ISRCTN41639707 https://doi.org/10.1186/ISRCTN41639707

An online consensus-building study to agree on key policy measures for assessing whether the England Rare Diseases Action Plans (developed by the Department of Health and Social Care) are effective

Submission date	**Recruitment status**	[X] Prospectively registered
16/03/2025	No longer recruiting	[_] Protocol
Registration date	**Overall study status**	[_] Statistical analysis plan
21/03/2025	Ongoing	[_] Results
Last Edited	**Condition category**	[_] Individual participant data
21/03/2025	Other	[X] Record updated in last year

Plain English summary of protocol

Background and study aims

In the UK, a rare disease is defined as one affecting fewer than one in 2,000 people, with over 7,000 rare diseases affecting over 3.5 million people. These diseases often have limited research, delayed diagnoses, and fragmented care. To improve care, the Department of Health and Social Care (DHSC) introduced the UK Rare Diseases Strategy in 2013, later updated in 2021 with the UK Rare Diseases Framework. The Framework has four priorities: faster diagnosis, increased awareness among health professionals, better coordination of care, and improved access to treatments and drugs.

The England Rare Diseases Action Plans implement the Framework. To assess their impact, clear measures are needed to see how well policies are working and help identify areas for improvement. This is where the eDelphi study comes in. A two-round online Delphi study will be conducted to identify and agree on key measures for evaluating the effectiveness of the Action Plans. The eDelphi method was chosen because it allows participants to remain anonymous, encouraging open and honest opinions.

The study will gather expert opinions to identify and prioritise measures for evaluating the success of the plans (including their validity, reliability, clarity, and how feasible it is to collect the necessary data). The study is part of a larger research programme, RareCare, which aims to design effective measures for evaluating the England Rare Diseases Action Plans and improving time to diagnosis for people living with rare disease. The study's findings will help policymakers track progress and improve outcomes for people with rare diseases.

Who can participate?

This study aims to recruit 50 individuals living with rare diseases or caring for someone with one and 50 people working in the rare disease field (paid or unpaid). By including people with lived experience, the study helps to ensure the measures chosen for evaluating rare disease policies reflect the real-world impact.

For participants with lived experience of a rare disease, the study will work with patient advocacy groups and aim to gather a diverse sample. The sample will comprise people with various rare diseases, varying ages, different stages of the patient journey, genetic and non-genetic conditions, while also including underrepresented groups. This diversity will help guarantee that the chosen measures are relevant to the broader rare disease community.
People working or volunteering in the rare disease field will be recruited based on their expertise and knowledge in healthcare policy, the rare disease sector, or relevant aspects of the healthcare system. They will include representatives from NHS management (national and regional commissioning), healthcare professionals, industry, education, patient advocacy groups, the broader health and care system, and the data and research sectors. A careful selection process will create diversity in geographic locations and expertise.
Eligible participants must have rare disease experience based in England, be 16 or older, have no financial conflicts of interest, be able to read and write in English, and have internet access.

What does the study involve?
The study will use a two-round eDelphi method to review and agree on the best ways to measure how well the England Rare Diseases Action Plans are working. The researchers carefully chose which potential measures would be included for rating in the Round 1 questionnaire. First, five experts checked and rated different measures compiled by government to monitor rare disease care. They looked at how clear these were, whether they could show real change, and how they might affect health inequalities. At the same time, researchers searched scientific studies and databases to find more potential measures that fit rare disease priorities. After reviewing all the information, potential measures were shortlisted for inclusion.
In the first eDelphi round, these measures will be grouped into the four Priority Areas listed above. Participants will be asked to rate shortlisted possible measures on a scale of one to nine, indicating their perceived value, and can also suggest additional measures. They will also be asked for personal information, such as demographics (age, gender, ethnicity, etc.), and experience with rare diseases (time since diagnosis, main field of work, etc). This information will help in describing the diversity and experiences of the study participants.
The answers from the first round will be analysed by determining whether the participants agree with each other. In the second round, participants will review the Round 1 results, compare their responses with the group consensus, and adjust their ratings if needed. After two rounds, a final set of policy measures will be selected. Any discrepancies in Round 2 ratings will be explored through follow-up research.

