BRIEFING PAPER

Exeter Policy Research Programme Evidence Review Facility: October 2025

From Evidence to Practice: A Scoping Review and Framework for Understanding Health Inequities in Rare Disease Diagnosis and Service Access.

RARE diseases are diseases which affect fewer than one in 2000 people. Although rare diseases are individually rare, they are collectively common, affecting around one in 17 people in the UK.[1] Evidence from studies conducted in the UK suggests that people living with a rare disease face many challenges with accessing health and social care services, as well as long delays for diagnosis.[2] These difficulties are exacerbated for some people within the rare disease community, including ethnic minorities, women, and according to socioeconomic status and where you live.[2]

As a result of challenges with accessing services, people with a rare disease may have worse health outcomes than people in the general population, contributing to health inequity. The UK Rare Diseases Framework and England's 2023 Rare Disease Action Plans are committed to addressing health inequities associated with rare diseases. [3-4]

"Although rare diseases are individually rare they are collectively common, affecting around one-in-17 people in the UK….Acting now to tackle health inequalities in rare diseases will strengthen the NHS and improve outcomes for all." NHS Confed Expo

We were commissioned via the NIHR Policy Research Programme to undertake research on the extent and nature of health inequity experienced by people with a rare disease with respect to diagnosis and service access. We approached this request in two parts:

- First, we undertook a scoping review which sought to identify evidence of health inequities experienced in the rare disease community with respect to diagnosis and service access;
- Secondly, we used the identified evidence to develop a framework which maps the experiences of the rare disease community onto the patient care pathway across diagnosis and service access, including both health and social care.

The second part of this work facilitates the identification of contexts in which specific types of inequity are experienced, which we envisage can be used to inform where targeted interventions can be developed to address these inequities.

What are health inequities?

Health inequities are differences in health opportunities and outcomes which are avoidable, systemic and unfair.[5] We used the PROGRESS+ tool in our research to assist in the identification of health inequities.[6] PROGRESS+ is an acronym used to identify characteristics that stratify health opportunities and outcomes. This includes: **Place of residence, race/ethnicity, occupation, religions, education, socioeconomic status, social capital, Plus (e.g. age, disability).** In addition we spoke to stakeholders and people with a rare disease to understand the types of experiences we should be looking out for in the research.

QR code for framework

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Exeter PRP Evidence Review Facility, University of Exeter
https://medicine.exeter.ac.uk/esmi/workstreams/exeterprpevidencereviewfacility/

Stage 1: Scoping review of the evidence-base

Why did we undertake this review?

Although there are well documented examples of health inequity in the rare disease community, there is limited overall understanding of the extent of the evidence. Understanding this is particularly important given the Rare Diseases Action Plan commitment to gathering the evidence to evaluate whether rare diseases should be incorporated into the CORE20PLUS5 framework. The CORE20PLUS5 aims to support integrated care systems to reduce health inequities for people with complex and long-term conditions at a local and national level.[7] **Our scoping review systematically identifies and describes the key characteristics and extent of evidence on health inequities shared across the rare disease community and experiences of inequities which relate to subgroups within the rare disease community, with regards to receipt of a diagno-**

How did we do this review?

Finding the literature: We searched for relevant studies using a combination of bibliographic databases, checking reference lists, forward citation searches, inspecting relevant journal contents pages, searching relevant websites, inspecting the included studies of relevant systematic reviews, and expert solicitation.

Eligibility criteria:

Study participants: people living with a rare disease and carers of people living with a rare disease.

Phenomenon of interest: experiences of inequity relating to receipt of a diagnosis or access to services, including experiences which occur across the rare disease community as a whole, and, experiences relating to characteristics which stratify health opportunities and outcomes as set out in the PROGRESS+ framework.[6]

Study type: UK primary research, and systematic reviews meeting the DARE criteria[8] and reporting data from at least one World Bank high-income country,[8] only studies/systematic reviews published in English and in 2010 or later.

Study selection and data analysis: Studies were screened independently by two reviewers using priority screening in EPPI-Reviewer software. Data-extraction was carried out by one person and checked. The data was then tabulated and narratively summarised.

What did we find?

Findings are divided into two parts summarizing experiences of inequity: (1) shared across the rare disease community and (2) subgroups within the rare disease community, with reference to PROGRESS+.

1. Types of inequity shared across the rare diseases community

Delayed diagnosis: Delays to a rare disease diagnosis may lead to inequity between people with a rare disease and the general population, who do not routinely experience similar delays to diagnosis.

