COORDINATING CARE

LEARNING FROM THE EXPERIENCES OF PEOPLE LIVING WITH RARE CONDITIONS

GENETIC ALLIANCE UK

ABOUT GENETIC ALLIANCE UK

Genetic Alliance UK is the largest alliance of organisations supporting people with genetic, rare and undiagnosed conditions in the UK. Our 200+ members and the people they support are at the heart of everything we do.

We advocate for fast and accurate diagnosis, good quality care and access to the best treatments. We actively support progress in research and engage with decision makers and the public about the challenges faced by our community.

We run two long standing projects:

Rare Disease UK, a campaign focused on making sure the new UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment.

SWAN UK (syndromes without a name), the only dedicated support network in the UK for families that have a child or young adult with an undiagnosed genetic condition.

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FOREWORD

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.

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.

In 2023 Genetic Alliance UK is highlighting how well-coordinated care can make a real difference to people's quality of life. We are publishing this report on Rare Disease Day 2023 to increase awareness of best practice among healthcare professionals and help them work together effectively to support people with rare conditions. We also hope that this report will help the NHS and governments across the UK to learn from what is already working well.

Genetic Alliance UK is an alliance of over 200 charities and support groups that advocate for people living with rare, genetic and undiagnosed conditions. We would like to thank the member organisations that put forward case studies for inclusion in this report for the amazing work they do to support people living with rare conditions, Addison's Disease Self-Help Group, Alex TLC, Alstrom Syndrome UK,Cavernoma Alliance UK, CGD Society, Huntington's Disease Association, Max Appeal, Rare Revolution, Tuberous Sclerosis Association and Unique.

We would like to thank the healthcare professionals and individuals who agreed to be interviewed for this report for sharing their stories to help improve care for individuals living with rare conditions and their families.

We would also like to thank our sponsors who have generously supported Rare Disease Day 2023 to help us raise awareness of the challenges facing people living with rare conditions.

Finally, we would like to thank you for taking the time to read this report. Whether you are reading this report in your office, your clinic, your university, your lab, or your home, it is your passion for learning about how to improve the lives of people with rare conditions that brings hope for the future to the individuals and families who are affected by them.

Louise Fish,
Chief Executive Officer, Genetic Alliance UK

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EXECUTIVE SUMMARY

The case studies presented in this report vividly demonstrate the value of care coordination both when they describe the presence of care coordination, and the impact of its absence. The benefits of care coordination are clear for people living with rare conditions and their carers, parents or relatives, for healthcare professionals, but also much more broadly for healthcare budgets, and providers of services outside of the healthcare environment.

The value of care coordination can be seen across the following themes:

Coordinating complex medical care

The efficiency of medical care coordination has direct positive health outcomes. In progressive rare conditions, time spent without optimal treatment has a cumulative effect on the impact of the condition, and efficient medical care coordination reduces this time period. Clinicians in the same room as each other, virtual or physical, can select medicines to treat one aspect of a condition carefully to ensure it does not worsen another symptom without weeks of correspondence.

Medical examinations can be planned to maximise the return of information, and minimise hospital time and resources, and the results can be discussed by a team of clinicians to ensure the best quality interpretation. Healthcare in primary and emergency settings are more effective, local care providers understand their role and information is available as it is needed.

Delivering well-organised logistical support

Families and clinicians both valued the administrative support that coordinated services bring. This role is distinct from the coordination of medical care, and can be delivered by a different member of the team. This role increases efficiency of the service, removes or reduces complexity for families and people, thereby removing a burden and potentially reducing stress.

In its absence, people are 'battling', dealing with primary and emergency care, and filling in information gaps between care providers. This is extremely stressful, damages physical and mental health, takes up a lot of time and reduces the ability for some to work. Opportunities are missed, interventions are delayed and health outcomes are worse.

Assisting an effective move from children's services to adult services

Transition coordination between paediatric and adult services is not an issue for all rare conditions, but where transition does occur, it is vital that it be coordinated effectively. It is not a single event or moment, but a process that takes place over a period of years. Focused oversight and planning are necessary to ensure transition is smooth and all aspects of care and needs are transferred effectively. Transition is between parents having an oversight role in care decisions to individuals advocating for themselves. It is also a change from the multi-system focused paediatrics to the single system specialisation of adult care – in some cases the transition can be to less coordinated care. This process needs to be adapted to the context of the individual's life, where education or extra-curricular activities may be priorities.