What are the possible benefits and risks of participating?
By taking part, participants could help to improve healthcare for people with rare diseases and support better government monitoring. There are no significant risks associated with taking part, but thinking about personal experiences with rare diseases may bring up difficult emotions for some people. There will be information and advice on how to contact support agencies at the end of both questionnaires.

Where is the study run from?
Coventry University leads on the study design, delivery, and data analysis. The study is being run in partnership with Consilium Scientific and Realise Advocacy Ltd.

When is the study starting and how long is it expected to run for?
The two rounds of the eDelphi study are expected to run from March to June 2025 (approximately). Once a participant provides their consent, they can access the questionnaire. Each round will be open for at least 2 weeks, and if response rates are low, we will extend this by a week. Up to three reminders will be sent to participants who have not completed the questionnaire.

The data analysis for Round 1 and preparation of feedback and questions for the next round (including possible rephrasing or refining of the candidate measures and adding those newly suggested) is estimated to take 3 weeks. Once completed, the second-round questionnaires will be made available to those participants who consented to participate in the second round.

Who is funding the study?
The National Institute for Health and Care Research (NIHR) funds this independent research [RareCare, NIHR205983]. The views expressed in this publication are those of the research team and not necessarily those of NIHR or The Department of Health and Social Care.

Who is the main contact?
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Additional identifiers

Clinical Trials Information System (CTIS)
Nil known

ClinicalTrials.gov (NCT)
Nil known

Protocol serial number
18424-01

Study information

Scientific Title
eDelphi study to identify consensus on policy metrics which should be included in future assessments of the England Rare Diseases Action Plans

Acronym
RareCare_eDelphi

Study objectives
Aims:
To develop a national consensus on the most relevant and appropriate high-level metrics that policymakers can use to effectively monitor the implementation progress of the England Rare Diseases Action Plans. This study also seeks to explore variations in consensus and priority levels across different policy domains and stakeholder groups while ensuring engagement with underserved and underrepresented communities.

Objectives:
1. To achieve stakeholder agreement on the most relevant and appropriate high-level policy metrics for assessing the England Rare Diseases Action Plans using the eDelphi method.
2. To analyse differences in consensus and priorities across policy domains and stakeholder groups.
3. To ensure the perspectives of underserved and underrepresented communities are meaningfully incorporated in the consensus-building process.
4. To provide policymakers with validated high-level metrics for effectively tracking progress in the England Rare Diseases Action Plans.

Ethics approval required
Ethics approval required

Ethics approval(s)
approved 19/03/2025, Coventry University Group Research Ethics Committee (Priory Street, Coventry, CV1 5FB, United Kingdom; +44 (0)24 7765 7688; ethics.uni@coventry.ac.uk), ref: P183377

Study design
eDelphi study

Primary study design
Observational

Study type(s)
Other

Health condition(s) or problem(s) studied
Any rare disease

Interventions
A long list of candidate metrics for the 36 Actions linked to the four Priority Areas of the UK Rare Diseases Framework (i.e., faster diagnosis; increased healthcare professional awareness, better care coordination and; improved access to specialist care, treatment, and drugs) was compiled from government logic models. The list was discussed in three workshops, and metrics irrelevant to the objectives, unclear in their measurement, or not suitable for tracking progress over time were removed. Additional metrics were identified through an evidence review and expert opinion.

In the first round of a two-round eDelphi study, shortlisted metrics are grouped into the four Priority Areas. An eDelphi panel of 50 people living with rare disease and their carers and 50 professional stakeholders (i.e., researchers, healthcare professionals, policymakers, commissioners, delivery partners, and industry representatives) will rate each metric on a 1-9 scale. The sample size allows for 20% attrition. Consensus for inclusion or exclusion will be based on 70% agreement (7-9 for 'extremely valuable', 1-3 for 'not valuable'), with fewer than 15% rating the metric at the opposite end of the scale.

Metrics will be included or excluded if consensus is reached in both groups; otherwise, they will remain under consideration in Round 2.

In Round 1, the eDelphi participants may propose additional metrics, which will be included in round 2. After two rounds, a final set of policy metrics will be selected. Any discrepancies in Round 2 ratings will be explored through follow-up research.

Intervention Type
Other

Primary outcome(s)
The number and list of candidate metrics achieving consensus for inclusion in the future monitoring of the England Rare Diseases Action Plans, assessed at the end of Round 2. Consensus is pre-defined as at least 70% of participants in both groups (people living with rare disease and professionals) rating a metric as extremely valuable (score 7, 8, or 9 on a 1–9 Likert scale), with fewer than 15% rating it as not valuable (score 1, 2, or 3) in the questionnaires.

