ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

on Rare, Genetic and Undiagnosed Conditions

Care coordination

Tuesday 16 May, 12:30-14:00

Parliamentarians in attendance: Liz Twist MP (Chair)

Christina Rees MP

The APPG held a meeting to discuss the report, Coordinating Care: learning from the experiences of people living with rare conditions published by Genetic Alliance UK. The meeting had attendees join in-person and via zoom.

MINUTES

Liz Twist MP, Chair of the APPG for Rare, Genetic and Undiagnosed Conditions

Liz Twist MP welcomed attendees and opened the meeting. Liz noted the launch of the Coordinating Care report at the Rare Disease Day reception that she hosted in February.

Coordinated care is working together across multiple aspects of care to help everyone involved in a patient's care to avoid repetition and achieve shared goals. It ensures the highest standard of care for a patient throughout their life by focussing on the needs of the person.

Many rare conditions are lifelong and complex. As a result, people affected by rare conditions often need support and expertise from a wide range of healthcare professionals.

Effective coordination of care is essential to help people living with rare conditions and their families minimise the impact of appointments on their busy lives and help healthcare professionals to work together effectively to provide high-quality and joined-up care.

Often care coordination is left to the individual and their family. It can add further stress, taking up time and energy while trying to manage a rare condition. Where care coordination is provided, it has shown to have a positive impact.

Louise Fish, Chief Executive, Genetic Alliance UK

Louise Fish gave an overview of the findings of the Coordinating Care report. Many rare conditions are lifelong and complex. As a result, people affected by rare conditions often need support and expertise from a wide range of healthcare professionals such as GPs, specialist hospital consultants, specialist nurses, physiotherapists, occupational therapists, speech and language therapists, and learning disability nurses. This can mean having multiple appointments across different settings and on different dates.

There are various factors that need to be managed between these appointments, and the burden and challenges of so many appointments and interactions with the health service affect people with rare conditions in many ways. We wanted this report to draw these challenges out, so that we can begin to address them.

Our prior knowledge about care coordination comes partly from our work on the CONCORD study, which examined how to describe coordinated care in the health service, and showed that the majority of people living with rare conditions do not have coordinated care.

We brought together case studies of how people are addressing care coordination challenges now, and we also have case studies of people who are coping without care coordination and how their lives are affected.

The report identifies five common themes where care coordination could help to improve their care experience:

- Coordinating complex medical care
- Delivering well-organised logistical support
- Assisting an effective transition from children's services to adult services
- Bridging the gap between healthcare and other services, such as education and benefits
- Integrating support from rare condition charities and support groups

So this report demonstrates that effective coordination of care is essential to help people living with rare conditions and their families minimise the impact of these appointments on their busy lives and help healthcare professionals to work together effectively to provide high-quality and joined-up care.
We hope that this report will help the NHS and governments across the UK to learn from what is already working well.

Sophie Kennginham, Mother to Teddy who lives with a rare chromosomal condition

Sophie Kenningham shared her experience of a lack of support coordinating care for her son Teddy who has a microdeletion on chromosome 4Q. Teddy has low muscle tone, he is

short in stature. He has poor coordination. He has a speech motor planning disorder and very poor speech. He also has an autism diagnosis.

To address the condition, Teddy has neuro developmental physiotherapy twice a week and speech therapy once a week. He is under endocrinology, neuro disability, ENT and orthopaedics at Great Ormond Street. He has a squint, wears glasses and is seen at Moorfields Eye Hospital.

Sophie shared the negligent delivery of the diagnosis to her family when Teddy was 14 months old. Following diagnosis, they were given no information or next steps for their son's care. Teddy's care was passed over to the community paediatrician who has failed to deliver on referral requests made by Sophie, she has had to find backwards routes to accessing the right teams.

"This highlights the need for a clinical lead for people with a rare disease. I should not be having to take this unorthodox route to getting Teddy seen by the appropriate medical people."

Sophie explained the challenges to accessing the right care. For example, healthcare professionals have been unaware of the necessary treatment and care needed to address Teddy's condition. In some circumstances this has been very dangerous to Teddy's health. She has frequently had to turn to legal solutions to secure the right care and even then, it is not being delivered.

