



**Health
Information
and Quality
Authority**

An tÚdarás Um Fhaisnéis
agus Cáilíocht Sláinte

Protocol for review of national rare disease strategies in selected countries

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About the Health Information and Quality Authority

The Health Information and Quality Authority (HIQA) is an independent statutory authority established to promote safety and quality in the provision of health and social care services for the benefit of the health and welfare of the public.

HIQA's mandate to date extends across a wide range of public, private and voluntary sector services. Reporting to the Minister for Health and engaging with the Minister for Children, Equality, Disability, Integration and Youth, HIQA has responsibility for the following:

- **Setting standards for health and social care services** — Developing person-centred standards and guidance, based on evidence and international best practice, for health and social care services in Ireland.
- **Regulating social care services** — The Chief Inspector within HIQA is responsible for registering and inspecting residential services for older people and people with a disability, and children's special care units.
- **Regulating health services** — Regulating medical exposure to ionising radiation.
- **Monitoring services** — Monitoring the safety and quality of health services and children's social services, and investigating as necessary serious concerns about the health and welfare of people who use these services.
- **Health technology assessment** — Evaluating the clinical and cost-effectiveness of health programmes, policies, medicines, medical equipment, diagnostic and surgical techniques, health promotion and protection activities, and providing advice to enable the best use of resources and the best outcomes for people who use our health service.
- **Health information** — Advising on the efficient and secure collection and sharing of health information, setting standards, evaluating information resources and publishing information on the delivery and performance of Ireland's health and social care services.
- **National Care Experience Programme** — Carrying out national service-user experience surveys across a range of health services, in conjunction with the Department of Health and the HSE.

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1 Purpose and Aim

The purpose of this protocol is to outline the process by which the Health Information and Quality Authority (HIQA) will conduct an international review of national rare disease strategies in selected countries. The information contained in the review will inform the development of a new national rare disease strategy in Ireland, through supporting the work of the Department of Health.

2 Process outline

It is important that a standardised approach to the process is developed and documented, to allow for transparency, aid project management and to mitigate risks.

Four distinct steps in the process have been identified and will be completed. These are listed below and described in more detail in sections 2.1-2.4:

1. Defining the scope
2. Search of relevant international sources
3. Review and extract relevant information on national rare disease strategies
4. Summarise the findings.

2.1 Defining the scope

A rare disease is defined as a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2,000 persons living in any World Health Organisation-defined region of the world.⁽¹⁾ Similarly, the European Union (EU) considers a disease to be rare if it affects fewer than 5 in 10,000 people in the European population.⁽²⁾ This EU definition was adopted in Ireland's first National Rare Disease Plan, which ran from 2014 to 2018.⁽³⁾

In 2010, the European Project for Rare Diseases National Plans Development (EUROPLAN) published recommendations as guidance for policy makers and other stakeholders involved in the development of national rare disease strategies.⁽⁴⁾ Within the current review, documents identified will be assessed broadly against the EUROPLAN definition of a national plan or strategy: "a set of integrated and comprehensive health and social policy actions for rare diseases (with a previous analysis of needs and resources), to be developed and implemented at national level, and characterised by identified objectives to be achieved within a specified timeframe".⁽⁴⁾

Further criteria outlined in section 2.3 will be then applied to identified documents to assess inclusion eligibility.

2.2 Search of relevant international sources

Information on national rare disease strategies from a select group of countries will be identified primarily from government resources (websites, reports and press releases), with representatives from key national-level organisations contacted for confirmation and additional resources, as appropriate. The following countries have been selected for inclusion in this review:

EU/EEA

- Austria
- Denmark
- Finland
- France
- Germany
- the Netherlands
- Portugal.

Non-EU

- Australia
- England
- Northern Ireland
- Scotland
- Wales.

These countries were selected on the basis of having published a national rare disease strategy after or at a similar time to or the publication of the *National Rare Disease Plan for Ireland 2014 – 2018*,⁽¹⁸⁾ combined with other factors such as geographical proximity to Ireland, similar population size, organisation of health services, European Union membership and or availability of documents in English.

Additionally, information from the *National Rare Disease Plan for Ireland 2014 – 2018* will be extracted to provide comparison to the selected countries.⁽³⁾ The associated resources that will be searched for each selected country are detailed in Appendix 1. This list is not exhaustive and will be expanded as necessary, should information on national rare disease strategies be available elsewhere.

The list of countries may be updated to include additional countries should this information be deemed relevant to the review. Similarly, some countries may be removed from the list if information on national rare disease strategies cannot be extracted due to a lack of information, or if a published national rare disease strategy cannot be identified. This will be documented.

