RARE DISEASE UK PATIENT EMPOWERMENT GROUP

Recommendations for the UK Rare Diseases Framework

Following the publication of the UK Rare Diseases Framework in January 2021, Rare Disease UK's Patient Empowerment Group (PEG) has held five meetings to conduct a deep dive into the four priorities and five underpinning themes set out in the Framework. The meetings explored each of the priorities to determine next steps to support people affected by rare conditions. This document summarises the key recommendations which arose in these meetings.

We have moved the topic of discussion to the most relevant priority or underpinning theme.

Summary

The Patient Empowerment Group recommends that the action plans based on the UK Rare Diseases Framework include:

1. The development of a system which closely monitors the diagnostic odyssey and implements a flagging system.
2. A restrucutring of the UK approach to newborn screening - including an expansion the number of conditions screened for and introducing a committee dedicated to newborn screening with members who have specialties in rare conditions.
3. A new method of integrating rare conditions into the whole journey of medical professional training and education must be devised.
4. Implementation of the recommendations laid out in the CONCORD project.
5. Exploration of the potential of repurposing drugs.
6. Supporting clinical guidelines developed by patient organisations.
7. Encouraging the use of telemedicine.
8. Integrating the voice of the rare disease community into every level of decision-making
9. Encouraging collaboration both with the EU and internationally.
10. Establishing a common language and data sharing structure for research into rare conditions which is compatible with international methods.
11. Incentivising clinicians to become more involved in rare conditions, including research and particularly into ultra-rare conditions

12. Launching a **rare disease specific UK registry**

Priority 1: helping patients get a final diagnosis faster

Detecting the diagnostic odyssey
The diagnostic odyssey presents a barrier to treating a rare condition. On average, it takes four years for someone to be diagnosed with a rare condition. Given that the diagnostic odyssey can be lengthy, it is important that people with rare conditions are treated throughout their diagnostic journey. However, without a diagnosis it is not possible for many people living with a rare condition to access specialist care. This leaves people feeling abandoned and 'stuck' in their diagnostic odyssey.

'In total 62% of respondents without a diagnosis have been undiagnosed for more than five years since they first contacted a healthcare professional about the onset of their symptoms' - Rare Experience 2020 Report, P. 14

PEG recommends that a project is launched to lay the foundations of a process to monitor an individual's journey to diagnosis and a flagging system for when an individual passes a determined time to diagnosis. The project should identify the needs of the community from such a system and determine the process and practicalities.

Undiagnosed conditions fit into two categories:

1. People who are living with an identifiable rare condition. They could be diagnosed if the right diagnostic tool was applied or they met the appropriate clinician to make a diagnosis.

   'There appears to remain a significant portion (45%) of those without a diagnosis who have not had the benefit of genetic testing to identify the condition that affects them' - Rare Experience 2020 Report, p. 19

2. People who are affected by a condition which has not been properly identified and characterised either through identification of the causative gene or through a clinical definition by a clinician – a syndrome without a name. They cannot be diagnosed until this condition is characterised.

The recent Autumn Budget and Spending Review 2021 demonstrates a commitment to developing whole genome sequencing for newborn screening. 80% of rare conditions are genetic, of which 70% start in childhood. The focus on genomics therefore caters for the majority of people living with a rare condition.

However, the current structure of the UK National Screening Committee is not equipped to support the level of change that will hopefully come from this project. The committee themselves have admitted that they are under equipped with such a small team to fully evaluate all evidence presented in an application. Additionally, there is a lack of

consideration for the difficulty in producing evidence due to the absence of expert opinion in rare diseases across the committee.

So that this investment effectively serves the community, **PEG recommends that the UK National Screening Committee should be restructured**. ArchAngel Newborn Screening campaign advocates for a change in the structure and bureaucracy of the UK National Screening Committee.

