Department of Health & Social Care

Introduction

Thank you for taking the time to complete this questionnaire.

The aim of the survey is to gather feedback from the community on the draft actions that have been developed for inclusion in England's 2022 action plan for the UK Rare Diseases Framework.

The questionnaire is aimed at members of the rare diseases community (e.g. people living with a rare disease / patient or public voice representatives / carers / family members / clinicians / industry professionals / healthcare professionals / researchers) or representatives from organisations involved in rare disease work. Because the questionnaire focuses on England's action plan, we would ask that you only complete it if you (or your organisation) are based in England, and/or if your organisation is involved in work across the UK.

As we are particularly seeking input from members of the rare disease community with an interest in rare disease policy, existing knowledge of the rare disease landscape is desirable (although not essential) for answering this questionnaire.

It should take about 30-45 minutes to complete.

* You cannot save your answers and return to the questionnaire, so if you prefer you can download the questionnaire as a Word document, fill it in at your own pace, and then copy your answers into the online questionnaire to submit.

Privacy notice

All responses to the questionnaire are anonymous.

In addition to your opinions, we will also ask you for some personal information such as whether you are a person living with a rare disease or a healthcare professional, your age, ethnicity and gender.

We will feed back the results of this questionnaire to delivery partners and will ask them to evidence how they will be addressing the points raised from the analysis of the questionnaire.

By filling out this questionnaire, you agree to your data being used in this way. If you wish to withdraw from the questionnaire, you can do so at any time and your answers will not be saved.

For further information, please see our privacy policy here.

Background

The UK Rare Diseases Framework, published in January 2021, replaced the 2013 UK Strategy for Rare Diseases. The Framework outlines the Government's priorities for rare diseases over the next 5 years: helping patients get a final diagnosis faster, increasing awareness of rare diseases among healthcare professionals, better coordination of care, and

improving access to specialist care, treatment and drugs. The Framework will be implemented through nation specific ‘Action Plans’, which will detail the commitments that each nation will put in place to meet the shared priorities of the Framework.

The England Rare Diseases Framework Delivery Group is responsible for developing, agreeing and monitoring the action plan for England. Its membership comprises representatives from delivery partner organisations responsible for implementing the Framework for England. These delivery partners have put forward a series of draft actions that aim to collectively meet the priorities of the UK Rare Diseases Framework. A summary of the draft actions is available here for reference while you complete this questionnaire.

The 2022 Rare Diseases Action Plan for England will be published early next year, describing actions which will begin during that year. It will clearly indicate who is responsible for each action, and provide details of milestones and timeframes. The action plan will be the first step towards the implementation of the Framework and will be followed by a series of annual updates with new actions that will build upon progress made and take into account any new opportunities.

Part 1

This part of the survey asks you to consider each individual action proposed for the 2022 Rare Diseases Action Plan for England. There are 23 actions in total. The questions are not compulsory and you are welcome to concentrate your answers on the actions that are most relevant to you.

1

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Undiagnosed conditions Working with doctors toctest new approaches for patients with undiagnosed rare conditions. Examples could include a one-stop paediatric clinic or a more targeted adult neurology clinic. **Priorities: 1** (Helping patients get a diagnosis faster)	NHS England	NHSE will test the approaches with patient groups before piloting.	Development of a pilot approach(es) by April 2022 Selection of sites by summer 2022 Evaluation of sites to follow	To be developed

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes the commitment to test new approaches for patients with undiagnosed rare conditions. We would welcome more information about how patients will be selected and whether this is a one-off activity or the start of a cycle of continual improvement.

2

Action	Delivery partner	Patient involvement	Milestones	Impact measures
<u>Developing a genomic testing strategy</u> Developing the testing strategy for the NHS Genomic Medicine Service (GMS). This involves overseeing the rollout of a range of DNA sequencing tests that will play an important role in diagnosis of rare diseases. **Priorities: 1** (Helping patients to get a faster diagnosis); **2** (Improving	NHS England	Patients are full members of all the groups which oversee the GMS at a national level, and are involved in all seven NHS GMS Alliances (which work to integrate genomics and personalised medicine into clinical care across different geographical regions). This ensures NHS England meets its legal duties to involve the public in decisions through consultation and engagement. NHS England are also working with	**2021/22** • Phase 2 and phase 3 clinical indications for whole genome sequencing will be launched. • By December 2021 the Test Directory will be updated to include new applications, including for rare disease for 2021/22. A key milestone will be when all seven NHS	Patient Level Contract Monitoring (PLCM) data will show that the genomic testing strategy is being delivered across England, with increased activity as the range of conditions on the test directory is expanded, and developments in technology are introduced in the NHS GMS.

access to specialist care, treatments and drugs)	partners to hear seldom heard voices (e.g. minority groups, children and young people).	Genomics Laboratory Hubs (GLHs) offer the additional indications included in the updated National Genomic Test Directory.	

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

Gene People welcomes the development of a testing strategy for the NHS Genomic Medicine Service. We would ask that legal duties are seen as minimum requirements, not aspiration. We suggest an impact measure on the number of engagements with people with seldom heard voices.

