



Rare Diseases  
Ireland



# Newborn Bloodspot Screening (NBS) - 'heel prick' test -

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# Rare Diseases in Ireland....

## What is a RARE DISEASE?

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

**1 in 17 people**

live with a  
rare disease



**300,000  
in Ireland**



**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**



Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rath A.  
Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* 2020 Feb;28(2):165-173.

# Impact on children in Ireland....

**4.2%** children diagnosed with a rare disease **by age 17**

**~2,000** (3.7%) teens with a life-long rare disease **transition each year** to adult care

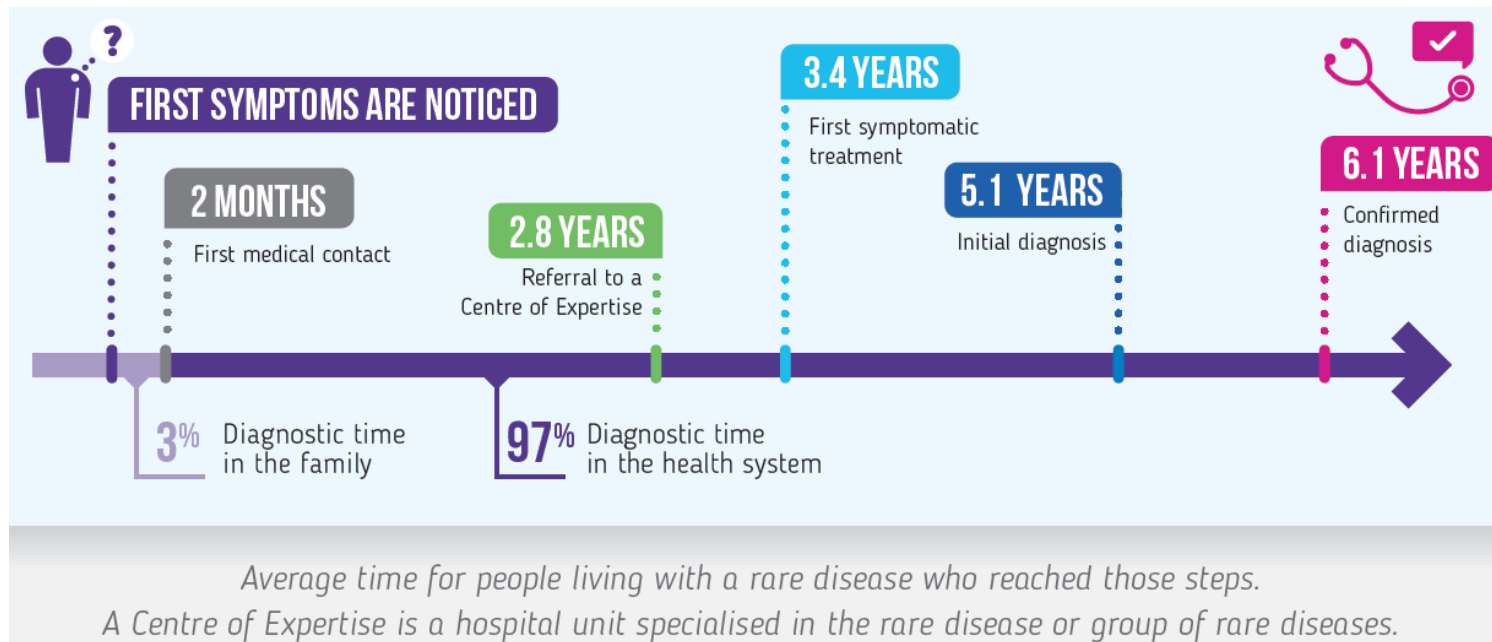
**11.8% mortality** by age 18 in those living with rare diseases