- **Change the structure and the positioning of newborn screening** within the wider screening body and separate out newborn screening to give this area of screening a committee which stands alone, focused on newborn screening with the potential to engage relevant experts on a per condition basis.
- **Streamline the evidence review process** to make it relevant to rare diseases rather than the wider population programmes, and to ensure that it would accept a wider range of evidence from validated and reputable sources.
- **Establishment of a set timeframe** so that the process can be carried out with efficiency and accountability.

PEG also recognises that biochemical screening for rare conditions is not mentioned in the UK Rare Diseases Framework. While whole genome sequencing is an important step to achieving diagnosis for all, it is still not fully developed. 20% of rare conditions are not genetic and there are several genetic conditions which are better diagnosed via biochemical screening. However, there has been a complete reluctance to add new conditions to the programme with no new conditions being added to the heel prick blood test since 2017 in Scotland, 2015 in England and 2009 in Northern Ireland. It is therefore important that the action plans include steps to **support the expansion of the list of conditions tested for in the heel prick blood test.**

Regarding the second category of undiagnosed people, the **findings from the Welsh syndromes without a name (SWAN) clinic pilot should be used in the other nations** to implement an infrastructure to support those without a positive diagnosis and to connect them with research..

PEG's response to the second category of those with a syndrome without a name is answered under the underpinning theme of Pioneering Research.

Priority 2: increasing awareness of rare diseases among healthcare professionals

PEG recommends that healthcare professionals are better informed on rare conditions as a group and the common challenges which they face as opposed to trying to improve knowledge of specific rare conditions.

The Rare Experience 2020 Survey revealed that patients do not expect healthcare professionals to be experts in every rare condition as this is unrealistic but they need to be willing to learn from the patient and understand their experiences.

'Nearly half (45%) of diagnoses for respondents to the Genetic Alliance UK Rare Experience Survey were made by doctors who specialise in specific rare conditions. This demonstrates the importance of the referral pathway. For these diagnoses to be made the primary and secondary healthcare providers need to continue the referral chain to the specialist clinicians who are able to make these diagnoses.' - *Rare Experience 2020 Report- P. 17*

PEG identifies the following as gaps in knowledge for healthcare professionals:

- General awareness of rare conditions
- Tools and resources available for diagnosis
- Care pathways and methods for referral - there needs to be more rigid guidelines on pathways to ensure consistency in the service
- Issues of intersectionality and rare diseases

'There appears to be an issue with patients with rare conditions being misdiagnosed with mental health conditions, as some respondents indicated that their misdiagnosis was sometimes due to physical symptoms being treated as psychological symptoms'. *Rare Experience 2020 Report, P. 17*

A new method should be devised to integrate rare conditions into the whole journey of medic training and education, tailored to the different HCPs depending on the service that they supply to the patient.

Medics 4 Rare Diseases provides education in the Rare Disease field for medical students and doctors in training via 'Rare Disease 101' on the common challenges faced by people affected by rare conditions. The programme identifies the following as ways to address the lack of awareness of rare conditions in the healthcare system.

- **Implementing broader general awareness of the challenges to rare conditions, the impact on the patient and where to find information on rare conditions consistently in undergraduate medical degrees** - as stated in the 2013 strategy for rare diseases.

- Opportunities for **tailored continual professionals development for different healthcare professionals.**
- **Whole-system signalling is required.** The health system needs to tell its workforce that rare diseases are relevant and important. In order for this to happen we need data, education, guidelines and tools as well as pathways.

'On the whole, respondents felt that healthcare professionals did not have sufficient information about their rare condition. In particular, people reported a lack of knowledge of healthcare professionals outside of the hospital system'. *Rare Experience 2020 Report, P. 22*

To further support healthcare professionals in the diagnosis and treatment of rare conditions, they need to be provided with the correct tools. PEG identifies the following as areas which need to be highlighted in the action plans.

- Access to patient organisations
- Tools to manage complex rare disease cases
- Clearly defined pathways
- Increasing incentives for HCPs

Priority 3: better coordination of care

Everyone diagnosed with a rare condition will need a care pathway. The action plans should focus on improving care coordination for those that already have it, and also addressing the fact that there are those that do not have access to coordinated care.

