The key to unlocking the Rare Disease Framework lies in Priority Two

by Dr Lucy McKay , CEO of Medics4RareDiseases

Medics4RareDiseases is driving an attitude change towards rare diseases amongst medical students and doctors in training in order to reduce the diagnostic odyssey and improve the patient journey. We've been doing this in some form or another for 10 years after starting as a medical student society. Therefore we whole heartedly agree that systemic change is need within healthcare if we're going to achieve the best outcomes for those living with a rare disease. It also means we have experience in making this happen. I believe that while the UK Rare Disease Framework has identified four key priority areas those creating the Action Plans need to realise that one of those Priorities is the key for unlocking the other three: raising awareness of rare diseases among healthcare professionals. This is going to be the hardest obstacle to concur but it will allow for the other three priorities to truly come into their own. Here I lay out how we all need to approach "awareness".

The UK Rare Diseases Framework 2021

On 9th January 2021 the greatly anticipated UK Rare Diseases Framework was published by The Department of Health and Social Care. This important update to the original 2013 UK Strategy outlines key priorities that need real action in order to improve the lives of those living with rare diseases, as well as supporting the personal and professional communities around them.

Representing Medics4RareDiseases I took part in the Big Conversation announced by Baroness Nicola Blackwood in July 2019 that resulted in the creation of this new Framework. My message to the team at the DoHSC was that medical education in the basics of rare disease is essential to build on the commitments made back in 2013. You cannot expect change to be implemented when those you need to implement it aren't aware of the problem you're trying to solve. Early and consistent medical education about rare disease will prepare the medical workforce to work alongside other stakeholders to achieve any future goals. I also advocate the retiring of the meaningless and powerfully obstructive phrase *"common things are common"* from medicine.

The Framework published this month represents Phase One of two and it focuses on four key priorities, as well as five underlying themes. It "sets out a high-level vision for each of these priority areas, shared by all UK nations, providing a strategic direction for the UK's work on rare diseases across the next 5 years, at which point it will be reviewed." During Phase Two each nation will decide on an action plan that they will implement in order to achieve this vision. This is where the real change will come from and the M4RD team plans to put all the knowledge and experience we've gained over the last 10 years to help inform Phase Two.

The Framework is a testament to the progress M4RD has made

The four key priority areas will not be surprising to anybody who has attended an M4RD event - which is reassuring in consistency but shows that progress is still slow.

UK Rare Diseases Framework 2021

4 PRIORITY AREAS:

1. Helping patients to 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 treatment and drugs

Priority 2 is the raison d'etre of Medics4RareDiseases and so it is great to see it clearly identified as a Priority by DoHSC. M4RD has consistently messaged that it is unfeasible for any medical professional to know about over 7000 rare diseases therefore we need to take a non-disease specific approach to medical education about what it's like to live with a rare disease and how medics can help shoulder some of the burden. I was glad to see the new Framework make reference to this way of thinking which demonstrates that the rare disease community are thinking more strategically about how we approach this issue than each individually competing for time on curricula. Medics4RareDiseases focuses on medical professionals in particular because they are key to diagnosis, care coordination and treatment. We ask them to #daretothinkrare and focus on the common challenges faced by people with different diseases.

The inclusion of this statement in The Framework demonstrates progress and is also exciting for me personally. When I first started public speaking on the issue this was a new concept and even at rare disease events I could receive resistance to it. However this model of thinking isn't new in medicine or healthcare.

We have other patient models or scenarios that are not united by an underlying condition but rather similar needs, risks and impact that patients share. For example every medical student should be able to start a management plan for an admission of an immunocompromised patient, whether this is due to systemic disease, infection or an iatrogenic cause.

We must strive for a medical profession that can similarly hear "rare disease patient" and think of coordinated care, multi-system disease, research, mental health support, etc. This is an achievable goal and M4RD is already putting the hard graft in to make it happen with events, medical student engagement and soon Rare Disease 101.

This leads to some key questions that must be defined and answered before the work progresses to Phase 2 which I will lay out below.

QUESTIONS THAT NEED ANSWERING FOR PRIORITY TWO

Increasing awareness of rare diseases among healthcare professionals

WHO
Who are you targeting?
Doctors? Nurses? AHPs?
Diagnosers? Coordinators?

WHAT
What does "awareness" look like?
What do you want "awareness" to achieve for patients?

HOW
How is "awareness" going to be achieved? How will we know it's been achieved?

WHEN
When can actions be implemented?
Short and long term actions?

WHO do we mean when we talk about "Healthcare Professionals"?

Priority 2 is a lofty goal but it is achievable. "Healthcare professionals" equates to a huge group of people consisting of nurses, paramedics, midwives and doctors and 14 allied healthcare professions. As we create the actions on the action plan we must define the **WHO** for each element.