**58.6% deaths** in those **<15 years old** have an underlying rare disease

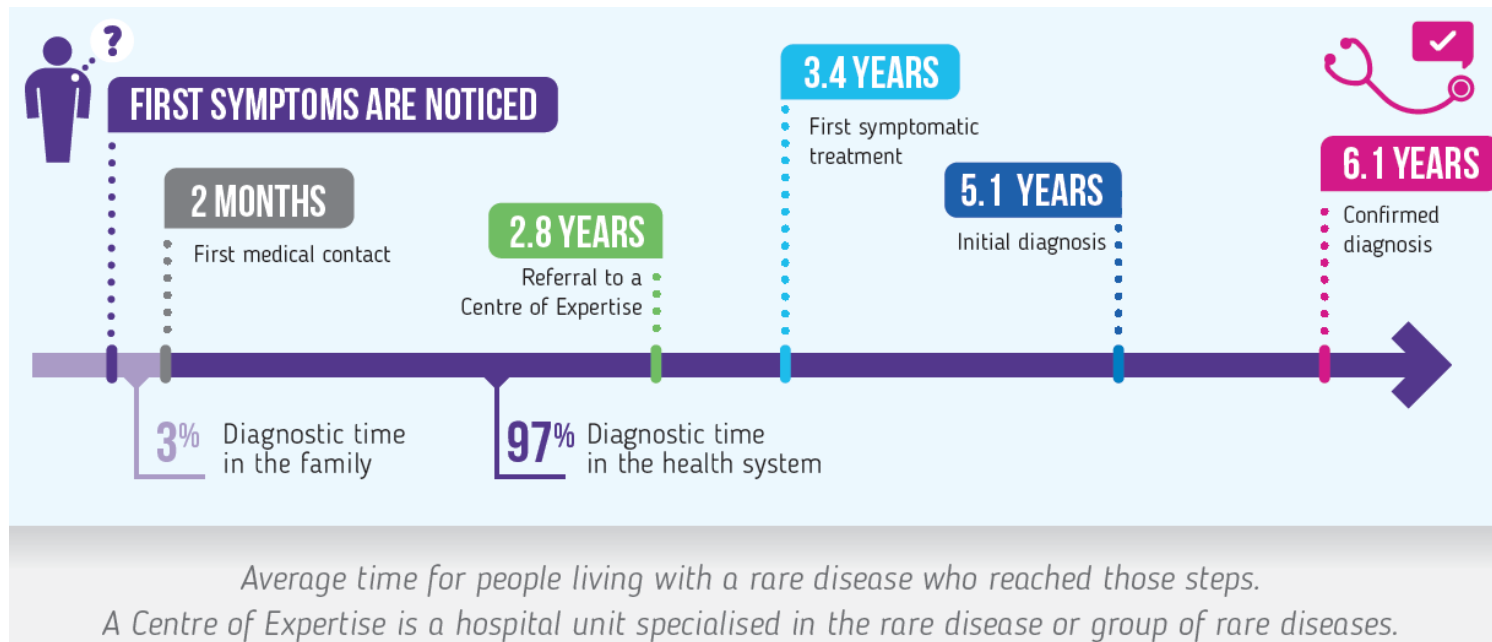
**Most rare diseases are life-long, chronic and disabling, and sometimes progressive and life-threatening.**

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

# Diagnostic odyssey in Ireland



# Diagnostic odyssey in Ireland



**Average time to confirmed diagnosis across Europe: 4.7 years**

# Impact of diagnostic odyssey

- **Missed opportunity** to access most appropriate treatment early
- Deterioration in **physical & mental health**
- Exposure to **potentially harmful treatment**
- **Undermines confidence** in healthcare system
- **Wasteful** of scarce resources
  - Healthcare provider time
  - Cost of unnecessary treatment



**60%**

have been **misdiagnosed** with another **physical disease**.



**72%**

have been **misdiagnosed** with another **psychological disease** or had their symptoms not taken seriously.



**34%**










had **8 consultations or more** with healthcare professionals before their diagnosis was confirmed.

Rare Barometer – EURORDIS-Rare Diseases Europe – 2024 May.