'The patient / carer is the one who coordinates the majority of care for 71% of the respondents. The next most common coordinator for our respondents was 'shared responsibility across professionals with no single coordinator' – 12%. Taken together, at least 83% of respondents did not have a single care coordinator provided by the health service.' Rare Experience 2020 Report, P. 30

In this way, there is a need for an **assessment of current care coordination in the NHS** first.

The CONCORD project explores whether and how care of people with rare conditions is coordinated in the UK, and how patients, families and professionals would like them to be coordinated. PEG recommends that the **findings from the CONCORD project are used** and converted to measurable realistic actions.

The CONCORD project identified six categories in which care can be coordinated (The lettered bullet points are specific points raised by PEG):

1. **Ways of organising care:** National centres, local centres, hybrid options - there is a preference towards national specialist centres and a hybrid model.
2. **Ways of organising those involved in care:** research showed that this ranged from little collaboration to high levels of collaboration e.g. condition specific clinics. There was a middle ground of collaboration e.g. this could be between a point of transition between paediatric and adult services.
    a. Flow charts to help address/identify a care pathway for those who are undiagnosed.
    b. There is a disconnect between genetic services and then what happens next. Care coordination should start from the moment that an individual enters the health system.
3. **Responsibilities:** There are different types of roles involved in coordination. This could range from an administer level e.g. organising your appointment and having a point of contact to someone that is responsible for your care overall. GPs were involved in providing care at a local level and charities played a huge role in providing care and support too.
    a. Safety nets need to be introduced to limit rare conditions being forgotten.
4. **How often appointments take place:** varies depending on the condition and the individual's needs. Some conditions need more regular monitoring.

5. **Access to records:** some people wanted information to be filtered to HCPs on a need to know basis, others were happy to have information shared with all HCP involved in their care. People living with rare conditions also wanted access to these records.
    a. Facilitating access for HCPs is important to prevent gaps in care.
    b. There should be a general increase in communication between HCPs as well to improve sharing information and reduce duplication of efforts.
6. **Mode of communication:** digital appointments, face-to-face appointments, letters following an appointment. Some highlighted the benefits of digital communication but also mentioned that it can't replace face-to-face appointments, especially for getting a diagnosis.
    a. Telemedicine has proved highly beneficial to the community if used at appropriate times. The option should be given to patients. The technology needs to match the need e.g. video calls as opposed to phone calls

'Generally, for respondents who had had the opportunity to make use of various technologies, they considered them more useful than not … Respondents' negative experiences with technology centred around phone or video conferences not being as suitable for them as face to face consultations, especially if any physical examination was needed' *Rare Experience 2020 Report, P. 48*

The key areas identified by PEG where care coordination is currently lacking is the transition from paediatric to adult care and the transition from diagnosis to treatment.

'On the whole respondents who had experienced transition between child and adult services were not very satisfied with the experience. Just a third (32%) of these respondents were either satisfied or very satisfied with transitioning between child and adult services, almost a half were unsatisfied or very unsatisfied with the experience. For those who had transitioned between child and adult services there was a sense that they were having to 'start all over' again.' - *Rare Experience 2020 Report, P. 47*

Priority 4: improving access to specialist care, treatment and drugs

PEG drew on some key points of interest relating to access to specialist care treatment and drugs.

PEG recommends that the action plans support the writing of guidelines and promote their use. To do this the actions plans should:

- Support research to supply guidelines with evidence
- Supply greater incentives for clinicians to become involved in rare conditions.
- Support the approval of patient organisation developed guidelines.

More Centres of Excellence that cover related 'groups' of rare diseases are needed to improve access to specialist care. These extra centres can help to better understand the conditions as well as improve standards of care and reduce the travel burden.