"The impact of the lack of care coordination has had a huge toll on my mental health, my family's emotional well-being and also our family finances. It shouldn't be that a family are so stretched and stressed, trying to get a child with a rare disease access to the right therapy and medical care that they end up having a tribunal and needing an advocate to help them, it shouldn't be that as I am now in charge of Teddy's care, the exchanges with the services need to be forceful and strong in order that we get an appointment. There will be numerous families in the same situation as us who aren't able to navigate this."

Liz Twist MP

Liz Twist commented on the shocking experience that Sophie shared, being left to her own devices. She also noted that the APPG's work on alert cards may be able to help with some of Sophie's challenges around health professionals being aware of the rare condition.

Bryony Bery, Mother to Delphi who lives with tuberous sclerosis complex

Bryony shared the journey that her family had been on to receive a diagnosis of tuberous sclerosis complex and associated neuropsychiatric disorders and subsequent care for her daughter Delphi during the Covid-19 pandemic.

Delphi is a very determined and creative girl who has given herself the very apt nickname ‘Dynamo’. At seven years old, she experienced a cluster of seizures on the way to A&E. “I thought she was dying”. It was at this point that Delphi received a diagnosis.

TSC is a rare genetic disorder which can impact just about every part of the body. Currently for Delphi the main manifestations of the condition are her epilepsy, autistic spectrum disorder, and severe anxiety, but this list is not exhaustive and could change at any moment.

It is exhausting for the whole family - it affects everyone. Notably, Delphi’s sister really struggled and began to shut down after the diagnosis. Bryony explained the challenges of negotiating a new diagnosis of a rare condition which included complex care, ongoing adjustments to treatments and changes in behaviour in Delphi alongside the Covid-19 pandemic.

Bryony shared that medical care for Delphi is excellently organised between hospitals. Health professionals work to align appointments and access the best care. For example, Delphi is sent to a separate hospital for MRI scans due to the facilities there which provide a calming space and help her through the process - the health professional consider the behavioural challenges which come with TSC when caring for other parts of the condition. Care coordination currently relies on the dedication and wider thinking of individual health professionals.

However, Bryony finds coordination outside of the NHS extremely difficult. Delphi recently received an ASD diagnosis. It is very difficult to get support through the school because Delphi is excellent at masking during the day but leaves her exhausted and deregulated when she is at home. Prior to diagnosis she was rejected twice by Child and Adolescent Mental Health Services.

The biggest challenge for Bryony and her family is to build connections with health professionals, local authorities and education to help them to understand Delphi’s condition so that they can address it.

Bryony noted the support she has received from several charities including the Tuberous Sclerosis Association, Hope Epilepsy, Wandsworth Autism Advisory Service, Wandsworth Information, Advice & Support Service and Contact.

The next area of concern for Bryony and her family is the transition from paediatric to adult care. Bryony fears that they will lose the great support and specialist medical care that they currently receive if the handover is not done correctly.

Dr Sam Amin, Consultant Paediatric Neurologist, University Hospitals Bristol

Dr Amin is sharing his work as part of the Tuberous Sclerosis Complex Clinics Network. Dr Amin notes that 1 in 45 000 people are reported to be affected by TSC, this figure is likely underestimating the prevalence due to the number of people who are left undiagnosed.

TSC requires a multidisciplinary team of at least 20 health specialists, it is a very complex condition. The combination of a complex condition requiring complex care means that things can easily go wrong.

A forum formed through the British Paediatric Neurology Association provided the basis for a group to encourage professionals in the UK to specialise in treating these conditions, including TSC. This facilitates the discussion of complex cases, dissemination of guidelines, development of research proposals and the planning of care audits.

Dr Amin notes that this forum would work most effectively where there are properly funded designated clinics. The current funding arrangement for TSC care creates inequality based on what individual clinics can obtain funding for. A properly funded TSC service for the whole of the UK would deliver equitable high quality care, including the recruitment of epilepsy nurses, TS nurses and care coordinators. All the centres would be able to address all aspects of the condition and provide the service from a central location.

Liz Twist MP asked if this work was replicated anywhere else. There are other interest groups for other rare conditions which require paediatric neurological input.

Discussion

Portia Thorman, SMA UK, shared the challenges she faced with her son who has spinal muscular atrophy type 1. They have to go to London for various appointments which can be very difficult as her son requires a lot of equipment to travel including a ventilator. It would be extremely helpful if these appointments could be coordinated under one trip. One nurse has initiated a Whatsapp group which helps to coordinate patient appointments.

