COMMUNITY CHECK-IN NOTES

Tuesday 9 February: Rare Disease 101

Organisations represented:

- Medics 4 Rare Disease
- Alex TLC
- Behcet's UK
- Duchenne Family Support Group
- HSP Support Group
- PKD Charity
- Unique
- Wolfram Syndrome UK

Genetic Alliance UK staff attending:

- Lauren Roberts, Director of Support
- Nick Meade, Director of Policy
- Amy Hunter, Director of Research
- Farhana Ali, Public Affairs Manager
- Natalie Frankish, Policy and Engagement Manager - Scotland
- Emma Hughes, Policy and Engagement Manager - Wales
- Sophie Peet, Policy Officer

Nick Meade welcomes everyone and introduces the session with M4RD (Medics 4 Rare Disease).

Nick Meade – introduction:

- Nick introduces Lucy McKay, the CEO of M4RD, who will share slides of her presentation after the launch of Rare Disease 101 on 16 February. The slides will be sent out as part of the GA-UK mailer next week.
- One of the key priorities of the UK Rare Diseases Framework is to increase awareness of rare diseases among medical professionals. Lucy's project Rare Disease 101 is to educate medics about rare diseases.

Lucy McKay – presentation:

- Thanks to GA-UK for inviting her to talk about RD 101.
- She will first explain her background about how she ended up in the rare disease advocacy setting. Thanks to M4RD sponsors and donors.
- The purpose of RD 101 is to give the medical professionals the opportunity to: understand rare disease during their clinical career; to help them consider rare diseases when the clinical picture doesn't fit the label it's been given, when it doesn't look right, not dismissing something rare that they haven't seen or heard of before; to appreciate the challenges of living with a rare disease, and understand the extra burdens on top of having a rare condition; to benefit from the excellent resources already available, a recent research paper found that only 4% of doctors had ever used a rare disease resource, an easy gain can be made on that; support and be supported by rare disease patient groups can support clinicians and patients, a lot of power if we triangulate this.
- Lucy explains her background. She doesn't have a rare disease. Her brother had a rare disease, but he died before she was born. Her mum funded the patient group MPS Society (Society for Mucopolysaccharide Diseases). She spent her childhood travelling around the world with her mum, very involved with rare disease advocacy.
- Lucy became a doctor and the two worlds collided. She was experiencing medicine with the knowledge of rare disease, with a community's story behind her, going through medical training and seeing how it could be. She started a society to do something about it. She is a second generation rare disease advocate, which has made her really passionate about the cause.
- Over the years, she had been listening and taking in the experiences of people with rare conditions with the knowledge of how medical training and services work. How to improve the patient experience.
- RD 101 is basic medical education in rare disease. With >1000 rare diseases, it won't be possible for doctors to learn about every rare disease. But they can learn the basics and what it is important to do, and skills to have. For example, researching opportunities and access to treatment that might become available later. RD 101 is online and free to medical and other healthcare professionals. It is targeted to those who know little about rare diseases, especially doctors as they are particularly relevant for diagnosis and prescriptions. In the future, it can be used as a model to educate other healthcare professionals.
- They have identified patterns of what people go through, diagnostic tools (tools in place so that doctors can do something once they are aware of rare conditions), genomics (NHS genomics services), patient advocacy groups (main player in a patient's care), basics on how to support patients and education (spread the message about rare disease and know where to go).
- Thanks to GA-UK and other rare disease communities. RD 101 has relied on patient groups like GA-UK and their surveys have been crucial to put the message of patients out there.
- Lucy shows RD 101 website, it is the first education of its type so constructive criticism is helpful. It gives a basic understanding of what it means to have a rare disease. There is an opening message from Lucy to start, then the different lessons e.g. Introduction to rare disease. She plays a video explaining what is a rare disease, followed by some knowledge checks. The knowledge in RD 101 is not new to patient organisations but it would be very new for the people that the course is targeted to.