'Respondents who were able to access specialist centres spoke very positively about them. They valued being able to see multiple healthcare professionals in one trip. Having access to doctors who are experts in their rare condition also meant that they then do not have to repeatedly explain their condition'. - Rare Experience 2020 Report, P. 36

'Nearly half (48%) of respondents said there was a specialist centre for their condition, around 1 in 5 (21%) said there was not and nearly a third (32%) were unsure'. Rare Experience 2020 Report, P. 36

Repurposing drugs is a potential route to offer treatments more cheaply and efficiently but there is currently a lack of incentives and regulatory flexibility.

Repurposing needs to be followed up with attention to the supply also. There is currently a petition supported by PEG to establish a national UK manufacturer of essential, off-patent, generic medicines (such as hydrocortisone, liothyronine or insulin) within the NHS. Make these medicines at close to production cost, in order to prevent the unethical practice of price gouging.

PEG also recognises that repurposing medical devices is missing from the Framework and how that can be implemented.

Underpinning themes

Patient voice

The Framework needs to **promote a representative voice in decision-making processes throughout the NHS system** by facilitating the voice of the rare disease community at every level of decision making by providing training and ensuring that the voice is fully integrated into decision-making processes from the start.

- Lacking representation for youth voice and Black and Asian minority ethnic communities
- Meetings should be made accessible ie.e outside of usual work hours as many representatives have full time jobs / education.
- Training on becoming a patient public voice representative is needed to attract different people and could contribute to addressing diversity concerns.

The community voice needs to be present at every level. It is important to include the community voice in the development of the integrated care systems.

National and international collaboration

Collaboration between the UK and EU should continue and information sharing should be bilateral. We need to take the best practices from ERNs and see if we can replicate this in the UK.

Collaboration must also extend beyond Europe, looking for opportunities internationally which could benefit people with rare conditions.

Pioneering research

Due to the nature of rare conditions, there is limited research in this area which has restricted the evidence being fed into diagnosis and treatment development thus having a significant impact on people with rare conditions.

'Because any given rare disorder affects so few patients, companies often are reluctant or unable to invest the years of research and millions of dollars necessary to develop, test and bring individualised gene therapy treatments for a single disease to market.' - Dr Rutter.

The action plans should make efforts to **incentivise research for clinicians**, in particular research into those conditions which have previously been neglected. Research must be of high quality, in line with international standards and translational.

The framework should **develop a method of sharing ongoing research projects for rare conditions** to more easily identify gaps in research and prevent duplication of efforts. Also, so that patient groups can support this research.

When producing research, PEG recommends that the action plans **promote the consistent use of a common language** to align with international standards in order to facilitate the use of the evidence.

The action plans should further support the 'less usual' formulas or methods of analysis and assessment to be accepted and acknowledged as evidential due to the challenges which the rarity of the population can cause.

Digital, data and technology

For this theme, PEG identifies that a **UK wide registry for rare conditions is necessary** for accurate data capture. The goal is for these registries to better support patient-focused research.

- To link internationally, they must be well structured and be consistent - metrics need to be based on an authoritative and international standard in order to achieve consistency in the data.
- This registry should expand on the work done under other relevant registries such as National Congenital Anomaly and Rare Disease Registration (NCARDRS), Congenital Anomaly Register and Information Service (CARIS) and Congenital Anomalies and the Rare Diseases Registration and Information Service for Scotland (CARDRISS).
- The registry must be well funded.

> Respondents on the whole wanted to take part in research but they wanted it to be easier to find out about research projects and to be made aware of the results when the research was complete. - Rare Experience 2020 Report, P. 45

Wider policy alignment

There is a distinct lack of mention of the UK Rare Diseases Framework in other policy documents.

For instance, there needs to be a bridge between the Framework and the integrated care system to ensure that the ICS are fully equipped to respond to rare disease issues. Equally, the National Institute for Health and Care Excellence (NICE) methods and process review should consider the aims of the UK Rare Disease Framework in its methods and process review.

PEG also recognises that not all points of interest raised in the National Conversation are covered in the UK Rare Diseases Framework. As such, it is important that the action plans highlight these other issues as well as the priorities identified in the Framework.

The UK Rare Disease Framework is independent of policies that are already taking place. As such, the framework should introduce new policies which expand on previous policy.