Medicines & Healthcare products Regulatory Agency

Consultation on Rare Disease Framework

Thank you for your interest and engagement with this proposal for a new regulatory framework for rare disease therapies. Please read the supporting framework document before contributing to the survey. We are keen to receive as much feedback as possible but most of the questions are optional so you do not need to answer all of them. There are 15 questions, you will be able to return to the survey if you have to break off before completing it.

About you (mandatory question)

Please tell us a little bit about you so that we can better understand your responses.

Which best applies to you:

- ○ I am responding as an individual
- ○ I am responding on behalf of an organisation

Are you?

- a member of the public
- a patient wth a rare disease or family member or carer of a patient with a rare disease
- a medical doctor
- another healthcare professional
- Other (please specify)

Where do you live?

- England
- Northern Ireland
- Scotland
- Wales
- I live outside the UK
- Prefer not to say
- Other - please specify

My organisation is a

- ○ Pharmaceutical or Biotechnology Trade Association
- ○ Pharmaceutical or Biotechnology Company
- ○ Patient organisation or disease charity
- ○ Health and Care organisations (e.g. NHS)
- ○ Professional Body
- ○ Clinical Research Organisation
- ○ Academic Research Organisation
- ○ Other

Please tell us the geographical area(s) your organisation covers

- ☐ England
- ☐ Northern Ireland
- ☐ Scotland
- ☐ Wales
- ☐ Outside the UK
- ☐ Other - please specify

Name of organisation (optional)

The first three questions are about the overall aim of the framework and the involvement of patient views.

1. In you view, is it desirable for more pharmaceutical treatments for rare diseases to be brought into a regulated pathway rather than used 'off-label' as many are at present? Please explain your answer.

2. The consultation sets out challenges associated with the development and regulation of treatments for rare diseases. Please identify any other challenges you are aware of that have not been considered or elaborate on those already identified. (optional)

3. Patient engagement is a key feature of the framework. How can patients and their families and carers contribute most effectively to the regulatory process? Please explain your answer. (optional)

The next section has nine questions about elements of the framework

4. A fundamental question is how much evidence is enough for a decision to be made on the use of a medicine? Using novel approaches to evidence generation the pathway aims to strike a balance between the nature and amount of data needed to support the safety and efficacy of a treatment before patients can access it and the time it takes to generate that data. Do you agree that the proposed approach has found the appropriate balance?

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

Please explain your view on this fundamental consideration (optional)

5. Defining rarity is a challenge due to the limitations of using a fixed prevalence (frequency) limit, changing disease classifications and the evolving nature of genetic and molecular disease definitions. Whilst retaining some flexibility, the framework is targeting diseases with a prevalence of around 1 in 50,000 of the UK population, which is deliberately distinct from the orphan designation threshold of 25 in 50,000. Do you consider that this threshold considered appropriate to encourage development of treatments for rare diseases ?

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

Please provide your rationale if another threshold should be considered (optional)

6. Do the Scientific Opinion and Designation steps add a tangible incentive for research and development of rare therapies? Are they best delivered at predefined points or on a flexible basis depending on the the individual development? Please explain your rationale. (optional)

7. Do you agree that open registration of all clinical trials which are linked to the pathway and transparent sharing of safety and efficacy data—especially from early-phase studies—will help in any of the following ways? (optional)

- [ ] To foster public trust
- [ ] To enhance scientific learning
- [ ] To support sustainability of the framework
- [ ] To support development of treatments for rare (and more common) conditions

Please add any further comments here (optional)

8. Proposals for managing market exclusivity have not been addressed in the framework document. How should market exclusivity be managed to balance the potential benefits of incentivising investment with the risk of creating unintended barriers to future innovation? (optional)

9. Should decisions regarding market exclusivity and reimbursement be made on the basis of a combination of pre-determined transparent criteria rather than fixed prematurely within the framework? (optional)

10. The scope of the framework includes authorised medicines that are being repurposed (being used to treat a different condition). Do you agree that the framework is suitable for these cases?

- ○ Strongly agree
- ○ Tend to agree
- ○ Undecided
- ○ Tend to disagree
- ○ Strongly disagree

Please tell us why you have given that response (optional)

11. Below we have listed six elements of the proposed framework. To what extent do you agree with each of these as an important element?; We are interested to hear of your further thoughts on them.

an adaptive regulatory pathway for very small populations (optional)

- ○ Very important
- ○ Important
- ○ Moderately important
- ○ Slightly important
- ○ Not important

Further comments (optional)

defined entry criteria (such as rarity, severity, unmet need and biological plausibility (optional)

- ○ Very important
- ○ Important
- ○ Moderately important
- ○ Slightly important
- ○ Not important

Further comments (optional)

early regulatory engagement and designation (optional)

- ○ Very important
- ○ Important
- ○ Moderately important
- ○ Slightly important
- ○ Not important

Further comments (optional)

the use of prior knowledge, platform data and adaptive evidence generation approaches (optional)

- Very important
- Important
- Moderately important
- Slightly important
- Not important

Further comments (optional)

ongoing safety monitoring with periodic review (optional)

- Very important
- Important
- Moderately important
- Slightly important
- Not important

Further comments (optional)

the potential to progress to full marketing authorisation over time (optional)

- Very important
- Important
- Moderately important
- Slightly important
- Not important

Further comments (optional)

12. Do you have questions or observations about any aspects of the framework including those we have not specifically asked about? (optional)

The last three questions ask about your view on the impact of the framework

13. In your view, will the flexibility and adaptability of the proposed pathway provide sufficient encouragement to developers of treatments for rare diseases to bring them within a regulated pathway? Please explain your answer.

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

Please add any further comments here and if you don't think this aim will be achieved please explain why.

