



Rannán na nOspidéal 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
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Dublin 8

28th November 2022

Deputy Peadar Tóibín
Dáil Eireann
Leinster House
Kildare Street
Dublin 2

PQ 56155/22- To ask the Minister for Health the definition of "rare disease" utilised by his Department and the HSE; and the number and names of the rare diseases entailed under these respective definitions provided in tabular form.

Dear Deputy Tóibín,

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.

A 'rare disease' is defined in Europe as a life-threatening or chronically debilitating disease affecting no more than 5 people per 10,000. There are an estimated 6-8,000 known rare diseases affecting up to 6% of the total EU population, (at least 30 million Europeans), and at least 300,000 Irish people during their lives. This definition is utilised by the Department of Health and the HSE.

Orphanet, the European portal for rare diseases and orphan drugs, hosts information on rare diseases including an inventory, classification and encyclopaedia of rare diseases and genes involved, a directory of patient organisations, and a directory of expert centres. Please see link to a list of rare diseases currently hosted on Orphanet which includes names and ORPHAcodes of 6227 rare diseases – [Orphanet Rare Disease List - 2022](#). ORPHAcodes are recognised as the most appropriate nomenclature for clinical coding of rare diseases in Europe in a dedicated recommendation from the European Commission Expert Group on Rare Diseases (CEGRD, 2014).

For prevalence and incidence data on more common rare diseases, please see link to Prevalence and incidence of rare diseases: Bibliographic data: https://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf. Incidence and prevalence data provided are worldwide estimates and European estimates where global data is not available.

I trust this information is of assistance to you but should you have any further queries please contact me.

Yours sincerely,

A handwritten signature in black ink, appearing to read 'Brian Dunne'.

Brian Dunne
General Manager, Acute Operations