2.3 Review and extract relevant information on national rare disease strategies

The review will extract relevant information on national rare disease strategies from official documents, reports and press releases, such as those published by government or state agencies and or bodies.

A national rare disease strategy will be included if it:

- relates to 'rare disease' as per the prevalence-based definitions adopted by Rare Diseases International (fewer than or equal to 1 in 2,000 people)⁽¹⁾ or the European Commission (fewer than 5 in 10,000 people)⁽²⁾ or similar
- meets the EUROPLAN definition of a national plan or strategy, such that it:
 - is a set of health and social policy actions for rare diseases
 - may include a previous analysis of needs and resources
 - contains actions that are integrated, that is, developed in a way to identify complementarities, maximise synergies and avoid duplications
 - is comprehensive, that is, the actions outlined in the plan aim to fulfil patients' main health and social care needs and are not limited to one single area, but rather several areas that are linked and support each other
 - is developed and to be implemented at national level, and
 - is characterised by identified objectives to be achieved within a specified timeframe⁽⁴⁾
- is published on or before 30 November 2023.

As all aspects of national rare disease strategies may not be contained within a single document, and make take various forms, relevant documents may include, but are not limited to:

- broad overview strategies (which may include themes, priorities and actions, for example)
- strategy implementation plans and or frameworks
- strategy evaluation plans and or frameworks
- legislation, rules, regulations and mandates
- policies.

Within these documents, relevant information may include, but is not limited to:

- timeline (to which a strategy applies)
- aims
- themes and or priorities (for example, prevention of rare diseases and screening for rare diseases)

- deliverables and or targets (for example, information around desired strategy outcomes, measurable targets and outcome measurement methods)
- implementation actions (for example, information about the types of actions to be carried out and with whom responsibility for implementation lies)
- governance and organisational structures
- funding model(s) (for example, government funding, EU funding and research-specific funding)
- references and or links to relevant initiatives such as:
 - screening programmes (including newborn screening)
- personalised medicine, genomics, genetic counselling, models of care and or care pathways (for example, how services are structured and delivered to population groups with certain conditions, pathways of transition from paediatric to adult care services, and or access to services abroad):
 - workforce (for example, workforce training, education and development)
 - European Reference Networks
 - participation in and or alignment with other EU initiatives
 - health information (for example, rare disease patient registries and data sharing with the European network of population-based registries for the epidemiological surveillance of congenital anomalies ((EUROCAT))
 - orphan medicines
 - rare disease research
 - alignment beyond the healthcare sector (for example, links to sectors such as education, employment and social protection)
- any additional information.

When identifying relevant documents the following are considered out of scope:

- rare disease strategies focused on global regions, such as the *Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases*⁽⁵⁾
- for countries which operate regionally, such as Australia, individual state or territory rare disease strategies, such as the *Western Australia Rare Diseases Strategic Framework 2015 – 2018*,⁽⁶⁾ are considered out of scope.

Furthermore, in relation to data extraction, information on the processes and methods used to develop national strategies will be considered out of scope within the current review. Information on public and patient involvement (PPI) will not be extracted as a distinct element. However, a number of the EUROPLAN recommendations relate to 'empowerment of patients' organisations',⁽⁴⁾ and

therefore it is anticipated that PPI may be integrated throughout many aspects of national strategies or plans. See Appendix 2 for the data extraction template.

Non-English documents and websites from the included countries will be translated where necessary via Google Translate or similar, with this noted as a potential caveat.

Any changes to the approach outlined in this document, will be noted in the report as a protocol deviation.

2.4 Summarise the findings

National rare disease strategies, where available for the included countries, will be documented and presented. Information extracted from within these strategies, as outlined in section 2.3, will be compared across the selected countries, and similar and contrasting elements will be presented descriptively. Any elements of the included strategies not addressed in the *National Rare Disease Plan for Ireland 2014 – 2018* will be identified.⁽³⁾

3 Quality assurance process

The review question will be undertaken in accordance with HIQA's HTA Directorate Quality Assurance Framework and led by an experienced member of the team. Data extraction for each country will be carried out by one reviewer and checked by a second reviewer for inaccuracies. The report will be reviewed by at least two members of the senior management team to ensure processes are followed and quality is maintained. To further ensure quality and accurate interpretation of the information included, an Expert Advisory Group (EAG) of relevant national and international experts in the field of rare disease will be created. Input from this group will be sought as appropriate, and a draft of the protocol and report will be circulated to them for review.