Bridging the gap between healthcare and other services

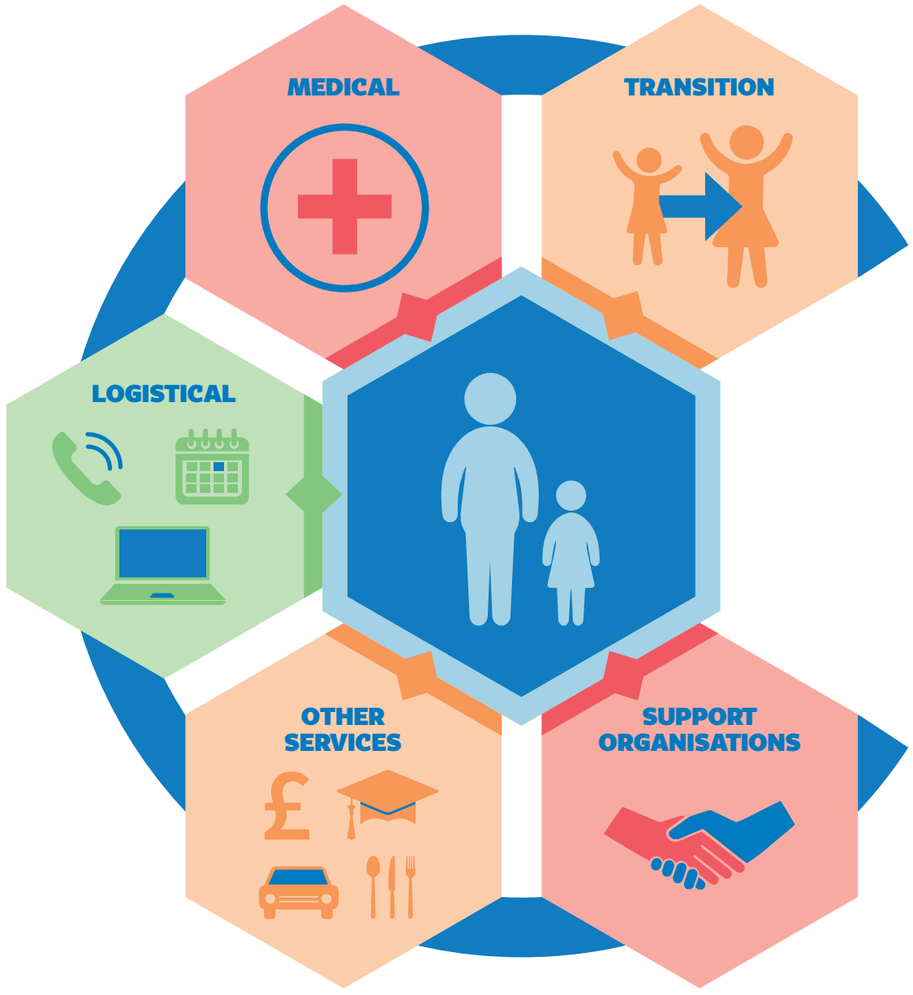
The biggest signal from this cohort of case studies was the value of coordination between health services and other agencies in people's lives. Education and access to social support and benefits were key themes that appeared in the majority of our discussions. Information exchange between healthcare providers and other services validates a person's needs. This support can reduce the burden of form-filling and 'battling' that some families face in accessing appropriate education support. Advice from healthcare professionals on how to manage one's condition at school, college or university can help people living with rare conditions to thrive as students. Clear information describing what benefits and support services are available and how to apply for them would have been valuable to many of the people sharing their experiences in this report.

Integrating support from rare condition charities and support groups

The key role rare condition support organisations can play in maximising the potential of care coordination was shown in services for Alström syndrome and for tuberous sclerosis complex. The convening role, expertise in the experiences of people living with the conditions, as well as two-way dialogue with the community are key to the design of an effective coordinated service. The role of support organisations also came through in the absence of care coordination where the information provided by the voluntary sector became a vital reference point for families navigating their own journey.

Communication is key and improvements are always possible

Even those involved in services with the most secure long-term funding felt that there were improvements to be made. The scope can always be broader and the process more efficient, dialogue with people receiving coordinated care is important to drive this progress. Communication most broadly appears to be the thread that brings this complex area together. Whether it is the timing of a clinic, sharing test results, accessing health records, exchanging care responsibilities between clinicians, deliberating over optimum treatment, planning a weekend of tests, describing educational support needs or supporting a grant application, good communication is the fundamental basis for good coordination of care.



WHAT IS CARE COORDINATION?