Lack of knowledge: Lack of knowledge about rare diseases amongst health/social care professionals hinders diagnosis and accessing services, and can include dismissive attitudes towards symptoms of rare diseases.

Lack of information: Health and social care professionals did not always provide or signpost people with a rare disease to relevant or sufficient information about their condition.

Limited service provision: Challenges with accessing appropriate services, often related to limited services provision and specific challenges within different types of services: mental health services, emergency services, dentistry services, specialist services, social care services and services in general where these were not specified.

Limited services for undiagnosed conditions: Specific challenges for people with symptoms of a rare disease without a diagnosis (i.e. syndrome without a name) when accessing services.

Lack of care coordination: Experiencing a lack of care co-ordination which adversely affects the receipt of a diagnosis and accessing services after diagnosis.

2. Types of inequity relating to subgroups within the rare disease community (PROGRESS+ [6])

Place of residence: Challenges in accessing diagnostic tests and care for rare diseases where this requires travelling long distances to specialist tertiary services, particularly for those living in rural areas.

Race/ethnicity: Ethnic minority patients can experience dismissive attitudes towards their symptoms when receiving a diagnosis or accessing services. This can cause delays to diagnosis and treatment.

Gender: Women perceived dismissive attitudes towards their symptoms or symptoms of their children. They also felt there was a lack of support for sexual and reproductive health.

Socioeconomic status: People with a rare disease need to pay for private services to avoid delays to diagnosis and care, or to receive better care standards. They might find it challenging to pay for specialist equipment.

Age: Dismissive attitudes towards children's symptoms and pain delay's their diagnosis. Older age groups also experience delays to diagnosis. Transition to adult services is unsatisfactory, with children's services rated better than adult services.

Disability: Symptoms of a rare disease are sometimes erroneously attributed to pre-existing disability. Clinicians lack of understanding of people with a rare intellectual disability impacts their access to services.

Stage 2: Explanatory framework

Why did we develop this framework?

To bridge the gap between the findings of the scoping review and the understanding required to inform practical action, we undertook further work to develop a framework of the structural interrelation of inequities experienced by people with a rare disease across the patient care pathway. This could be used to inform the development of targeted interventions aimed at addressing these inequities, further supporting the commitment of the England Rare Diseases Action Plan 2023.[3]

How did we develop the framework?

- We undertook further analysis of the data from the scoping review, with reference to Diderichsen and Hallqvist's framework for describing pathways from social contexts to health outcomes. [8]
- We adapted the overarching societal context in Diderichsen and Hallqvist's framework to focus on *health policy, resourcing and organisation* and removed Diderichsen and Hallqvist's references to *specific exposure or disease/injury* at the individual/group level to focus on *social position and consequences of ill-health.*
- The framework aims to support the identification of *contexts* along the patient care pathway that are associated with *mechanisms (* types of inequity identified in the review e.g. lack of knowledge amongst health clinicians) leading to differential *outcomes (*e.g. delayed diagnosis*).*