Each of these professions will have a unique insight into a patient's experience and will play their own role in that patient's journey. Even within one profession, Medicine, a doctor's skills and experience can vary wildly depending on their training level, the rotations they've had, the hospitals they have worked in and the specialism they are pursuing. Therefore this one heterogenous group described as "healthcare professionals" needs breaking down in order to create specific actions that are achievable for each individual.

WHAT do we specifically want from healthcare professionals?

The approach needed for increasing awareness of rare diseases among healthcare professionals is going to be different depending on the profession and their experience, but it will also depend on one other factor: **WHAT** specifically are we trying to achieve with this "awareness"? This is where the other three priorities come into play: 1. Helping patients get a final diagnosis faster 3. Better coordinated care and 4. Improving access to specialist care, treatment and drugs.

We will need to target different professions and different groups within those professions in order to achieve the change needed for each of the other three priorities. A GP is going to need a different focus than a Clinical Geneticist when it comes to diagnosis. A surgical ward nurse is likely to need a different approach to a Specialist nurse in a Tertiary Centre in order to better coordinate care.

HOW can we approach Priority 2?

Priority 2 is not a small one. It needs a strategy of its own. I have been thinking about **HOW** to tackle this problem and suggest a two-armed approach. Arm 1 is a Healthcare Professional (HCP) -Led Approach (The **WHO** Approach). Arm 2 is a is a Priority-Led Approach (The **WHAT** Approach).

THE WHO APPROACH

STEPS

1. Who makes up this specific group?
2. What role does this group play in each of the other 3 priority areas?
3. With which Priority could we make the greatest improvement with this group?
4. What are the key messages we want this Group to hear to affect this priority?
5. What changes can they make today to further this priority?
6. What changes can they start implementing to achieve this priority within 5 years?

EXAMPLE

1. GPs
2. GPs are most likely to be the first point of contact in a patient's diagnostic pathway. They can be the difference between a timely diagnosis and an odyssey. GPs should also be recognised for the pivotal part they can play in coordinating a patient's care. GPs are likely to play a smaller role in Priority 4, although should be aware of the obstacles here so they can support coordinated care.
3. Concentrate on Priorities 1 and 3.
4. Prevalence of rare disease, relevance to their practice, opportunities arising from timely diagnosis, cost benefit of coordinated care for the patient and NHS.
5. Have a Practice Rare Disease Action Plan outlining how they will support the 2020 Framework and subsequent Action Plans.
6. Look at training and roles within the practice, thinking about opportunities such as having a GP with an Extended Role in Genomics.

THE WHAT APPROACH

STEPS

1. What is the greatest priority?
2. Which groups could we make the greatest gain from concentrating on?
3. What are the key messages we want them to hear to affect this priority?
4. What changes can they make today to further this priority?
5. What long-term changes are needed to achieve this priority within 5 year?

EXAMPLE

1. Helping patients to get a final diagnosis faster.
2. General practitioners, general paediatricians, neurologists and general internal medics.
3. Focus on prevalence of rare disease as a whole and relevance to their everyday clinical practice.
4. Providing tools for suspecting when someone has a rare disease (but not expecting them to suggest a specific rare disease). Providing consistent support for decision making and receiving expert/specialist advice.
5. Rare Disease 101 training in undergraduate and postgraduate studies.

We need patients and healthcare professionals both sharing experiences to unravel this issue

As always with my work, I am trying to bring together two pieces of cloth that make up the fabric of my life: my childhood following around a rare disease advocacy trailblazer, Christine Lavery, and my adult life training to be a doctor. When I started medical school I couldn't make these two parts of my life reconcile so I now spend my working life trying to make it happen.

So the above should be taken as a thought-experiment from a very interested onlooker with more than the average understanding of the rare patient experience and clinical medicine. We need patients **and** healthcare professionals to share their experiences in order to understand the obstacles standing in the way of each of the 4 priorities. 68% of the responses to the Government survey were from patients which reassures me that the priorities chosen therefore reflect true unmet need. However only 7% were from healthcare professionals so now we need to hear from those in the healthcare workforce to understand why this is.

It is imperative that those involved in creating the action plans, that follow this Framework, consult the frontline professionals that they target and who will be expected to implement those plans. If we want to make a real difference in this area we need to be bringing healthcare professionals into the conversation from the very beginning.

Medical Education, Medical Education, Medical Education

We need to now look to the group of people that are pivotal to three of its four priorities and the focus of the remaining priority - healthcare professionals. We know about the unmet need and now we need to equip those who can fulfil the needs to do so. This process starts with consulting healthcare professionals about why they believe these unmet needs still exist despite the efforts following the UK Rare Disease Strategy in 2013. My suspicion is it will be due to a basic lack of understanding of the relevance of rare disease to everyday clinical practice.

This is why I believe that everything we do in rare disease needs to be complemented by medical education in order to systematically improve the patient experience and reduce the diagnostic odyssey.

Written by Dr Lucy McKay on 26th February 2021
Rare Disease 101 is an online medical education module that will be free to access from the M4RD website from 16th February 2021.