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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Dublin 8

21st July 2022

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

PQ 36116/22* "To ask the Minister for Health the number of patients throughout the country allegedly suffering from a rare disease but whose condition remains undiagnosed; if such patients are deemed to be suffering from a rare disease; if a sufficiently coordinated action plan will be put in place to meet their requirements in early date; 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.

Currently there is no central mechanism in place such as a national rare diseases registry for rare disease surveillance, registration and reporting of diagnosed and undiagnosed patients. The accurate registration of diagnosed and undiagnosed rare disease population data requires the implementation of Orphacodes (the rare disease specific coding system recommended by the EC as best practice), the establishment of integrated electronic health records and the use of the unique patient identifier. The Orphacode system also includes codes for undiagnosed rare diseases. The development of rare disease registries is a key recommendation of the National Plan for Rare Diseases (2014-2018) and is central to monitoring the national prevalence and incidence of rare diseases. The development of rare registries is also a key function of the European Reference Networks (ERNs) (established under the European Commission Cross Border Care Directive 2011/24/EU). The ongoing integration of ERNs within our national health service will further contribute to capturing rare disease data.

Nonetheless, I can advise that a recent Irish study (published by the European Journal of Human Genetics), "An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature" (Gunne et al, 2022) has indicated that 4.5% (2,283) of children born in the year 2000 have an identified rare disease. Extrapolating this data to include adults would suggest that an estimate of 5.95% of the Irish population (1 in 17) likely have a rare disorder.

I trust this answers your question to your satisfaction.

Yours sincerely,

Helen Byrne
Assistant National Director, Acute Operations