3

Action	**Delivery partner**	**Patient involvement**	**Milestones**	**Impact measures**

Developing the NHS GMS infrastructure Supporting the implementation of the UK Rare Diseases Framework, Genome UK and the NHS Long Term Plan commitments for genomics, by continuing to develop the NHS GMS infrastructure. **Priorities: 1** (Helping patients to get a faster diagnosis)**; 2** (better coordination of care), **4** (Improving access to specialist care, treatments and drugs)	NHS England	Patients are full members of all the groups which oversee the GMS at a national level, and are involved in all seven NHS GMS Alliances (which work to integrate genomics and personalised medicine into clinical care across different geographical regions). This ensures NHS England meets its legal duties to involve the public in decisions through consultation and engagement. NHS England are also working with partners to hear seldom heard voices (eg. minority groups, children and young people).	An annual process for updating the Test Directory will be published. NHS GMS Alliances will have delivered a range of national and local transformation projects, in line with priorities identified in the NHS GMS Alliance business planning guidance. An updated Genomics Clinical Service Specification will be published.	An annual process for updating the Test Directory will have been implemented. NHS GLH and NHS GMS Alliance planning guidance will continue to be annually published to inform business planning. Annual business and financial planning for the NHS GMS Alliances will be implemented.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )

This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

We are concerned that the Impact Measures are in fact outputs and do not show how they contribute to the priorities. We welcome an annual update of the Test Directory; the rest of the actions are standard business procedure. A deadline for the GCSS publication is needed.

4

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Toolkit for virtual consultations Action that proposes the development of a toolkit to improve the use of videoconference and telephone clinic calls in services for patients with rare diseases. **Priorities: 2** (better coordination of care)	NHS England	Patients, family members and carers and supporting charities have been involved in all stages of the development of the toolkit.	Toolkit finalised by the end of December 2021 with a publication date of spring 2022.	The Highly Specialised Commissioning Team will monitor the uptake of the toolkit within the Highly Specialised Portfolio through annual clinical meetings. The toolkit can be further expanded into tertiary units which refer into services in the Highly Specialised Portfolio, and across services in NHS England specialised commissioning.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

There has been a mixed response from the rare condition community to remote clinics during the pandemic. A blended approach is needed so those who do not welcome remote clinics or with no access to digital are not disadvantaged. An evaluation should be in the implementation plan.

5

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Gene therapies Developing a strategy for gene therapies and other advanced therapy medicinal products (ATMPs). Based on horizon scanning by NHS England. **Priorities: 4** (Improving access to specialist care, treatments and drugs).	NHS England	TBC	Development of a strategy by summer 2022	TBC

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	(Y)	( )	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes this commitment, especially the link to horizon scanning. We would welcome clarity on how patients will be involved in the formation of the strategy and ideally in evaluating the impact of that strategy.

6

Action	Delivery partner	Patient involvement	Milestones	Impact measures
<u>Support for early access to drugs</u>				High percentage of drugs available at anticipated date of delivery
Action supporting access to drugs for patients with rare diseases through a number of processes: • Mapping the available access initiatives and understanding their	NHS England	TBC, noting that there is patient/patient group engagement in the evaluation of all drugs, whether by NICE or by NHS England	Develop and finalise mapping – spring 2022	Access initiatives mapped Commissioning processes to support rapid access to drugs developed

place in the pathway for evaluation of drugs. • Linking to the development of the Innovative Medicines Fund. • Identifying drugs and challenges with delivery, through horizon scanning. • Listing drugs that have been identified for access through the access initiatives. **Priorities: 4** (Improving access to specialist care, treatment and drugs).		Ongoing list of relevant drugs developed and maintained Measurement taken of whether or not there is access at the point of anticipated delivery

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

While patients are involved in individual drug evaluations, Gene People is keen that patients are involved in the actions listed to ensure their experience is embedded. Clarity on how these initiatives will link with the IMF would be welcome.

7

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Monitoring uptake of drugs Developing systems to measure (a) overall uptake of drugs for patients with rare diseases and (b) geographical mapping of access to drugs for patients with rare diseases. **Priorities: 4** (Improving access to specialist care, treatment and drugs)	NHS England	Patients, family members and carers will not specifically be involved in the development and implementation of this action, but they may be involved in taking forward actions as a result of the outcomes of the exercise.	Development of the processes. Running the two exercises. Writing a report on the outcomes of the exercises. Developing an action plan in response to the outcomes of the exercises.	For most services this exercise will be run on an annual basis as appropriate. It should then be possible to see improvements in (a) expected uptake and (b) improved geographical access.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	(Y)	( )	( )	( )	( )
This action is sufficiently ambitious.	( )	(Y)	( )	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes this commitment. It is crucial that treatments for rare conditions are available to all for whom they would be beneficial including those from underrepresented groups. We hope that the progress reports will be shared with the public.