Key secondary outcome(s))
1. The extent to which consensus levels differ between people living with rare diseases (or their carers) and professional stakeholders, assessed at the end of Round 2. This will be measured by comparing the percentage of participants in each group who rate individual metrics as extremely

valuable (scores 7, 8, or 9 on a 1–9 Likert scale) in the questionnaires.
2. The extent of variation in consensus on candidate metrics based on the demographic characteristics of Delphi panel members, assessed at the end of Round 2. This will be measured by analysing the percentage of participants in different demographic subgroups who rate individual metrics as extremely valuable (scores 7, 8, or 9 on a 1–9 Likert scale) in the questionnaires.

Completion date
26/04/2026

Eligibility

Key inclusion criteria
1. Have experience with rare diseases based in England
2. Be 16 years or older
3. Be able to read and write in English
4. Have access to the internet and a device to complete the questionnaire

Participant type(s)
Patient, Health professional, Carer, Employee, Service user, Other

Healthy volunteers allowed
No

Age group
Mixed

Lower age limit
16 years

Sex
All

Key exclusion criteria
1. Have self-declared financial conflicts of interest in assessing the impact of the England Rare Diseases Action Plans
2. Be a member of the RareCare project team, Research Advisory Group, or Patient Public Involvement and Engagement Group

Date of first enrolment
25/03/2025

Date of final enrolment
20/04/2025

Locations

Countries of recruitment
United Kingdom

England

Study participating centre
Consilium Scientific
Mabledon Place
Hamilton House
London
United Kingdom
WC1H 9BB

Study participating centre
Alstrom Syndrome UK
4 St Kitts Close
Torquay
United Kingdom
TQ2 7GD

Study participating centre
Beacon: for rare diseases
66 Devonshire Road
Cambridge
United Kingdom
CB1 2BL

Study participating centre
Cambridge Rare Disease Network
BCS
5 Station Court
Station Road
Great Shelford
Cambridge
United Kingdom
CB22 5NE

Study participating centre
Genetic Alliance UK Limited
The Clock Tower
5 Farleigh Court
Old Weston Road
Flax Bourton
Bristol

United Kingdom
BS48 1UR

Study participating centre
Metabolic Support UK
Centurion House
Deansgate
Manchester
United Kingdom
M3 3WR

Study participating centre
NRG Collective Limited [RARE Revolution]
Suite 2006
Letraset Buildings
Wotton Road
Ashford, Kent
United Kingdom
TN23 6LN

Sponsor information

Organisation
Coventry University

ROR
https://ror.org/01tgmhj36

Funder(s)

Funder type
Government

Funder Name
National Institute for Health and Care Research

Alternative Name(s)
National Institute for Health Research, NIHR Research, NIHRresearch, NIHR - National Institute for Health Research, NIHR (The National Institute for Health and Care Research), NIHR

Funding Body Type
Government organisation

Funding Body Subtype
National government

Location
United Kingdom

Results and Publications

Individual participant data (IPD) sharing plan
The datasets generated during and/or analysed during the current study will be stored in a publicly available repository. Data will include anonymised quantitative responses, and the final list of agreed-upon metrics. Only anonymised and non-identifiable summary data will be shared publicly. For that reason, it is likely that the data will be in aggregated format with at least five other individuals in each group. Any data containing personally identifiable information will be excluded from public repositories and retained securely within Coventry University, accessible only by authorised personnel.

The preserved data will be stored in an open-access repository that meets the relevant standards for data storage, such as the UK Data Service or a similar recognised repository. The data will be assigned a DOI (Digital Object Identifier) to facilitate citation and long-term access. The repository will allow for secure long-term preservation and ensure that the data remains publicly available in a format that complies with FAIR (Findable, Accessible, Interoperable, and Reusable) principles. Consent will be obtained by participants prior to data collection for anonymised, summary data to be held in a long-term repository for future access/analysis. Permission will be sought from the NIHR prior to publication, and any embargoes or restrictions on data sharing will be discussed with relevant stakeholders before submission.

IPD sharing plan summary
Stored in publicly available repository

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Study website	Study website	11/11/2025	11/11/2025	No	Yes