14. In your view, will the pathway achieve its aim of supporting innovation while safeguarding patients and maintaining confidence in regulatory decision-making?

- ○ Strongly agree
- ○ Tend to agree
- ○ Undecided
- ○ Tend to disagree
- ○ Strongly disagree

Please add any further comments here and if you don't think this aim will be achieved please explain why. (optional)

15. In your view could this framework reduce barriers to patient access, including time and cost, and in turn help reduce overall treatment costs?

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

Please add any further comments here and if you don't think this aim will be achieved please explain why.

Equality Impact

Do you think the proposals risk impacting people differently, or could impact adversely on any of the protected characteristics covered by the Public Sector Equality Duty set out in section 149 of the Equality Act 2010 or by section 75 of the Northern Ireland Act 1998? (optional)

- ○ Yes
- ○ No
- ○ No opinion

Satisfaction Survey

If you do not wish to leave your feedback about the survey please select the 'Next' button.

The Rare Disease Framework document was easy to understand (optional)

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

It was easy to contribute to this consultation (optional)

- ○ Strongly agree
- ○ Agree
- ○ Neither agree or disagree
- ○ Disagree
- ○ Strongly disagree

Review your answers

About you (mandatory question)

Which best applies to you:	Unanswered	Change
Are you?	Unanswered	Change
Where do you live?	Unanswered	Change
My organisation is a	Unanswered	Change
Please tell us the geographical area(s) your organisation covers	Unanswered	Change
Name of organisation	Unanswered	Change

The first three questions are about the overall aim of the framework and the involvement of patient views.

In you view, is it desirable for more pharmaceutical treatments for rare diseases to be brought into a regulated pathway rather than used 'off-label' as many are at present? Please explain your answer.	Unanswered	Change

The consultation sets out challenges associated with the development and regulation of treatments for rare diseases. Please identify any other challenges you are aware of that have not been considered or elaborate on those already identified.	Unanswered	Change
Patient engagement is a key feature of the framework. How can patients and their families and carers contribute most effectively to the regulatory process? Please explain your answer.	Unanswered	Change

The next section has nine questions about elements of the framework

A fundamental question is how much evidence is enough for a decision to be made on the use of a medicine? Using novel approaches to evidence generation the pathway aims to strike a balance between the nature and amount of data needed to support the safety and efficacy of a treatment before patients can access it and the time it takes to generate that data. Do you agree that the proposed approach has found the appropriate balance?	Unanswered	Change
Please explain your view on this fundamental consideration	Unanswered	Change

Defining rarity is a challenge due to the limitations of using a fixed prevalence (frequency) limit, changing disease classifications and the evolving nature of genetic and molecular disease definitions. Whilst retaining some flexibility, the framework is targeting diseases with a prevalence of around 1 in 50,000 of the UK population, which is deliberately distinct from the orphan designation threshold of 25 in 50,000. Do you consider that this threshold considered appropriate to encourage development of treatments for rare diseases ?	Unanswered	Change
Please provide your rationale if another threshold should be considered	Unanswered	Change
Do the Scientific Opinion and Designation steps add a tangible incentive for research and development of rare therapies? Are they best delivered at predefined points or on a flexible basis depending on the the individual development? Please explain your rationale.	Unanswered	Change

Do you agree that open registration of all clinical trials which are linked to the pathway and transparent sharing of safety and efficacy data—especially from early-phase studies—will help in any of the following ways?	Unanswered	Change
Please add any further comments here	Unanswered	Change
Proposals for managing market exclusivity have not been addressed in the framework document. How should market exclusivity be managed to balance the potential benefits of incentivising investment with the risk of creating unintended barriers to future innovation?	Unanswered	Change
Should decisions regarding market exclusivity and reimbursement be made on the basis of a combination of pre-determined transparent criteria rather than fixed prematurely within the framework?	Unanswered	Change
The scope of the framework includes authorised medicines that are being repurposed (being used to treat a different condition). Do you agree that the framework is suitable for these cases?	Unanswered	Change

Please tell us why you have given that response	Unanswered	Change
an adaptive regulatory pathway for very small populations	Unanswered	Change
Further comments	Unanswered	Change
defined entry criteria (such as rarity, severity, unmet need and biological plausibility	Unanswered	Change
Further comments	Unanswered	Change
early regulatory engagement and designation	Unanswered	Change
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ongoing safety monitoring with periodic review	Unanswered	Change
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the potential to progress to full marketing authorisation over time	Unanswered	Change
Further comments	Unanswered	Change
Do you have questions or observations about any aspects of the framework including those we have not specifically asked about?	Unanswered	Change

The last three questions ask about your view on the impact of the framework

In your view, will the flexibility and adaptability of the proposed pathway provide sufficient encouragement to developers of treatments for rare diseases to bring them within a regulated pathway? Please explain your answer.	Unanswered	Change
In your view, will the pathway achieve its aim of supporting innovation while safeguarding patients and maintaining confidence in regulatory decision-making?	Unanswered	Change
Please add any further comments here and if you don't think this aim will be achieved please explain why.	Unanswered	Change
In your view could this framework reduce barriers to patient access, including time and cost, and in turn help reduce overall treatment costs?	Unanswered	Change

Equality Impact

Do you think the proposals risk impacting people differently, or could impact adversely on any of the protected characteristics covered by the Public Sector Equality Duty set out in section 149 of the Equality Act 2010 or by section 75 of the Northern Ireland Act 1998?	Unanswered	Change

Satisfaction Survey

The Rare Disease Framework document was easy to understand	Unanswered	Change
It was easy to contribute to this consultation	Unanswered	Change

Thank you for your engagement, we aim to publish our response to the survey in the early autumn.

This survey is now closed.