8

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Medicines Repurposing Programme The Medicines Repurposing Programme identifies and develops opportunities to repurpose medicines, meaning medicines used in ways not included in the original licence. The remit of the programme includes, but is not restricted to, rare conditions and diseases.	NHS England	The Medicines Repurposing Programme Steering Group includes two patient and public voice representatives plus a representative of the Association of Medical Research Charities. When a medicine enters the programme we will set up a Working Group including two patient and public voice representatives. Patient and research organisations can propose	A plan for medium- and long-term deliverables is in development. In the short term: - spring 2022: publish key documents including how to propose a medicine to the programme. - summer 2022: start licensing work on first pilot medicine.	A plan for impact evaluation is in development.

Priorities: 4 (Improving access to specialist care, treatment and drugs)	medicines to the programme.	- Ongoing: adopt additional medicines into the programme.	

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes this brief action plan for repurposing medicines and looks forward to the full detail.

9

Action	**Delivery partner**	**Patient involvement**	**Milestones**	**Impact measures**
<u>Reducing Health Inequalities</u> Taking steps to reduce health inequalities in NHS Highly Specialised Services (HSS) through improved	NHS England	TBC	HSS team complete health inequalities training – March 2022	Clear description of any health inequalities for each HSS

data collection, training and sharing best practice. Committing to consider health inequalities at HSS annual clinical meetings, in service development and commissioning decisions, and in provider selection processes. **Priorities: 4** (Improving access to specialist care, treatment and drugs)	Discussion of health inequalities with all services – April 2022 to March 2023 Repeat of geographic access exercise – Sep 2023 Explore how consideration of health inequalities can be incorporated into future HSS procurements – Sep 2023	Action plan in place to mitigate any health inequalities Reduction in measured health inequalities

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	(Y)	( )	( )	( )	( )
This action is sufficiently ambitious.	( )	(Y)	( )	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes any steps made to reduce health inequalities, particularly those for people affected by rare conditions. We look forward to seeing a reduction in health inequalities.

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Develop an educational resource for clinicians that can be embedded into existing IT infrastructure Developing and integrating an educational resource ('GeNotes') that will provide healthcare professionals with relevant and concise 'just-in-time' information to support patient management. In addition to linking to the NHS Genomic Test Directories, it will signpost to extended learning opportunities for clinicians. This action will be delivered in three phases: 1. User testing and publication of a minimal viable product (MVP). The MVP will include resources for	Health Education England	HEE will be looking to involve patients/family members/carers in contributing and reviewing content in phase 2 of this project. We would also be looking for input from patients/family members/carers into the evaluation strategy. We will initially be approaching HEE's People's Advisory Forum as well as members of the rare disease community to be involved.	Phase 1: March 2022 – completion of private beta testing phase March/April – launch public beta site (MVP) Phase 2: April 2022 onwards Phase 3: Starting from October 2022	Impact evaluation will form phase 3 of this action, and details are still to be determined. There will be process evaluation data from the private beta testing and public beta site, around clinician's behaviour in using the resource, usefulness of the information and anticipated impact on practice.

paediatricians, oncologists and GPs amongst others.
2. Scaling up of content production for other specialities.
3. Evaluation of the use and effectiveness of the resource.

Priorities: 2 Increasing awareness of rare diseases amongst healthcare professionals.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People would like to see coordination with patient groups that have developed materials and organisations, eg Medics4Rare. We recommend that referral pathways, listing of clinics and professionals with expertise in specific rare diseases and sign-posting to support are included.

11

Action	Delivery partner	Patient involvement	Milestones	Impact measures
<u>Inclusion of rare disease in UK health professional education and training frameworks: Establishing the evidence base</u> Action to ensure that rare disease competencies and learning outcomes are embedded in NHS education and training frameworks across all relevant specialities including general practice training and those involved with emergency care.This action will be formed of three steps: 1. Carry out a content review of current UK health professional education and training frameworks (to include curricula,	Health Education England	We anticipate that patients/family members/carers will be involved in step 1 and step 2 of this action. Involvement in step 1 will be to advise on search terms used in the content review. For step 2, we anticipate patients/family members/carers will be members of any working group that is established to identify the rare disease competencies required for each education and training framework document. We will initially be approaching HEE's People's Advisory Forum as well as members of the rare disease community to be involved.	Completion of Step 1 by April 2022. Additional milestones TBD, but work is likely to commence in 2022/23 financial year.	Report and action plan outlining: content within education and training frameworks, rare disease competencies, gap analysis identifying deficits and recommendations to address deficits.

proficiency standards etc.) to determine the current rare disease
2. Using existing generic curricula and competencies and working with professional organisations, curriculum developers and other stakeholders (including patients/family members/carers), identify the minimum level of rare disease competency needed for each education and training framework document.
3. Undertake a gap analysis to identify deficits in the inclusion of rare diseases in the frameworks.

Priorities: 2 Increasing awareness of rare diseases

amongst healthcare
professionals.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

This appears to be a positive step forward and Gene People looks forward to the outcome and evaluation.

