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National Rare Disease Strategy for Ireland Public Consultation

Fields marked with * are mandatory.

National Rare Disease Plan for Ireland - Public Consultation

Public Consultation on the development of a new National Rare Disease Strategy for Ireland.

New National Rare Disease Strategy

A ‘rare disease’ is defined in Europe as a life-threatening or chronically debilitating disease affecting at most only 5 people out of every 10,000. However, there are more than 6,000 known rare diseases affecting up to 6 per cent (6 out of every 100) of the total EU population (at least 30 million Europeans). This means it is common for people in the population to be affected by one of the rare diseases.

At any time, about 300,000 people in Ireland are living with a rare disease. Rare diseases present unique challenges for patients, their families, and the wider healthcare community. This is why we need to take a national, strategic approach to rare diseases.

First National Rare Disease Plan published in 2014

The first National Rare Disease Plan for Ireland 2014-2018 was published in 2014. That Plan contained 46 recommendations focused on:

improving diagnosis identifying centres of excellence setting up dedicated rare disease treatment pathways.

Progress with this plan included:

- setting up the National Rare Disease Office in the HSE

- publishing a Model of Care for Rare Diseases
- Ireland joining 18 out of the 24 European Reference.

(European Reference Networks are virtual networks involving healthcare providers across Europe. They aim to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, and knowledge and resources.)
However, there were other areas:

- that did not progress as much as expected
- where new technologies and advancements overtook the recommendations in the.

Time to update vision and aims
It is now time to renew our focus on rare diseases and update our vision and aims. We want to make sure that a new strategy meets the needs of people living with a rare disease and their families.
This consultation looks at what areas are important to the many stakeholders across the rare disease community, healthcare providers, the public and others. We aim to make sure that the new National Rare Disease Strategy responds to their needs.

Identifying improvements that will have the biggest impact
We would like to find out which improvements would have the biggest impact on rare disease services in Ireland as they affect people you represent.
We will use the results of this consultation to help the National Rare Disease Steering Group develop a new National Rare Disease Strategy for Ireland. This Strategy will also be influenced by:
further evidence reviews expert advice experience of people living with a rare disease.
We want to make sure that any recommendation:

- is achievable
- is effective
- addresses the needs of the rare disease community.

You can get more information on the National Rare Disease Steering Group here:
https://www.gov.ie/en/collection/85f8b-minutes-and-agendas-from-meetings-of-the-national-rare-disease-steering-group/#2023

Privacy Policy

As we are in the process of developing new Rare Disease Strategy, we are seeking your views in this public consultation. To do so, we must have a lawful basis to use the personal information you supply.
The lawful basis under the GDPR for processing your information is because: 6(1)(e): processing is necessary for the performance of a task carried out in the public interest or in the exercise of official authority vested in the controller 9(2)(i): Processing is necessary for reasons of public interest in the area of public health.
We will treat all information you give us sensitively, and we will only use the information you have provided to us to assist in developing a new Rare Disease Strategy.
Any information provided will be stored securely, with access to your personal data limited to relevant team members only. Only aggregated data will be shared with the National Rare Disease Steering Group.

* We will hold all information until December 2024. After that, we will dispose of the information, as we are obliged to do, in line with the National Archives.

- (●) I confirm I have read the above text

About You

* Are you currently a resident of/is your organisation based in Ireland?
  - (●) Yes
  - ( ) Yes - Northern Ireland
  - ( ) No

* Are you responding as a member of the public or on behalf of an organisation?
  - ( )

- [ ] Member of the public
- [x] On behalf of an organisation

About You

About You

* Which one of the following best describes your organisation?
  - [ ] A public health body
  - [ ] An advocacy group for people living with rare diseases
  - [ ] An advocacy group for carers
  - [x] Non-Governmental Organisation (NGO)
  - [ ] Medical organisation
  - [ ] Industry providing products or services for people living with rare diseases (e.g. Pharmaceutical or biotechnology company, laboratory, digital tools etc…)
  - [ ] Commercial lobby group

If comfortable, please share with us the name of your organization:

200 character(s) maximum

Pavee Point Traveller and Roma Centre

Current Rare Disease Services in Ireland

Does the condition or the syndrome your organisation represents have a name?

- [x] Yes
- [ ] No
- [ ] Don't know
- [ ] Prefer not to say

If comfortable, please share with us the name of any conditions or syndromes your organisation represents?

We represent Travellers with higher incidences of inborn errors of metabolism and/or immunity.

