COMMUNITY CHECK-IN NOTES

Tuesday 12 January:
UK Rare Diseases Framework

Organisations represented:

- Addison's Disease Self-Help Group
- Alpha-1 UK Support Group
- Alstrom Syndrome UK
- ArchAngel MLD Trust
- Ataxia Patients' Charity Wales
- Behcet's UK
- Cure Myotonic Dystrophy UK
- Duchenne Family Support Group
- Fibrous Dysplasia Support Society UK
- Hereditary Spastic Paraplegia Support Group
- Metabolic Support UK
- Nerve Tumours UK
- OcuMel UK
- Pitt Hopkins UK
- The Pituitary Foundation
- PKD Charity
- PSC Support
- Neurological Alliance
- Niemann-Pick UK
- NSPUK
- PIP-UK
- Ring20 Support Group
- Ryburn SMA UK
- SMS Foundation UK
- TOFS
- UKPIPS
- The UK Mastocytosis Support Group
- Unique
- Wolfram Syndrome UK
- World Alliance of Pituitary Organisations

Genetic Alliance UK staff attending:

- Nick Meade, Director of Policy
- Emma Hughes, Engagement and Policy Manager, Wales
- Natalie Frankish – Engagement and Policy Manager, Scotland
- Farhana Ali – Public Affairs Manager
- Sophie Peet, Policy and Public Affairs Officer
- Jennifer Jones, Researcher
- Jan Bochinski, Fundraising Manager
- Izzy Rundle, Engagement and Support Officer

Nick Meade welcomes all to the meeting on the UK Rare Diseases Framework launched on Saturday 9 January. This meeting will be recorded for people that were unable to attend.

Nick Meade – Presentation:

- Introducing Genetic Alliance UK: organisation committed to improve the lives of patients and families affected by genetic, rare and undiagnosed conditions. An alliance of over 200 patient organisations. Rare Diseases UK is a national campaign that built the momentum behind the first policies addressing people with rare conditions, and now makes sure the strategy is implemented and continued. GA-UK also runs SWAN UK, a network supporting families raising children with undiagnosed conditions.

- On Saturday 9 January, the UK Rare Disease Framework was published along with a blog by Lord James Bethell.

- Structure of Framework: four priorities supported by five underpinning themes (see below), and eight principles that aim to ensure crossboard collaboration and maximise the benefits of the framework. The Framework is the first phase, phase two will be about how the Framework is implemented.

- The Framework isn't very long, but there are a few nuggets of commitment which have been picked out in the presentation slides. The first one is positive messages for newborn screening including a reference to the UK being able to examine newborn sequence, references to Genome UK, and also a quote about the need to support patients with non-genetic rare diseases (a gap that has resulted in some people missing out), as well as a measurable commitment to improve.

- Improving the awareness of rare diseases among healthcare professionals – providing education and resources to help them recognise rare conditions, additional genomic training so clinicians can recommend appropriate tests, and references to how important it is for healthcare professionals to be conscious of rare diseases.

- Better coordination of care – for care to be effectively managed, and for patients to experience better coordination of care with a commitment to improvement.

- Commitment to improving access to rare disease treatments and to international collaboration, which is vital for the future of progress in rare conditions, and to improve the pathway to treatment.

- The underpinning themes include: patient voice is at the heart of decision-making and the commitment to address inequality in patient voice; national and international collaboration – recognition that is essential to support patient care, continuing collaboration with rare disease

people around the world; pioneering research – list of activities going on in the UK and their aims; digital, data and technology – make use of tech benefits and for registration services to continue to play a key role in rare disease policy.

- A few comments – The Framework has been built to follow the national conversation on rare conditions, picking up a lot of different themes and priorities. The approach of taking the top 4 priorities and using them to build the Framework was justified to improve focus while other priorities may be covered somewhere else. But it's possible that important points have been missed, inevitably there are gaps, and our community is encouraged to point them out in the discussion. With the previous focus on genomic services, it was thought that diagnosis would come a long way, but there has been little progress. There is also a lack of reference to the journey of someone living with rare conditions.

- Principles – Commitment to regularly review commitments every one to two years and a close collaboration with our community.

- Next steps – Phase one, the Framework has been published. Phase two, to develop an action plan, reviewed every one to two years. So far, there is no commitment, so Rare Disease Uk will focus on putting pressure on the 4 nations to deliver action plans.