12

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Extending the remit of the Genomics Education Programme 6-part action that looks to extend existing workstreams in the Genomics Education Programme. These include:	Health Education England	Details still to be confirmed, but patients/family members/carers will be invited to contribute to parts 2, 3, 4 and 6.	Workplans, including milestones, for each of the parts of this actions are still being developed. Some parts of the action are business as usual for the GEP and will form part of the GEPs annual workplan.	Details to be determined alongside the workplan but will include qualitative and quantitative data where appropriate. All of our resources are evaluated to assess their

• Extend the remit of the Genomics Education Programme to include non-genomic rare diseases • Increase awareness of rare disease among ‘patient-facing’ NHS staff and provide targeted resources for healthcare professional groups integral to the care of patients with rare disease • Develop resources to support the implementation of the NHS Genomic Medicine Service. • Enhance patients/family members/carers’ involvement in the GEP • Establish links with Integrated Care Systems to understand workforce issues to		effectiveness and application to practice and access to webpages evaluated via google analytics.

inform workforce planning and new ways of working.
- Develop a rare disease network and digital hub

Priorities: 1 Helping patients to get a faster diagnosis, **2** Increasing awareness of rare diseases amongst healthcare professionals.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	(Y)	( )	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes the commitment to extend the GEP and looks forward to receiving more details. We would also welcome detail of how the GEP will be marketed to HCPs.

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Develop analytical approaches within the National Congenital Anomaly and Rare Disease Registration Service to assess geographical variation and health inequalities in the use of high cost drug treatments for rare diseases (Ties into NHS-E action about monitoring drug uptake.) Enabling better understanding of the population uptake and impact of NHS England's high-cost drug commissioning policies and relevant NICE Technology Appraisals. **Priorities: 4** (Improving access to specialist care, treatments and drugs).	NHS Digital (previously Public Health England)	National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) has strong links to some of the patient groups that are eligible for high-cost drug treatment including: • Rare Autoimmune Rheumatic Disease Alliance (RAIRDA) • Wilson's Disease Support Group (WDSG-UK). We will engage with these groups (and others if needed including the relevant NHS England Clinical Reference Groups) with the support of our clinical colleagues and the National Disease Registration Service's comms team throughout. We will also	TBD	Impact will be measured by actions arising from discussion, and interpretation of results with relevant NHS England Clinical Reference Groups, Specialist Societies, NICE, patient charities and peer reviewed publication (where appropriate).

	seek advice from the Rare Diseases Forum if appropriate.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

Health inequalities are not limited to geographical areas. Gene People asks that health inequalities in their broadest sense are included and that evaluation of success is measured by a decrease in inequality of access and uptake.

14

Action	**Delivery partner**	**Patient involvement**	**Milestones**	**Impact measures**
Increasing the knowledge of rare diseases in research through publication of	NHS Digital (previously Public Health England)	NCARDRS has strong links to patient groups that are involved	NCARDRS will collaborate on and publish at least six papers describing novel findings or methods	Impact will be measured using Altmetric scores (a measure of the dissemination of a peer-

<u>epidemiology and research papers</u> Publishing high-quality epidemiological and research papers to increase the understanding of rare diseases and raise their profile. This will include papers looking at basic rare disease epidemiology, the impact of COVID-19 on people with some rare diseases and cancer-related risk factors or outcomes for people with some rare diseases. **Priorities: All** but mostly **2** (Increasing awareness of rare diseases among healthcare professionals).	in some of our ongoing work. For example: • The Registration of Complex Rare Diseases – Exemplars in Rheumatology (RECORDER) project. • The Rare Autoimmune Rheumatic Disease Alliance (RAIRDA) • NCARDRS works with multiple organisations on a project looking at histiocytic disease. This work has been partly funded by Histio UK. Histio UK are represented on the project steering group and the project team give updates at Histio UK hosted meetings. We will engage with patient representatives on existing and new projects with the support of our clinical colleagues and the National Disease Registration Service's comms team throughout. We will also seek	relevant to rare disease by 31/12/2022. NCARDRS will work with patient organisations to ensure visibility in their communities. Findings will also be disseminated through presentations at conferences and other relevant events/platforms.	reviewed paper via platforms like news outlets, social media, blogs, etc…).

	advice from the Rare Diseases Forum if appropriate.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

Gene People disagrees that the impact measurement be based on the opportunities to view for articles published: an impact measure would be akin to brand awareness surveys, ie how many of the target audience has prompted/unprompted awareness of a condition about which an article has been published.

15

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Scoping the role of rare disease registration in the delivery of the Rare Disease Framework Action Plan for England across all actions	NHS Digital (previously Public Health England)	We will work within existing partnership organisations' patient involvement structures and/or our own to ensure relevant expert patient input. For some actions,	1. With ERDFDG partners, complete review of their first round of actions 2. Agreement of partners' roles for actions where	These will be aligned to support the impact evaluation of partners actions.

Working with England Rare Diseases Framework Delivery Group (ERDFDG) partners to: • Understand if there is a role for national rare disease registration to support the delivery of their actions • Agree the role for national disease registration to deliver actions in partnership if appropriate • Begin the process to formalise data sharing and deliverable arrangements **Priorities: All**	steering groups or might be appropriate. These would include patient representation.	NCARDRS is involved 3. For actions where it is appropriate, start formal partnership agreement processes

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

Gene People suggests that there is greater scope for involving patients and patient organisations in this work. Even the smallest patient organisation maintains records on their community and there is scope for upskilling patient organisations to feed into registries on a more systematic way.