Portia also related to both Bryony and Sophie's comments around the need to educate local medical and other services. The GP for Portia's son has previously organised a 'team around the child' meeting where local services come together to discuss his care. However, Portia finds that she is left telling the group about what she has done, with little help to her son.

Chris Bedford Gay, FOP Friends, noted the challenges of information sharing between hospitals and encouraging health professionals to speak to one another. One of the people

in the FOP community went to A&E. The patient noted that neither the specialist centre nor the charity were contacted for support.

Chris also shared that Mike Penning MP had secured a three hour debate on FOP on 22 June. For more information contact chris@fopfriends.com

Bryony's comment that care was often dependent on friendly motivated clinicians resonated with Chris.

Louise Fish raised the highly specialised services provided through NHS England. There are currently 85 services for 500 people across the UK addressing rare conditions. Some of these services are NHS funded but they are often supported and bolstered via charities. An example of these services is given in the Coordinating Care report. There is a clinic provided for people with Alstrom syndrome. Most of the funding is provided by Alstrom Syndrome UK who also provide a care coordinator.

Louise also mentioned that the TSC Clinics Network has recently become a Rare Disease Collaborative Networks which are a NHS England initiative that identifies clinic networks that are emerging from the bottom-up. The NHS recognise the expertise in these networks so that other health professionals can turn to them for advice and support. However, this involves even more work for those dedicated health professionals.

The SWAN Clinic pilot in Cardiff serves people with conditions so rare that they currently haven't been identified. They are proving to be an excellent example of one centre providing a holistic approach to care. The DHSC is currently working on securing funding for a similar model. This is necessary for all SWAN families.

Liz Twist MP notes that the UK Rare Diseases Framework sets out aspirations but we need to make sure we have actions that are being delivered.

Liz Martin, William Syndrome Foundation and Pooja Takhar, Tuberous Sclerosis Association, notes that with the advances of medicine, there is a new group of people whose life expectancy is extending. This poses problems for their parents who have coordinated their care throughout their lives and fear who will do this after they pass away.

Steve White, Cure Usher, raised the importance of demonstrating the way that care coordination can save money for the NHS.

PROPOSED ACTIONS

The APPG have identified and will be working taking forward actions linked to several of the themes raised in this meeting.

The lack of awareness of rare conditions in Child and Adolescent Mental Health Services (CAMHS) was raised as a challenge for several rare condition communities. The APPG will engage with CAMHS to raise awareness of rare conditions in the service and make sure that their assessment processes sufficiently consider the nuances that follow rare conditions.

The APPG will also engage with the Integrated Care Systems on the above issue who are responsible for delivering the service. The APPG will engage with the Integrated Care Systems to raise the issue of connection between the education services and health care and the importance of such a connection for people with rare conditions.

It is noted that the responses to the call for evidence as part of the Mental Health and Wellbeing Plan will now be integrated into the Major Conditions Strategy which will replace the Plan. It is important that the responses submitted to the call for evidence regarding the Mental Health and Wellbeing Plan will be appropriately considered and incorporated into the new strategy. The APPG will submit questions asking for clarity on how these responses will be used and how the new strategy will cater for rare conditions.

The APPG will continue with its work to raise the importance of NHS certified medical alert cards for rare conditions in Parliament. The APPG will follow up with the Minister for Social Care, Helen Whately MP regarding her response to written question 183537. The APPG will also request a meeting with the Minister to discuss the issue further.

The APPG will continue to plan a meeting to raise the issue of alert cards towards the end of this year in which an update on these actions can be given. The APPG will invite relevant stakeholders to join the meeting including a representative form the Care and Respond project; the patient passport that has been developed in Wales.

ZOOM CHAT

Karen Whitehead MBE: Hi - Karen here, on behalf of our Pregnancy Associated Osteoporosis (PAO) Rare Disease Patient Group - my story is here https://www.raredisease.org.uk/rduk-news/karens-story/ where I've blogged about my issues and difficulties with Care co-ordination.

Eve Smith: Hi I'm Eve I was the first Gloucestershire ref for SWAN in 1998.. in 1996 my son gained a dx of ATRX syndrome. He is now 26 and an inpatient MSU. He has been totally misunderstood all of his life. He has asd and adhd dx also.

