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

Acute Operations
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Kilmainham
Dublin 8

11th July 2022

Deputy Bernard J. Durkan,
Dáil Eireann,
Leinster House
Kildare Street
Dublin 2

PQ 36105/22* "To ask the Minister for Health the degree to which research continues into rare diseases in this country by means of the highest possible level of scientific research, reference to centres of excellence globally; the extent to which, in Ireland that it is possible for persons suffering from rare diseases to obtain early diagnoses and remuneration as appropriate; and if he will make a statement on the matter".

Dear Deputy Durkan,

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.

The Health Research Board (HRB) is a State Agency under the Department of Health and is responsible for funding, co-ordination, and oversight of medical research. In the last 10 years the HRB has invested €14M in rare disease research. Since 2006, the HRB has operated a joint funding scheme with Health Research Charites in Ireland (HRCI) which funds internationally competitive and innovative research in areas of strategic relevance to HRCI-registered research charities.

The HRB is a member of the European Joint Programme on Rare Diseases, specifically contributing co-funding for successful Irish research partners. Discussions are at an advanced stage to shape a new EU Partnership on Rare Diseases, which will begin in 2024 and ensure continuity on the European Joint Programme on Rare Diseases. HRB are actively participating with the DoH, HSE and others to ensure participation from Ireland.

The HRB announced a new Clinical Trials Network in Rare Diseases earlier this year which will connect clinicians, researchers, trialists, patients and others nationally, participate in European-wide funding calls and develop collaborative links through the European Reference Networks to further support and progress the research agenda in rare diseases. In addition, Ireland's membership of the European Clinical Research Infrastructure Network (ECRIN) gives access to much larger trials than could be delivered in Ireland alone and provides real opportunities for trials in rare conditions.

In relation to obtaining early diagnoses, all infants born in Ireland are offered new-born bloodspot screening between 72 and 120 hours after birth. New-born bloodspot screening is a positive contributor to the early diagnosis of nine specified and serious rare diseases in Ireland and has an extremely high uptake rate.

The National Screening Advisory Committee (NSAC), based in the Department of Health, is responsible for reviewing the evidence and making recommendations to the Minister for Health whether to add new conditions to the New-born Bloodspot Screening Programme. The NSAC has developed its own criteria for appraising the viability, effectiveness and appropriateness of a screening programme which includes requests to add new conditions or make changes to an existing screening programme.

There are a number of supports available to families of NBS screened disorders to help relieve some of the financial burden which can include the following:

- Babies identified with Phenylketonuria (PKU) and Cystic Fibrosis (CF) are entitled to a long term illness (LTI) card
- Babies detected as having any of the other conditions identified via NBS can be assessed for a medical card
- If deemed not eligible for a medical card; medications, specific named dietary products etc. fall under the Drug Payment Scheme

I trust this answers your question to your satisfaction.

Yours sincerely,

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Brian Dunne
General Manager, Acute Operations