16

Action	Delivery partner	Patient involvement	Milestones	Impact measures
<u>Clinical research interface</u> Establishing the Clinical-Research Interface in order to increase the number of diagnoses from genome data, and providing evidence to support the Genomic Medicine Service in developing its diagnostic test directory. **Priorities: 1** (Helping patients get a final diagnosis faster), **4** (Improving access	Genomics England	Genomics England has a highly engaged Participant Panel who meet regularly to advise us on research participant priorities. Members of the Panel also sit on key committees including the Ethics Advisory Committee and the Access Review Committee which oversees data access processes for the National Genomic Research Library (NGRL).	December 2021 – diagnostic discovery process becomes routine, with a researcher awareness campaign in place and the NHS Diagnostic Discovery Oversight group overseeing the regular return of new diagnoses. July 2022 – patients from the Genomic Medicine Service who opt into the NGRL have started to receive diagnostic discovery results and recontacts for registry / trial eligibility	Number of diagnostic discovery results and recontact for trials or eligibility grows month on month.

to specialist care, treatment
and drugs)

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	(Y)	( )	( )	( )	( )
This action is sufficiently ambitious.	( )	(Y)	( )	( )	( )

Any additional comments (max 300 characters)

Gene People supports this action and the clear approach to impact measurement taken.

17

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Designing an ethically approved research pilot scheme for using whole genome sequencing to screen for genetic	Genomics England	The Genomics England participant panel is providing key input from the rare disease community in development of the project and its key goals. The steering group for this	Funding confirmed – January 2022 Framework for decisions related to the initial selection of genes to be returned to	Engagement opportunities offered and number of stakeholders involved Results of testing the bioinformatics pipeline

<u>conditions in healthy newborns</u> The programme will involve a series of stages of 1) engaging relevant communities to understand key design themes and questions 2) working with different groups to develop the design and test the feasibility of some aspects of it with different groups 3) Implementing the pilot at a small number of NHS trusts 4) Scaling up the pilot to recruit and sequence the relevant number of babies to provide the health data required (potentially up to 200,000). **Priorities: 1** (Helping patients get a final diagnosis faster); **3** (Better coordination of care); **4**	project has the deputy chair from the Genomics England participant panel and is also seeking two other community representatives. In addition, there is a dedicated engagement lead and Human Centred Design Researcher who will focus on investigating and testing concepts and approaches with public representatives to understand their attitudes, reactions and priorities.	participating families constructed – summer 2022 Pilot Trusts identified and engaged – summer 2022 Bioinformatics pipeline for newborn genome analysis developed and tested – end of 2022 Ethical approval obtained and patient information developed – end of 2022	on control datasets – estimated sensitivity and specificity for key conditions Number of families recruited to the pilot Number of clinically confirmed diagnoses returned to families Number of false positive genome results

(Improving access to specialist care, treatment and drugs)

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	(Y)	( )	( )	( )	( )
This action is sufficiently ambitious.	( )	(Y)	( )	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes this action and its clear approach. We ask for close involvement of patient organisations in this pilot over and above the additional two roles referred to in the action. We would like to emphasise the need for timely support being available for families having this testing.

18

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Developing a refreshed rare diseases strategy Bringing together a number of different activity strands across healthcare and	Genomics England	Genomics England has a highly engaged Participant Panel who meet regularly to advise us on research participant priorities. Members of the Panel also		Publishing the strategy will be followed by an engagement phase where impact can be measured e.g. through the number of companies using the

research within Genomics England to ensure that best outcomes are achieved for participants. This will include engaging with industry partners on how the Genomics England dataset can best meet requirements for development of therapeutics and support for rare disease clinical trials. **Priorities: 4** (Improving access to specialist care, treatment and drugs)	sit on key committees including the Ethics Advisory Committee and the Access Review Committee which oversees data access processes for the National Genomic Research Library.	National Genomic Research Library to work on development of therapeutics for rare disease.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	( )	(Y)	( )	( )
This action is sufficiently ambitious.	( )	( )	( )	(Y)	( )

Any additional comments (max 300 characters)

Gene People would suggest that the impact measures are the number of new treatments in the discovery pipeline and the number of conditions that did not previously have treatments that now do within a certain timeframe.

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Refined criteria for routing topics to Highly specialised Technologies program Supporting the access/rapid access to drugs for patients with very rare diseases through a number of refinements in the highly specialised technologies program including: • Develop refined clearer precise criteria • Consultation on refined criteria • Assess impact of proposed changes • Review and respond to consultation comments • Amend criteria in response to	National Institute of Health and Care Excellence (NICE)	Patients and other stakeholders have been involved already in helping shape the criteria. A stakeholder meeting was held in April 2021. A consultation exercise was undertaken between Feb-April 2021 to help inform the refining of the criteria. A further consultation is now in process and closes 13 October 2021	Develop and agree final HST routing criteria – December 2021 • Intention to publish new manual in January 2022 • Implementation February 2022	• Number of responses to consultation from rare disease patient groups • Final refined criteria signed off • Assessment of time between topic being considered and final routing decisions being made

	consultation comments
Priorities: 4 (Improving access to specialist care, treatment and drugs)	

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	( )	( )	( )	(Y)
This action is sufficiently ambitious.	( )	( )	( )	( )	(Y)

Any additional comments (max 300 characters)

Gene People is concerned by the refinement to the HST routing criteria as NICE has stated that previously eligible conditions will become ineligible. This is not contributing to the aims and ambition of the Rare Diseases Framework.