Mike Cain: Louise raised an interesting point. People with Rare Disease need to meet and if clinic can be timed to match then so much the better. Ellen Whatlet MP has written a letter which I have seen that all rare Neurological conditions will be treated in Neurological Centres. These by no means can know the requirements of ppl with rare conditions.

Specialist clinics exist and it is logical that patients are directed there esp if the clinic is doing research so care can be kept updated and share with LOCAL care cord wo can ensure services are provided

Katie Woolsey: absolutely agree Sophie, we're hitting the same type of barriers accessing care for our 4 year old daughter, Megan. sending a hug !

Samantha Barber: Thanks so much Sophie for a powerful presentation.

Karen Harrison: Thank you for sharing your journey Sophie, and so sorry to hear the battles you continue to have

Kym Winter: Thank you for sharing your experiences so openly Sophie - including how the challenges you encountered very understandably impacted on you are your husbands mental wellbeing.

Polly Moyer : Thank you, Sophie - my heart goes out to you.

Claire Andersen : Thank you Sophie for such a powerful and moving testimony, which unfortunately reflects the experiences of far too many families.

Jill Harkin : Thank you Sophie for sharing this personal story and the challenges you've faced. I'm sorry for your ongoing struggles. I wonder if there's an opportunity here to pilot a case manager/care coordinator role that's funded by a shared resource? Like a MacMillan cancer nurse type model for rare disease care coordination?

Karen Whitehead MBE :I have gone to the rare Bone Disease service at Addenbrooke's hospital in Cambridge and helped them set up this service, as a rare patient representative. Rare disease patients were disrupting their normal clinics and services and requiring more time, appointments and support. Now they have a Rare Bone Disease Nurse co-ordinating the Care of Rare Bone Disease Patients and they give us unlimited clinical time with a specialist and have a specialist rare bone disease helpline can call anytime with queries, etc. We have a Whatsapp group of rare bone disease patients at Addenbrooke's hospital with my rare bone disease, PAO and we do meet up regularly, separately from the hospital, we patients all support each other eg. one PAO Mum is speaking publicly about her rare bone disease this Sunday at her baby's baptism and we are all going to support her & cheer her on.

Claire Andersen : Seeing so many different professionals, who may have quite different approaches to the same medical concern(s), and having to go back to the beginning and repeat medical history etc. each time adds to the stress experienced by families.

Eve Smith: Yes I have been my sons carer since 1996 it's been a struggle and traumatic not knowing how ATRX will evolve. It's multiple bodily issues of concern. Holland is the only country working on this leaving Uk far far behind.

Eve Smith: ATRX is very rare and debilitating and my son has suffered all of his life. I contacted NHSE for help but nothing was offered. Why are LAs locking away ppl like my son who need extra experience, support and care. This is a national scandal.

Karen Whitehead MBE: Us PAO Mums are trialling this new Rare Disease Patient Passport via Cambridge Rare Disease Network. I've moved to Somerset and its been a nightmare trying to explain my medical history and about my rare bone disease to new healthcare professionals. But the passport does seem to be helping.

Karen Whitehead MBE : Yes, thank you Sophie & Bryony, it is really brave of you both to share your & your families stories like this....

Polly Moyer: Thank you, Bryony.

Samantha Barber: thank you Bryony for another impactful presentation - what a mix of care you and your daughter have received

Katie Woolsey: Hug to you Bryony :)

Karen Whitehead MBE :Cambridge Rare Disease Network Patient Passport

Rachel Rimmer : Thank you Sophie & Briony for sharing your experiences

Eve Smith: ATRX - there are only around 100 in the uk - not common at all…yet left alone

Karen Whitehead MBE: There is a Welsh Passport too, anybody anywhere in UK can sign up for. This is helpful, because ambulances, etc. can access this. I've signed up for this too... Its here https://www.careandrespond.com/passport/

Karen Whitehead MBE: PAO is apparently ultra-rare, estimated as 1 in 100,000 although I personally think it is underdiagnosed !

Karen Whitehead MBE: Thank you to Dr Amin too, for his support of and interest in rare diseases and his presentation.

Nigel Over: I'm in Scotland so health is devolved but happy to share my experience for learning from. As a teenager my son Matthew (now 25) was at one point supported by 52 professionals. It was a challenge to have to coordinate and attend appointments / consultations. At the appointments I was constantly repeating information. I found that the information and data was retained by individual professionals / supporters. There

needs to be more joined up thinking of information retention and sharing as well as effective coordination, which has always come down to what I can do as a parent.

