

Rare Diseases in Ireland

A disease that affects fewer than 1 in 2,000 people

live with a rare disease





6,000+ distinct rare diseases

72% genetic origin



● ● • 70% with first symptoms appearing in childhood

5.100+ ultra-rare diseases each one affecting < 5 people in Ireland



Rare diseases in children living in Ireland

4.2% children diagnosed with a rare disease by age 17 ~2,000 (3.7%) teens with a lifelong rare disease transition each year to adult care

Children with rare diseases use 25 times more as compared to their peers

11.8% mortality by age 18 in those living with rare diseases 58.6% deaths in those <15 years old have an underlying rare disease

51.9% children's are used by the 4.2% living with a rare disease



Most rare diseases are life-long, chronic and disabling, and sometimes progressive and life-threatening.

Gunne E, Lambert DM, Ward AJ, Murphy DN, Treacy EP, Lynch SA. An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature. Eur J Hum Genet. 2022 Nov;30(11):1211-1215 Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rare Diseases Rath A. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Ireland Eur J Hum Genet. 2020 Feb;28(2):165-173.



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