20

Action	**Delivery partner**	**Patient involvement**	**Milestones**	**Impact measures**
Proposed additional flexibilities in standard technology appraisal for	National Institute of Health and Care Excellence (NICE)	NICE has undertaken a range of consultations with	• Consultation close 13 October 2021	TBC

technologies for rare diseases A multi-part action proposing changes to the standard technology appraisal process, including: • Replacing the end-of-life criteria with a severity modifier • Accepting a greater degree of uncertainty when evidence generation is difficult, including rare diseases • A broad package of methodological improvements that are relevant to rare diseases, which will provide greater clarity and improved support for a comprehensive evidence base **Priorities: 4** (Improving access to specialist care, treatment and drugs)	a wide range of stakeholders Patients and other stakeholders have been involved already in helping shape the proposals. A consultation exercise was undertaken between to help inform the proposal for consultation. A further consultation is now on and closes 13 October 2021	• Intention to publish new manual in Jan 2022 • Implementation February 2022

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	( )	( )	(Y)	( )
This action is sufficiently ambitious.	( )	( )	( )	( )	(Y)

Any additional comments (max 300 characters)

Given the limitations on the NICE Methods review and the need for NICE to maintain a cost neutral approach Gene People is skeptical about the positive impact of the changes for the rare disease community. We welcome the severity modifier and would like to see the range of modifiers expanded.

21

Action	Delivery partner	Patient involvement	Milestones	Impact measures
NICE Process proposals that could have positive impacts for access for rare diseases. A range of process proposals that are relevant for all technologies evaluated at NICE and could have positive impacts for access	National Institute of Health and Care Excellence (NICE)	NICE has undertaken a range of consultations with a wide range of stakeholders Patients and other stakeholders have been involved already in helping shape the proposals. A consultation exercise was	• Intention to publish new manual in January 2022 • Consultation close 13 October 2021 • Implementation February 2022	TBC

for rare diseases. These include: **Topic selection process (within topic selection manual)** • HST topic refined routing criteria • Creation of a topic selection oversight panel (TSOP) including 2 lay representatives • Clarity on topics that can be considered for NICE guidance for therapeutics, medical devices and diagnostics • Transparency in decisions made at TSOP • Formal challenge process for routing decisions **Evaluation process (within process and methods manual)**	undertaken between February - April 2021 to help inform the proposal for consultation. A further consultation is now underway and closes 13 October 2021.	

- Earlier selection of patient experts
- Multiple Highly Specialised Technologies (M-HST)
- Commercial and managed access approaches
- Introduction of technical engagement step to HST
- Patient involvement in the development of a managed access agreement
- Virtual meetings to support patient participation where travel may be challenging
- Feedback from NICE on patient group input

Summary of information of patients

Priorities: 4 (Improving access to specialist care, treatment and drugs)

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	( )	( )	( )	(Y)
This action is sufficiently ambitious.	( )	( )	( )	( )	(Y)

Any additional comments (max 300 characters)

Gene People is deeply concerned that this action has no stated impact measures when the changes are at the point of implementation. We have commented on the potential for delays in the process through M-HST which can be catastrophic for patients in the NICE consultation.

22

Action	Delivery partner	Patient involvement	Milestones	Impact measures
Implementing UK Vision for Clinical Research Delivery Ensuring that the implementation of the Government's vision for the future of UK clinical research delivery is linked up with the UK Rare	DHSC Research Faculty, Infrastructure and Growth	The Recovery, Resilience and Growth Advisory Group includes patient and public involvement and engagement representation. Individual actions in the implementation plan also include patient and public involvement and engagement	We have published our implementation plan for 2021-22. Timelines for Phase 2 (from 2022) will be somewhat dependent on the nature of the spending review/budget announcements in 2021 (single or multi-year etc).	TBC

Diseases Framework and ensuring that this work is considered through a ‘rare disease lens’. **Priorities: 4** (Improving access to specialist care, treatments and drugs)	representatives in their design and delivery.

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	**Agree**	**Neutral**	**Disagree**	**Strongly Disagree**
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People understands the Future of UK Clinical Research Delivery vision is a general vision and not focused on rare diseases. We would welcome more detail is needed about how this vision will include rare disease.