Katie Woolsey: how can care coordinators become more common place

Eve Smith: That could have helped my son to have 1 clinic for everything- sounds amazing.

Karen Harrison: We (Alex TLC) have been part of setting up the new NHS England Inherited White Matter Disorders Service and Registry, this service consists of 5 centres of including one adult centre with 2 hubs for adults, we will be providing support at the clinics.

Karen Whitehead MBE :I just wanted to query, on mine and the Pregnancy Associated Osteoporosis Mums, we to date have mainly been very well looked after by a few key hospitals and specialists & often travelled long distance (eg. Addenbrooke's in Cambridge, Oxford, RNOH, Glasgow in Scotland, etc.) but now, under the new Rare Disease Framework, it is being suggested we should see new Rare Disease Hospitals in our area of country - but these hospitals and specialists are not knowledgeable about our particular rare bone disease. How many rights as patients do we have to stay at our long standing hospitals? eg. I've been a patient at Addenbrookes and well looked after for over 10 years and they are suggesting under the new Action Plan I have to be seen in Bristol?! Is this necessarily the case? Its not just me, this is just my example for you.

Rachel Rimmer : There is a huge disparity of services available between Paediatric services and Adult Care

Karen Whitehead MBE :I am used to co-ordinating my own care and I just email updates to the Rare Bone Disease Nurse at Addenbrookes who is now coordinating our care. But some of the young Mums have found her co-ordination help really useful. She works like the Macmillan nurses in Oncology. There is a Rare Nurse Network, of nurses who specialise in rare diseases.

Kym Winter: I strongly suspect that rare conditions as a category are poorly understood/recognised at services like CAMHS(which are admittedly hugely stretched). Has there been any contact made at all with national CAMHS Leads to begin establishing rare conditions as a distinct group with particular and unique needs?

Nigel Over:The clinicians with knowledge on SMS are scattered across England. It would be great to have an annual clinic where the professionals can come together but can't see this coming together soon just because of low numbers of families affected. Each rare disease needs to be considered as a Highly Specialised Service due to the geographical spread of families and the expertise that sits with a few dedicated professionals. It cannot

be practical for every health area to have the in depth knowledge to support all genetic disorders within their respective areas.

Rachel Rimmer:I love the question from a GP “Who is your care coordinator?” For most adult patients and parents to reply “ME”

Karen Whitehead MBE: I do think mental health support for patients with rare diseases is lacking. We've just organised a Trauma Workshop for our PAO Mums and it was heartbreaking listening to their stories.

Karen Whitehead MBE: My new GP told me they had never heard of my rare disease and that they did not believe it existed !

Rachel Rimmer: Some centres may have Paediatric Psychologists attached, if they’re “lucky”. But move to adult care and the reliance heavily relies on a patient requesting MH support from their GP who then adds them to the never ending waiting lists

Kym Winte: There is still a lot of work to do in integrating mental health support into RD care - good care co-ordination also playing an important preventative /supprtive function

Amy Hunter: The new Framework Evaluation research funded by NIHR will begin early 2024. Applications still being processed.

Karen Whitehead MBE: It is wonderful there is going to be a new Parliamentary debate like this. I'd like to see patients able to see specialists who really are specialists and knowledgeable in their particular rare disease. ie. not being transferred to a supposed new rare disease service in their part of the country, where the specialists don't know or understand their particular rare disease. There are only a few UK specialists truly expert and knowledgeable about my rare disease.

Karen Whitehead MBE: Is it possible to get an official Rare Disease Patient Passport - at present different charities / groups / areas seem to be DIY'ing but the Government could help standardise & make this more official ?

Katie Woolsey: thank you so much, this has been very interesting and gives me more hope for my daughter's future care x

Rebecca Middleton: Thank you everyone, and thank you for sharing your experiences.

Samantha Barber: Thanks everyone for a great and action-focused meeting.

Jonathan Gibson: Thank you very much

Polly Moyer: Thank you, everyone.

Nigel Over: Thank you for sharing stories and such an interesting discussion.

Karen Whitehead MBE: Thank you for a really interesting and useful meeting and thank you for all your clear hard work on behalf of us rare disease patients.

Kym Winter: Thanks everyone - and to GAUK for all you do