- What are the learning outcomes? She would like medical professionals to have a trusted place to go for accurate information about rare disease, not the normal place where they usually get their medical education. From there, they would be more likely to go to patient groups or umbrella organisations like Genetic Alliance UK. Doctors can facilitate the patient's journey through healthcare. They are hoping medical professionals rise to the challenge of coordinating their patient's care, taking it on board as part of their profession, making sure that they know their patients' management plan. Hoping doctors prepare for possibilities ahead in terms of diagnostics and treatment, doing research on patient's condition, communicate to colleagues that rare disease is relevant to mainstream medicine, recognise when someone might have an undiagnosed rare disease.
- #ThinkRare is the idea that if you've spent your career, if you've been conditioned to not think about rare disease, that will hinder you. #ThinkAgain – maybe there is something unifying about their list of symptoms, many patients experience that their diagnosis changed when they changed location. #ThinkAgain – what's changed and we didn't know 10 years ago, options for diagnostics and management have changed, it is important to try to have an open mind. Innovation is outstripping education, but innovation without education is just a vanity project, we need to make that innovation accessible to the patients.
- Handover to Jennifer Jones (Researcher at Genetic Alliance UK), who is going to highlight how Genetic Alliance UK and other patient organisations have contributed to the creation of RD 101

Jennifer Jones (Genetic Alliance UK) – presentation:

- There were over 6,000 responses to the survey for the Rare Experience Report we launched in December 2020. She codes frames, the most common code is the lack of awareness of rare diseases – 10% of comments mentioned problems with healthcare profession awareness.
- They were looking for quotes that would speak to healthcare professionals. Also, including some positive views, good cases are happening and can serve as a model. She went through RD101 and worked together with Lucy. Primary care (GP) was the second most common issue raised by patients.

Lucy McKay – presentation:

- Example of RD 101 slide explaining common challenges of people living with a rare disease. Seeing multiple specialists that don't talk to each other and disagree – poorly coordinated care. A lack of effective treatment or access to treatment. Having to repeat yourself, start from the beginning for a long period of time, a burden of explanation, which changes the dynamic between patients and doctors. Medical professionals have less information than the patient and scepticism about rare diseases within the healthcare system.
- She shows a quote mentioning the lack of interaction between specialists, getting conflicting advice.
- Jen contributes – some quotes were great at creating a picture of their experiences, there is a commonality of experiences despite the uniqueness of rare diseases.

- There is a burden of explanation as patients have to explain their condition over and over again to healthcare professionals, frustrating to patients when they're looking for answers. Especially when many patients have had negative experiences when talking about their condition. Quote saying that healthcare professionals looked completely blank so they didn't continue looking for answers – people are not getting the care they need because of the lack of information in the healthcare system.
- Another quote – healthcare professionals should listen to patients with rare disease, take them seriously and treat them with respect. They don't feel safe when a care provider doesn't have knowledge of their rare condition. "You look well" – denying the lived experience, challenge of invisibility due to invisible condition or because the condition doesn't or because can't have voice heard.
- Slides include pop-ups with quotes so that medical professionals can hear from patients.

Discussion:

- HSP participant: He doesn't understand why patients go through so many consultants.
- Lucy: The problem is that rare diseases present at different times, symptoms will appear over time, which can be similar to a common condition. There must be an understanding that time will be the teller, and that is when you need to act, when there is a mounting picture that the patient's experience doesn't fit a common condition. A softer finding might put it all together, as it can suggest there is something else going on. There is a list of 'red flags' – softer signposting that there is something going on here: the parent has been told to have parental anxiety but now the child has multi-system problems, there is multi-specialist involvement, family history, multiple visits.
- Unique participant: Great presentation, important to get this message out. She has been teaching medical students at UCL for 15-20 years and people coming back and telling her that they haven't forgotten.
- Lucy: The hard thing is to get people to look at it. This information can be put in places where doctors usually get their information from. When do you approach people? Medical students and early training – if we can talk to them, we can get peer-to-peer education so there is some organic spread. And also GPs, their training is shorter. Doubled the entrance to student voice price. CPD is crucial, but higher up in the to-do list.
- Duchenne group participant: Thank you. Previously she tried to educate medical students about neuromuscular conditions. Are you going to link up?
- Lucy: RD101 is not the last place to go, it's just the beginning. From there, people should go to Genetic Alliance UK and then go to a more specialised patient group. It is a gateway to all of the resources that are already there. There is a problem with succession planning in the workforce, it needs to be started early. Why are people so interested in cancer? Because they're really exposed to it, we can demonstrate all the excellent research and progress in rare disease as well.
- Duchenne group participant: Wanted to get there with education, make it more general and then specialise.

- Lucy: When educating about a specific rare condition, it could be useful to also teach the basics about rare disease and signpost to RD101, thus addressing inequity within rare disease. 16 February is the launch of RD101.
- Participant: GPs need to know when to refer, GPs hold the key to unlock to go further. There needs to be some focus within the generic stuff to get that interest going, having focus points.
- Lucy: I agree on the second point and it will come over time. Awareness is great, but what do they need to do with it? What are the options?
- Nick: What is the hashtag so we can help disseminate? #RD101

Nick thanks everyone for attending and closes the meeting.