- Action plans – It is important that each nation creates action plans that are specific and measurable, reviewing them every one to two years, the more comprehensive, the fewer gaps. We will publish a report with must-haves and desirables for the action plans for the four nations. We want to hear from you about research, report, findings, etc., any gaps in framework to put in place all the evidence so that the nations can write action plans easily.

- Challenges to get actions plans in the UK. England: stretched by covid-19, no decision on the implementation plans, no deadline or commitment yet.

Emma Hughes – Presentation on Wales:

- First published a delivery plan in Wales for mental health, stroke, etc. which is being reviewed – first phase. Second phase: how delivery plans will be, rare diseases will be more difficult due to different conditions, but plan to produce quality statements in high-level documents.

- Commitment to develop action plans for rare disease, reviewing every two years, although unsure if they'll stick to that timeframe. Next meeting in February to update about quality statements. Discussion on Monday about the Welsh cross-party group with Dr Graham Shortland OBE, welcome for anyone to come along.

- First time around, no funding received for rare disease – big gap, how to address this to make a commitment. Email in chat for anyone that wants the link to join.

Natalie Frankish – Presentation on Scotland:

- Commitment to implement the framework. Change of minister – Mairi Gougeon. No formal commitment to produce a Sottish action plan, but positive signals and there will be one by the sound of things.

- Challenge – no funding attached. There is a cross party group within Scottish parliament – to produce a report. Challenges include: measuring success of plan, no mechanism to evaluate what or how well was happening

Nick Meade – Northern Ireland and end of presentation:

- No news, GA-UK doesn't have as many contacts in Northern Ireland. If anyone in the group has any, would be grateful to hear. Call for contributions: policy@geneticalliance.org.uk email to share policy suggestions/comments. Thank you.

Discussion – on specific nations, comments/gaps seen from what has been described in the Framework:

- Actions should be measurable to identify if progress is being made. How do we actually identify people with rare conditions in the UK? No comprehensive registry, there are registration services and NHS hospital records but they don't really mark who has a rare condition. How can you measure if diagnosis has been increased or coordination has been improved without looking at specific pockets? And how do we ensure that the patient voice is truly represented across all the rare conditions? How do we get everyone involved in the conversation instead of just the regular few patient organisations?

- We need to know the incidence of the rare disease, we need to know what we're measuring against. Action plan will depend on what happens in Scottish and Welsh elections coming up in March.

- Nick – definitely will be campaigning the need for action plans in Scotland and Wales.

- Participant – welcome the part about genomics and relationship between research and newborn screening. From discussion with scientists, we know that newborn screening is more accurate, but we are not to forget to continue the advocacy role. Genome sequencing won't be completely replaced – just flagging this.

- Nick – very good points, we shouldn't be distracted by shiny things far into the future.

- Participant – patient, going through various departments within the NHS, and also working for the NHS. Link with NHS so that rare disorders are included in records. Is there a way to work with the NHS to make the journey easy? It would help clinicians to make decisions and when looking at ideas for better treatment.

- Participant – an open approach for those implementing the new Framework for more patient voices. In the past 7 years, we've depended on a small number of patient representatives.

- Participant – who is doing online surveys? we need to make sure to include disadvantaged communities, welcome the collection of a list of patients.

- Participant – if more voices are included, that would be good to press the points you're making from a broader base.

- Nick – welcome new ideas, many of you have worked with us in specific topics. If you think a topic is important for your community e.g. flaws, information, etc. let us know to collect these issues into the report.

- Participant – campaigning for newborn screening for many years now, it is a shame that little progress has been made taking into account there are treatments to improve outcomes. Coordination of care is so important, helping families contact professionals.
- Nick – thanks, will include Concord into reports.
- Participant – the reference to international collaboration is important to address every rare disease. European Reference Network - powerful force but many people don't know about it.
- Nick – useful points in the chat, this input will be considered.
- Participant – what do people think about whether new points need to be included in the action plan even if they weren't in the framework? How do we try to include a wider range of patient voices?
- Nick – trying to get things missed into the action plans. Neurological Alliance has recently published a report on changes in policy they'd like to see.

Nick Meade closed the meeting, thanking all for coming.