The new UK Rare Diseases Framework Write-Up

by James Charlick, Assistant Administrator, following attendance at a Genetic Alliance UK Briefing Meeting on 11 January 2021.

In addition to this write-up report, see the News section for The Pituitary Foundation's Chief Executive's response to the launch of the new Framework.

On Saturday 9 January 2021, the UK Government published a new UK Rare Diseases Framework. This document sets out the underpinning themes and priorities for the next five years that must be acted upon if the lives of people with rare diseases are to improve.

The Framework is called 'Phase 1' in this two-phase approach, with 'Phase 2' being the action plans that each of the four devolved nations must draw up separately in the coming years under the Framework. Genetic Alliance UK is a group of over 200 organisations dedicated to supporting people affected by genetic conditions; Rare Disease UK is a subsidiary organisation of Genetic Alliance UK, and provides a united campaigning voice for the rare diseases' community. Campaigns run by Rare Disease UK contributed significantly to the implementation of a new Rare Diseases Framework.

The Framework is structured around three core sections: i) four most important priorities in the rare disease community; ii) five underpinning themes that will support the enactment of the four priorities; iii) eight core principles that allow continuity between the action plans of the four nations.

Here are the three core sections of the Framework in brief:

Phase 1

Priorities

1) Helping patients get a final diagnosis faster
2) Increasing awareness of rare diseases among healthcare professionals
3) Better coordination of care
4) Improving access to specialist care, treatments and drugs

Underpinning themes

1) Patient voice – make sure patients, families and organisations that support patients are at the centre of decision making; handling inequality.
2) National and international collaboration – rare disease research and healthcare practitioners collaborating will allow better information dissemination and increase the chance that patients' lives are improved.
3) Pioneering research – taking advantage of the UK's world-leading science industry and continuing to invest in rare disease research.
4) Digital, data and technology – utilise the benefits of revolutionary data science and easy access to registries, such as NCARDRS in England, CARIS in Wales and CARDRISS in Scotland.

Phase 2

Principles

1) Deliver the aims of the UK Rare Diseases Framework under each of the priorities and underpinning themes.
2) Consider where action plans can contain specific and measurable commitments under each focus area and regularly review commitments (every 1 to 2 years).
3) Develop policy commitments with expertise, in close collaboration with patients and others living and working with rare diseases
4) Ensure any impacts on health inequalities are considered when developing action plans
5) Ensure that the experiences of rare disease patients during the COVID-19 pandemic are reflected in the development of action plans and implementation of framework priorities and themes
6) Ensure that the voice of the rare diseases community is recognised across the system and that work, as part of the UK Rare Diseases Framework, is aligned with other relevant policy development, such as mental health and social care
7) Work collaboratively across nations to share knowledge and best practice
8) Review progress made towards the aims of the framework every 5 years and update priorities when necessary.

Further observations about the Framework

- The criteria for selecting the four most important priorities set out in the Framework is not discussed within the document itself. This raises the possibility that valid priorities of the rare diseases' community are not covered by the Framework and therefore receive less attention in the action plans developed by each nation.
- Clear and significant progress in the diagnosis of rare diseases had been made prior to the development of the new Framework. Nevertheless, the Framework focuses heavily on improving diagnosis where it could have prioritised aspects of the patient journey that traditionally receive less attention.
- The Framework emphasises the use of genomics in diagnosis of rare diseases. Although the Framework states that improvements in the diagnosis of non-genetic rare diseases should also be considered, it is clear that the Framework aims to establish genomic testing as a standard in the diagnosis of rare diseases. This creates the potential for non-genetic rare disease diagnostics to be side-lined.

Summary

The new UK Rare Diseases Framework sets out a two-phase commitment to improving the lives of people with rare conditions. The Framework lists 'high-quality/high-level' statements that, if acted upon, would deliver this commitment. Even though the publication of the new Framework is a positive development for the rare diseases' community, in practice, the Framework presents a challenge for the people and organisations responsible for its delivery.

The statements that make up the Framework are ambitious, detailing the ideal approach to improving the lives of people with rare conditions. However, as yet none of the four nations have set out a timeline or plans to fund the development of an action plan as part of Phase 2. Timely development of each nation's action plan will therefore require further pressure from the rare diseases' community.