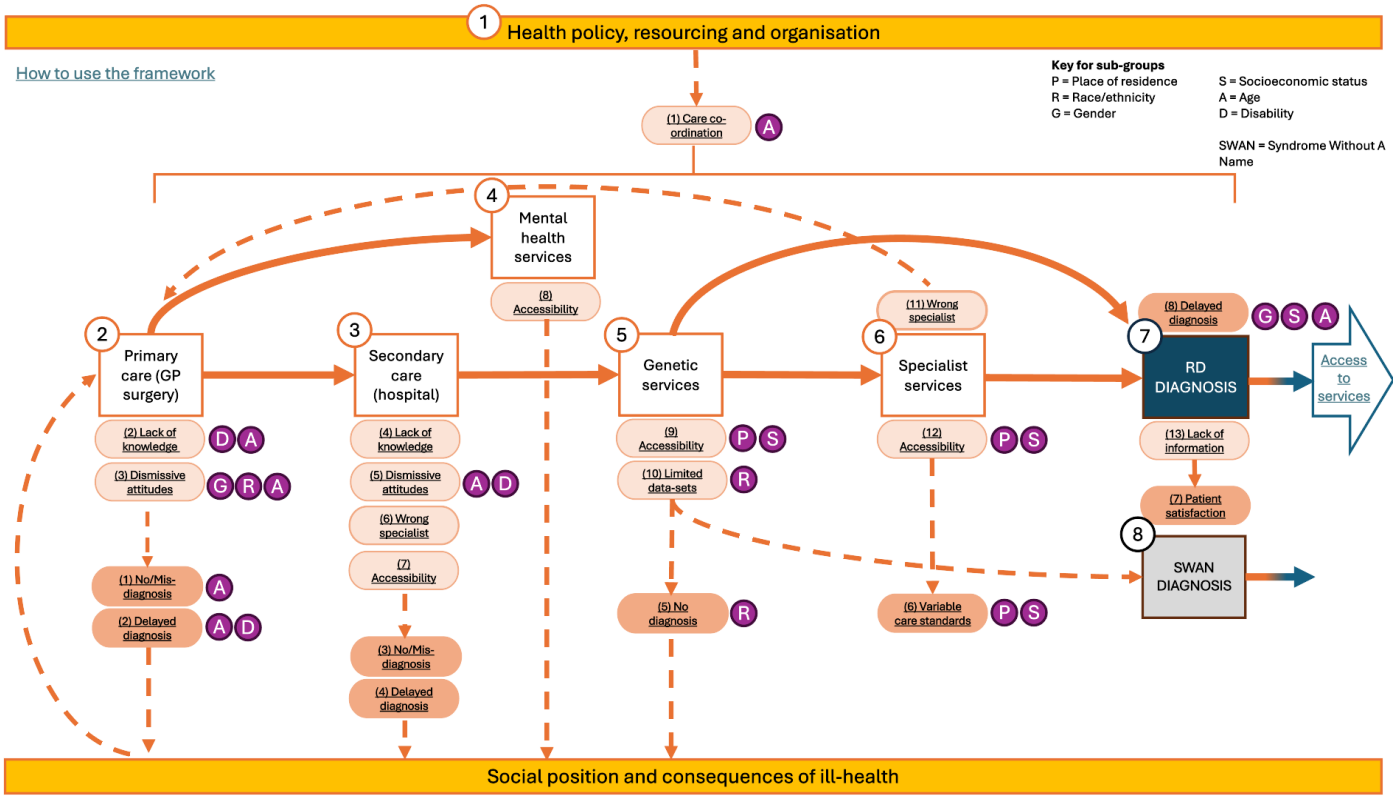

Fig 1. Framework mapping inequities onto patient pathway, and specifically to receipt of a diagnosis. Shows contexts, mechanisms and outcomes across the pathway.

Presentation of framework

- The framework is presented in a slide deck in two stages: (1) receipt of a diagnosis (shown in Fig 1 above) and (2) access to services.
- Clear boxes represent service context and are labelled with the name of each service.
- Mechanisms which lead to inequity within each context are presented in light shaded boxes.
- Outcomes arising from the mechanisms are presented in darker shaded boxes.
- Each of the contexts, mechanisms and outcomes is assigned a number for ease of reference. (The numbers do not necessarily indicate the order that someone moves through the pathway).
- Purple circular labels show which sub-groups are identified in the data as experiencing a particular type of mechanism or outcome. These labels correspond with the PROGRESS+ framework.[6]

What are the implications of this research?

Our findings draw attention to experiences which are shared across the rare disease community and are different to experiences in the general population, and also to differences in experiences within the rare disease community itself, with respect to receipt of a diagnosis and access to services. These experiences are indicative of health inequity.

The **review** has highlighted the following:

- More research into health inequity for people with less common rare diseases, including ultra-rare diseases, is needed.
- More comparative data would be helpful to establish the extent of inequities between the rare disease community and general population, and between subgroups within the rare disease community.
- There is a need for more research on inequities relating to accessing social care services and end-of-life care, which were relatively underrepresented in the data
- There are relatively few studies on the prevalence of ethnic minority data in genetic data sets, which we have been alerted to by topic experts as a major concern for equity relating to receipt of a diagnosis.

The **framework** has highlighted the following:

- Stages where the identified health inequities might occur across the patient pathway and how they relate to each other.
- Potential policy entry points across contexts within the patient pathway, where interventions aimed at addressing health inequities can be implemented.

Overall, the evidence gathered suggests that rare diseases should continue to be considered for inclusion in the Core20PLUS5 framework to increase action to reduce health inequalities.

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Link to full report:
Scoping review: https://zenodo.org/records/14892726

Framework: https://zenodo.org/records/16793150

Exeter PRP Evidence Review Facility

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