23

Action	**Delivery partner**	**Patient involvement**	**Milestones**	**Impact measures**

Improve the way that the Government makes decisions on newborn screening for rare diseases • Establishing a new and improved UK National Screening Committee (NSC) with greater join up with researchers and greater focus on targeted screening programmes, thus improving the evaluation of screening programmes for rare diseases • Transfer of hosting the UK NSC secretariat and evidence and expert screening advisory functions to Office for Health Improvement and Disparity in DHSC allowing greater collaboration with rare disease policy team • Work to draft a paper which compares UK NSC bloodspot screening policy processes and programme delivery regime to EURORDIS criteria for good practice to identify any areas for improvement. • Describe the sort of evidence the UKNSC would find useful when attempting to	DHSC Screening Policy Team	The UK NSC, which will continue to make all recommendations concerning screening in the UK, has several lay representatives in its membership It also has feeder groups which focus on specific age groups and there are expert clinicians and lay representatives on those too.	Study of policy and programme performance will confirm quality according EURORDIS criteria. New UK NSC set to commence work in Spring 2022	UK NSC regularly gathers qualitative feedback from its stakeholders on its engagement, which can be used as one tool to understand the impact of these actions. Publication of and feedback on the testing, modelling and registry reports

understand how accurate proposed bloodspot tests are. This will allow better judgement of suitability of these tests for screening purposes and assist with setting out a research and development agenda
- Continue to develop the UK NSC's disease and economic modelling capability to support better evaluation of screening programmes for rare disease
- Investigate and describe how disease registries might be better able to inform evaluation of screening programmes for rare diseases by working with rare diseases charities to earn from their experiences

Priorities: 1 (Helping patients get a final diagnosis faster)

To what extent do you agree or disagree with the following statements?

Select the most applicable option in each row.

Grid showing question statements against rating options

	Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
This action will contribute towards implementing the Rare Diseases Framework.	( )	(Y)	( )	( )	( )
This action is sufficiently ambitious.	( )	( )	(Y)	( )	( )

Any additional comments (max 300 characters)

Gene People welcomes the commitment to faster diagnosis and the approach set out in this action. We look forward to the consultation on the proposed changes, as in our view, updating the processes for evaluating proposals for additional screening tests is long overdue.

Part 2

This section asks you to consider whether the proposed actions address each of the four priorities. Note that some actions address more than one priority so may be listed more than once.

When considering your answers, please keep in mind that this is the first step in implementing the UK Rare Diseases Framework, and that further actions will follow each year.

24a

Priority 1: Helping patients get a final diagnosis faster

- Develop a genomic testing strategy in the NHS to support diagnosis of rare diseases. (NHS England)

- Develop the NHS Genomic Medicine Service (GMS) infrastructure and review the service specification. (NHS England)

- Pilot new approaches for patients with undiagnosed rare conditions to enable faster diagnosis. (NHS England)

- Expand the remit of the Genomics Education Programme to incorporate the full spectrum of rare disease (not just those with a genomic basis), with the aim to increase awareness of rare diseases among health professionals. (Health Education England)

- Establish the Clinical-Research Interface to increase diagnostic yield from genome data, and provide evidence to support the GMS in developing its diagnostic repertoire. (Genomics England)

- Design an ethically approved research pilot scheme for using whole genome sequencing to screen for genetic conditions in healthy newborns. (Genomics England)

- Increase the understanding of rare diseases in research through publication of epidemiological and research papers. (NHS Digital (previously Public Health England))

- Scope the role of rare disease registration in the delivery of the Rare Disease Framework Action Plan for England across all actions. (NHS Digital (previously Public Health England))

- Improve the way that the Government makes decisions on newborn screening for rare diseases. (DHSC Screening Policy Team)

Thinking about these actions as a whole, to what extent do you agree or disagree that this group of actions addresses the priority?

- Strongly agree
- Agree
- **Neutral**
- Disagree
- Strongly disagree

Any additional comments (max 300 characters)

There are too many missing impact measures to be able to make a meaningful decision as to agreement or disagreement.

24b

If you selected disagree or strongly disagree, is this because:

- Additional actions are required to meet the priority
- **One or more actions listed should be more ambitious**
- There should be more evidence of the centrality of the patient voice

Other (please specify)
[ ]

25a

Priority 2: Increasing awareness of rare diseases among healthcare professionals

- Develop an educational resource for clinicians that can be integrated into existing IT infrastructure (e.g. GP digital patient notes). Proof of concept using Genomics as a case study. (Health Education England)

- Establish an evidence base for inclusion of rare disease in UK health professional education and training frameworks. (Health Education England)

- Expand the remit of the Genomics Education Programme to incorporate the full spectrum of rare disease (not just those with a genomic basis), with the aim to increase awareness of rare diseases among health professionals. (Health Education England)

- Increase the understanding of rare diseases in research through publication of epidemiological and research papers. (NHS Digital (previously Public Health England))

- Scope the role of rare disease registration in the delivery of the Rare Disease Framework Action Plan for England across all actions. (NHS Digital (previously Public Health England))

Thinking about these actions as a whole, to what extent do you agree or disagree that this group of actions addresses the priority?