# NBS leader to laggard...

- Feb 1966 (established)
  - first national screening program in the world
- Today
  - **Ireland screens newborns for less diseases than 75% of European countries**
  - **only 1 disease implemented into NBS program at HSE over last 5 years**

The NNBS provides a quality assured programme for the following conditions which are listed based on when screening commenced in Ireland:

	<b>1966</b> - Phenylketonuria (PKU)
	<b>1971</b> - Homocystinuria (HCU)
	<b>1972</b> - Classical Galactosaemia (CGAL)
	<b>1972</b> - Maple Syrup Urine Disease (MSUD)
	<b>1979</b> - Congenital Hypothyroidism (CHT)
	<b>2011</b> - Cystic Fibrosis (CF)
	<b>2018</b> - Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)
	<b>2018</b> - Glutaric Aciduria Type 1 (GA1)
	<b>2022</b> - Adenosine Deaminase Deficiency Severe Combined Immunodeficiency (ADA-SCID)



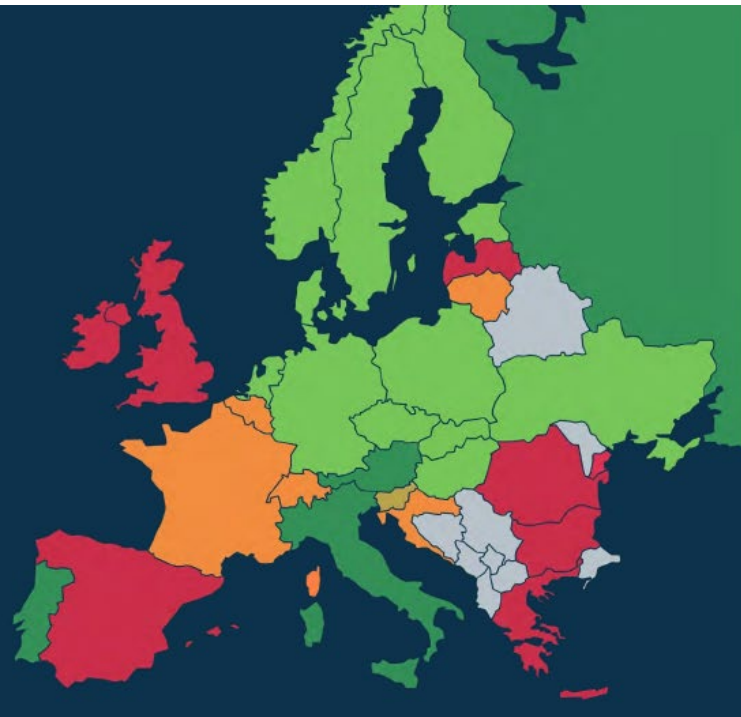
# Variation in screening (Europe)

## Variations in newborn screening across Europe<sup>1</sup>

Number of diseases screened for at birth

- **30+:** Russia, Portugal, Austria, Italy
- **20+:** Slovakia, Ukraine, Poland, Hungary, Czech Republic, Estonia, Germany, Finland, Denmark, Norway, Netherlands, Sweden
- **15+:** Slovenia
- **10+:** Croatia, Lithuania, Belgium, Switzerland, France
- **<10:** Ireland, Latvia, Bulgaria, Romania, Cyprus, Malta, Greece, Luxembourg, UK, Spain

**18 - average number diseases in NBS panels**





# Expanding NBS program

- 2-step process
  - NSAC recommendation/Minister approval
  - HSE implementation
- 9 conditions in NBS panel today
  - only ADA-SCID added at HSE over last 5 years
  - other SCID types & SMA approved, but NOT implemented
- 34 conditions awaiting NSAC assessment
  - Each assessment approx. 12-18 months

# Our ask...

- At least **double the number of diseases implemented in NBS panel over next 5 years (by 2029)**
  - HSE adopt systematic approach to implementing NSAC recommendations
  - DoH/NSAC coordinate assessment and approval at European level – eliminate duplication and 18-month assessment time

Maximise benefit of screening program:

**Save lives & health of  
~200 newborn babies annually**

# Thank you!



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