References

1. Rare Diseases International (RDI). Operational Description of Rare Diseases [Internet]. Rare Diseases International; 2023 [cited 2023 October 13]. Available from: <https://www.rarediseasesinternational.org/description-for-rd/>.
2. European Commission. Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases - Europe's challenges [COM(2008)679 final] [Internet]. Brussels: European Commission; 2008 [updated 2018 November 11; cited 2023 October 13]. Available from: <https://eur-lex.europa.eu/legal-content/en/ALL/?uri=CELEX:52008DC0679>.
3. Department of Health. National Rare Disease Plan for Ireland 2014 – 2018 [Internet]. Dublin: Department of Health; 2014 [cited 2023 October 12]. Available from: <https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf>.
4. EUROPLAN. Recommendations for the development of National Plans on Rare Diseases: Guidance Document [Internet]. Brussels: European Project for Rare Disease National Plans Development; 2010 [cited 2023 October 16]. Available from: http://www.europlanproject.eu/_europlanproject/Resources/docs/2008-2011_2.EuroplanRecommendations.pdf.
5. Asia-Pacific Economic Cooperation. Action Plan on Rare Diseases [Internet]. Papua New Guinea: Asia-Pacific Economic Cooperation; 2018 [cited 2023 October 16]. Available from: https://www.apec.org/docs/default-source/satellite/Rare-Diseases/APEC_ActionPlan.pdf.
6. Government of Western Australia Department of Health. WA Rare Diseases Strategic Framework 2015–2018 [Internet]. Perth: Department of Health, Western Australia; 2015 [cited 2023 October 13]. Available from: <https://www.health.wa.gov.au/~media/Files/Corporate/Reports-and-publications/PDF/Rare-diseases-strategic-framework.pdf>.

Appendix 1: List of countries and associated resources

The resources listed below will be searched for relevant data relating to national rare disease strategies (this list is not exhaustive and will be added to as necessary):

EU/EEA

- Austria
 - [Social Affairs, Health, Care and Consumer Protection](#)
 - [Pro Rare Austria](#)
- Denmark
 - [Ministry of Health](#)
 - [Danish Health Authority \(Sundhedsstyrelsen\)](#)
 - [Rare diseases Denmark](#)
- Finland
 - [Ministry of Social Affairs and Health](#)
 - [Finnish Institute for Health and Welfare](#)
 - [HARSO-Rare Disease Alliance Finland](#)
- France
 - [The Ministry of Health and Prevention](#)
 - [Public Health France](#)
 - [French Rare Diseases Alliance](#)
- Germany
 - [Federal Ministry of Health](#)
 - [German National Alliance for Chronic Rare Diseases](#)
- Ireland
 - [Irish Government](#)
- Netherlands
 - [Ministry of Health, Welfare and Sport](#)
 - [National Patient Alliance for Rare and Genetic Diseases](#)
- Portugal
 - [Directorate General of Health \(DGS\)](#)
 - [Doenças Raras Portugal](#)

Non-EU

- Australia
 - [Department of Health and Aged Care](#)
 - [Rare Voices Australia](#)
- England
 - [Department of Health and Social Care](#)
 - [Office for Health Improvement and Disparities](#)
 - [UK Health Security Agency](#)

- o [Rare Disease UK](#)
- o [Genetic Alliance UK](#)
- o [SWAN UK](#)
- Northern Ireland
 - o [Department of Health](#)
 - o [Health and Social Care Northern Ireland](#)
 - o [Northern Ireland Rare Disease Partnership](#)
- Scotland
 - o [Scottish Government](#)
 - o [Public Health Scotland](#)
- Wales
 - o [Welsh Government](#)
 - o [Public Health Wales](#)

Appendix 2: Sample data extraction template

Country (Reference)	Strategy information
Author(s) Title	
Timeline	
Overall aim(s)	
Themes and or priorities	
Targets (if specified) and measurement method(s) (where available)	
Implementation action(s), lead(s) and key performance indicator(s)	
Governance and organisational structures	
Funding model	
<i>References and or links to relevant initiatives</i>	<i>(If already extracted above, redirect to appropriate section(s). If no information is included in the strategy, list as 'not mentioned'.)</i>
Screening programmes (including newborn screening)	
Personalised medicine, genomics, genetic counselling	
Models of care/care pathways	
Workforce	
European Reference Networks	
EU alignment and participation	
Health information (including rare disease registries)	
Orphan medicines	
Rare disease research	
Alignment beyond the healthcare sector	
Any additional information (for example, background to the strategy or strategy development)	

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