What do you think are the greatest challenges faced by people living with rare diseases? (Choose your top 3)

Maximum 3 selection(s)

- [x] Time until a diagnosis is made or you get the right diagnosis. This includes access to genetic testing
- [ ] Lack of awareness of rare diseases amongst healthcare professionals
- [ ] Access to specialist medical care and treatment
- [ ] Access to disability services and supports
- [ ] Coordination of care between:
  GP and hospital care
  different hospital specialists
  health services and social care services
- [ ] Access to drugs and innovative technologies
- [ ] Availability of information about rare diseases
- [x] Availability and provision of wider support for rare diseases, for example:
  social care supports
  mental health support services
  special education supports
- [ ] Need to pay privately for wider support services like access to psychology support
- [ ] Ability to participate in research opportunities, including clinical studies and trials
- [ ] Opportunities to access medical opinions from international experts
- [x] Transition from child to adult services

Are there any other challenges which were not included which you would include in your top three?

2000 character(s) maximum

The lack of routine data collection with an ethnic identifier in healthcare systems is a significant challenge for Travellers who are diagnosed with rare diseases. Travellers are disproportionately affected by the incidence of rare disease, but little is known about the exact incidence, diagnostic pathway, treatment and health outcomes for Travellers without disaggregated data collection.

Pavee Point welcomes the introduction of SCID to be added to the National Newborn Screening Service; Ireland is far behind international counterparts in the introduction of screening for Hurlers Syndrome also. Given the increased incidence of these conditions among Travellers, they are disproportionately affected by delays in introducing tests to the Newborn Screening Service.

What do you think could be done to address the challenges which you selected?

4000 character(s) maximum

Travellers have a higher incidence of rare diseases such as inborn errors of metabolism and immunity in comparison to the majority population, with increased prevalence of conditions such as galactosaemia, hurlers syndrome, and SCID. Travellers experience health inequalities and poorer health outcomes compared to the general population indicating an urgent need to address this issue.
The previous “National Rare Disease Plan for Ireland” recognises the higher incidences of certain rare diseases among Travellers, however there are no specific targeted measures to ensure these conditions are screened, diagnosed and treated among Travellers in the plan. To address this, the new Strategy should explicitly include both targeted and mainstreaming measures to address the specific health needs of Travellers. Engagement with the Traveller health infrastructure, via the established Traveller Health Units and local Traveller organisations/Traveller Primary Health Care Projects will ensure actions in the Rare Disease Strategy are informed by Traveller Community Healthcare Workers who are familiar with the issues Travellers face. Pavee Point seeks partnership working in this area to ensure action plans are inclusive and culturally appropriate.
The particular issue of racism and discrimination within health services is well-established in relation to Travellers. Consanguinity is often identified by health providers as a key determinant for higher incidence rates of rare diseases amongst Travellers, however, careful consideration must be provided as Travellers report effectively being blamed for having children with rare diseases. This creates further stigma and disengagement, which can prevent Travellers from accessing services such as fast-tracked prenatal screening for galactosaemia.
Travellers also face significant barriers in accessing healthcare, with half of Travellers reporting poor functional literacy. When diagnosed with a rare disease, therefore, Travellers face compounding challenges in navigating the complex care pathways for rare diseases; coordinated care, with a specific focus on transitions from child to adult services, is of particular importance. An assigned point of contact in this care pathway should be allocated to patients to coordinate car and build trust for Traveller families.
The following actions are required:
- Introduce targeted measures to increase the provision of support to Travellers with rare diseases, given the marginalisation and discrimination they experience across all domains of society.
- Embed nationally the practice of collection and publication disaggregated data collection in healthcare systems, including an ethnic identifier to ensure Travellers have access to health screening measures.
- Anti-racism and discrimination training for health providers in line with existing policy and legal obligations (e.g.) Section 42, IHREC Act, 2015.Add SCID to the Newborn Screening Service.
- Engage with the existing Traveller healthcare infrastructure and local Primary Healthcare for Travellers Projects in order to introduce meaningful change for Travellers with rare diseases.
- Introduce a healthcare professional as a single point of contact for Traveller families throughout the disease care pathway.
- Provide targeted information for healthcare professionals, particularly maternity hospitals and GPs with Traveller patients on the availability of prioritised and fast-tracking of prenatal galactosaemia genetic testing for Traveller women who are pregnant. Resources need to be made available to roll this out nationally.

Your Experience of Rare Disease Services in Ireland

What do you think Ireland does well in supporting or caring for people with a rare disease?

4000 character(s) maximum

Contact

Contact Form