be assessed by healthcare professionals and people living with a rare disease.

European Reference Networks (ERNs) aim to enable discussion on rare and complex conditions that require specialised treatment, and concentrated knowledge and resources. Ireland is current a member in 18 ERNs. This coordination and sharing of knowledge across European networks will enable healthcare providers to access a larger pool of expertise and to improve diagnoses and treatment, in particular when the expertise is not available in Ireland. The Department of Health is supporting Ireland's participation in the current EU4Health ERN Integration grant 'JARDIN' which includes development of a signposting tool for use by families and GPs to improve access to diagnosis and treatment.

The NRDO is currently leading out on the development of a number of integrated care pathways for rare diseases, which aligns with the EU4Health JARDIN project. The care pathways aim to improve patient outcomes by facilitating more timely diagnosis and reduced waiting times, and by supporting delivery of integrated multi-disciplinary care.

In the context of treatment of rare diseases, in 2018, as recommended by the National Rare Disease Plan for Ireland 2014-2018, the Rare Diseases Technology Review Committee (RDTRC) was established. To date eight Orphan Medicinal Products (OMPs) have been referred to and reviewed by the RD TRC. Six of which received a positive reimbursement decision from the HSE Executive Management team

and are currently available for prescribing and reimbursement in accordance with HSE clinical guidelines.

In February 2023, Minister for Health Stephen Donnelly announced the development of a new National Rare Disease Plan. This new plan will set out the vision for rare disease services in Ireland and the actions required to achieve this including a focus on ERN national support and integration.

I trust this information is of assistance.

Yours sincerely,

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Catherine Clarke
Assistant National Director, Acute Operations