- Strongly agree
- **Agree**
- Neutral
- Disagree
- Strongly disagree

Any additional comments (max 300 characters)

No comment

25b

If you selected disagree or strongly disagree, is this because:

- Additional actions are required to meet the priority
- One or more actions listed should be more ambitious
- There should be more evidence of the centrality of the patient voice

Other (please specify)
[ ]

26a

Priority 3: Better coordination of care

- Develop a digital toolkit to facilitate and improve virtual consultations. (NHS England)

- Develop the NHS GMS infrastructure and review the service specification. (NHS England)

- Design an ethically approved research pilot scheme for using whole genome sequencing to screen for genetic conditions in healthy newborns. (Genomics England)

- Increase the understanding of rare diseases in research through publication of epidemiological and research papers. (NHS Digital (previously Public Health England))

- Scope the role of rare disease registration in the delivery of the Rare Disease Framework Action Plan for England across all actions. (NHS Digital (previously Public Health England))

Thinking about these actions as a whole, to what extent do you agree or disagree that this group of actions addresses the priority?

- Strongly agree
- Agree
- Neutral
- **Disagree**
- Strongly disagree

Any additional comments (max 300 characters)

Some of the actions are difficult to link to improved health outcomes for those with rare conditions.

If you selected disagree or strongly disagree, is this because:

- **Additional actions are required to meet the priority**
- One or more actions listed should be more ambitious
- There should be more evidence of the centrality of the patient voice

Other (please specify)
[ ]

27a

Priority 4: Improving access to specialist care, treatments and drugs

- Establish a number of processes to support early access to drugs. (NHS England)

- Develop a genomic testing strategy in the NHS to support diagnosis of rare diseases. (NHS England)

- Develop the NHS GMS infrastructure and review the service specification. (NHS England)

- Develop a strategy for gene therapies and other advanced therapy products. (NHS England)

- Develop systems to measure uptake of drugs for patients with rare diseases. (NHS England)

- Continued operation of the Medicines Repurposing Programme which identifies and develops opportunities to use medicines in ways not included in the original license. (NHS England)

- Take steps to reduce health inequalities in NHS Highly Specialised Services (HSS) through improved data collection, training, and sharing best practice. (NHS England)

- Establish the Clinical-Research Interface to increase diagnostic yield from genome data, and provide evidence to support the Genomic Medicine Service in developing its diagnostic repertoire. (Genomics England)

- Design an ethically approved research pilot scheme for using whole genome sequencing to screen for genetic conditions in healthy newborns. (Genomics England)

- Develop a refreshed rare diseases strategy. (Genomics England)

- Develop analytical approaches within the National Congenital Anomaly and Rare Disease Registration Service to assess geographical variation and health inequalities in the use of high cost drug treatments for rare diseases. (NHS Digital (previously Public Health England))

- Increase the understanding of rare diseases in research through publication of epidemiological and research papers. (NHS Digital (previously Public Health England))

- Scope the role of rare disease registration in the delivery of the Rare Disease Framework Action Plan for England across all actions. (NHS Digital (previously Public Health England))

- Refine the criteria for routing topics to Highly specialised Technologies program. (National Institute for Health and Care Excellence)

- Proposed additional flexibilities in standard technology appraisal for technologies for rare diseases. (National Institute for Health and Care Excellence)

- NICE Process proposals that could have positive impacts for access for rare diseases. (National Institute for Health and Care Excellence)

- Consider rare diseases through implementation of the UK Vision for Clinical Research Delivery. (DHSC Research Faculty, Infrastructure and Growth)

Thinking about these actions as a whole, to what extent do you agree or disagree that this group of actions addresses the priority?

- Strongly agree
- Agree
- Neutral
- Disagree
- **Strongly disagree**

Any additional comments (max 300 characters)

Gene People is deeply concerned by the potential impact of the NICE review on access to medicines for those with rare conditions.

27b

If you selected disagree or strongly disagree, is this because:

- **Additional actions are required to meet the priority**
- One or more actions listed should be more ambitious
- There should be more evidence of the centrality of the patient voice

Other (please specify)
[ ]

Part 3

This section asks for some basic information about you so we can ensure we are capturing a range of voices. These questions are optional. All responses are anonymous.

28

I am a(n)...(select all that apply):

- Person living with a rare disease
- Family member
- Carer
- Healthcare professional
- Researcher
- Industry professional
- **Volunteer/employee of a rare diseases patient organisation/charity**
- Patient/public voice representative in the field of rare diseases
- Prefer not to say

Other (please specify)
[ ]

29

I have responded to this questionnaire...(select one):

- Individually
- **Based on feedback collated from different people in my organisation**
- Prefer not to say

30

Age

- 17 or younger
- 18-20
- 21-29
- 30-39
- 40-49
- 50-59
- 60 or older
- **Prefer not to say**

31

Gender

- Female
- Male
- Prefer to self describe (below)
- **Prefer not to say**

I identify as...
[ ]

32

Ethnicity (Choose one option that best describes your ethnic group or background)

- White
- Mixed/ Multiple ethnic groups
- Asian/ Asian British
- Black/ African/ Caribbean/ Black British
- Chinese
- Arab
- Other ethnic group (please specify below)
- **Prefer not to say**

Other ethnic group:
[ ]

Thank you

Thank you for completing this survey.

Your responses will be used to inform the content of England’s 2022 rare diseases action plan. The voice of the rare diseases community is a key driving force in what we do, and we truly value your input. Thank you for your time.