Genetic Alliance UK's work on care coordination has primarily been in the context of the CONCORD (CoOrdinated Care Of Rare Diseases) study, so it is appropriate to start by answering the question – What is care coordination? – using CONCORD's answer:

Co-ordination of care involves working together across multiple components and processes of care to enable everyone involved in a patient's care (including a team of health care professionals, the patient and/or carer and their family) to avoid duplication and achieve shared outcomes, throughout a person's whole life, across all parts of the health and care system, including: care from different health care services... care from different health care settings... care across multiple conditions or single conditions that affect multiple parts of the body, the movement from one service, or setting to another. Co-ordination of care should be family-centred, holistic (including a patient's medical, psychosocial, educational and vocational needs), evidence-based, with equal access to co-ordinated care irrespective of diagnosis, patient circumstances and geographical location.

Reproduced from Walton H, Hudson E, Simpson A, Ramsay AIG, Kai J, Morris S, et al.

The CONCORD study

The experiences of the people featured in this report bring to life many aspects of the multi-faceted CONCORD project findings: about care coordination in the UK currently, about preferences of people living with rare conditions, and around how care could be coordinated taking into account personal circumstances and available services.

Good practice for care coordination exists in the UK, but is not widespread. A survey for the CONCORD study found that while about a third of people responding (~32%) had access to a specialist centre, only 10-12% had a care coordinator. Just 10% of affected adults taking the survey said they have a care plan in place. Parents with children with rare conditions are much more likely to have a care plan for them, but still fewer than half of our respondents reported this (44%).

The impacts of good versus poor coordination are discussed by the people featured in this report and they reflect the findings from interviews carried out for CONCORD. Interviewees shared experiences of uncoordinated care such as poorly organised appointments, ineffective communication between key professionals, and a lack of flexibility to allow coordination to meet people's changing needs. In many cases people reported having to coordinate their own care. Consequently, there can be barriers or delays to accessing the care people need and negative effects on physical health. Substantial impacts in other areas of life were reported, such as loss of earnings, disruption to schooling and work, and an emotional burden which can affect mental health.

Through in depth and structured consultation with healthcare professionals and people living with rare conditions and their families and carers, CONCORD found that effective coordination will inevitably involve a jigsaw of elements and considerations, and the reality of what is in place across the UK is mixed:

- Organisation of clinics and services can be through national centres, or local services, or a combination.
- Relevant healthcare professionals may be working effectively in fully-developed multi-disciplinary teams (MDTs), or at intermediate levels of collaboration, or with little collaboration.
- There may or may not be a point of contact, such as a care coordinator, who has an administrative and/or support role.
- Appointments might be organised on a regular schedule, only on-demand or both.
- Access to health records by both clinicians and patients varies greatly, from limited to full access.
- Clinicians use a variety of means to communicate with each other and with patients, which can be efficient or slow.

CONCORD interviewees, like the people featured in this report, highlighted the positive difference that can be made when services take account of these elements of coordination. For example, care coordinators can smooth the organisation of complex care, bridge the gap between healthcare and other services, and offer reassurance by offering a consistent point of contact. Being able to see several healthcare professionals in one location on one day reduces disruption to everyday life. Conversely, the advantages of well-coordinated local provision such as less travel and familiar surroundings can be welcome. Good communication was stressed by many, facilitated by, for example, 'Team Around the Child' meetings and agreed care plans (both long-term and for emergency situations). The potential for electronic/computer systems to drastically improve the effectiveness of communication between healthcare professionals and with affected individuals is recognised, but balanced by a frustration with current implementation.

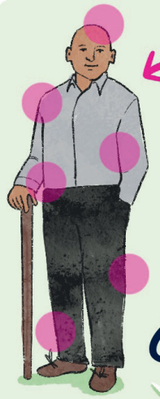
CONCORD outlined a series of 'models' of care coordination, designed for a wide variety of circumstances, but to support implementation of these approaches across the NHS there is a need for a thorough assessment of their benefits in relation to costs.

A short animation summarising the CONCORD findings was released for Rare Disease Day 2023. It can be found along with the scientific papers published to date and a recording of the final project webinar at geneticalliance.org.uk/CONCORD.

This project is funded by the National Institute for Health Research Health Services and Delivery Research Programme (project number 16/116/82). The views expressed are those of the authors and not necessarily those of the NHS, the NIHR, or the Department of Health and Social Care.

CONCORD STUDY

Coordinated Care of Rare Diseases



6-7000 UNIQUE RARE CONDITIONS

AFFECTING MULTIPLE SYSTEMS OF THE BODY

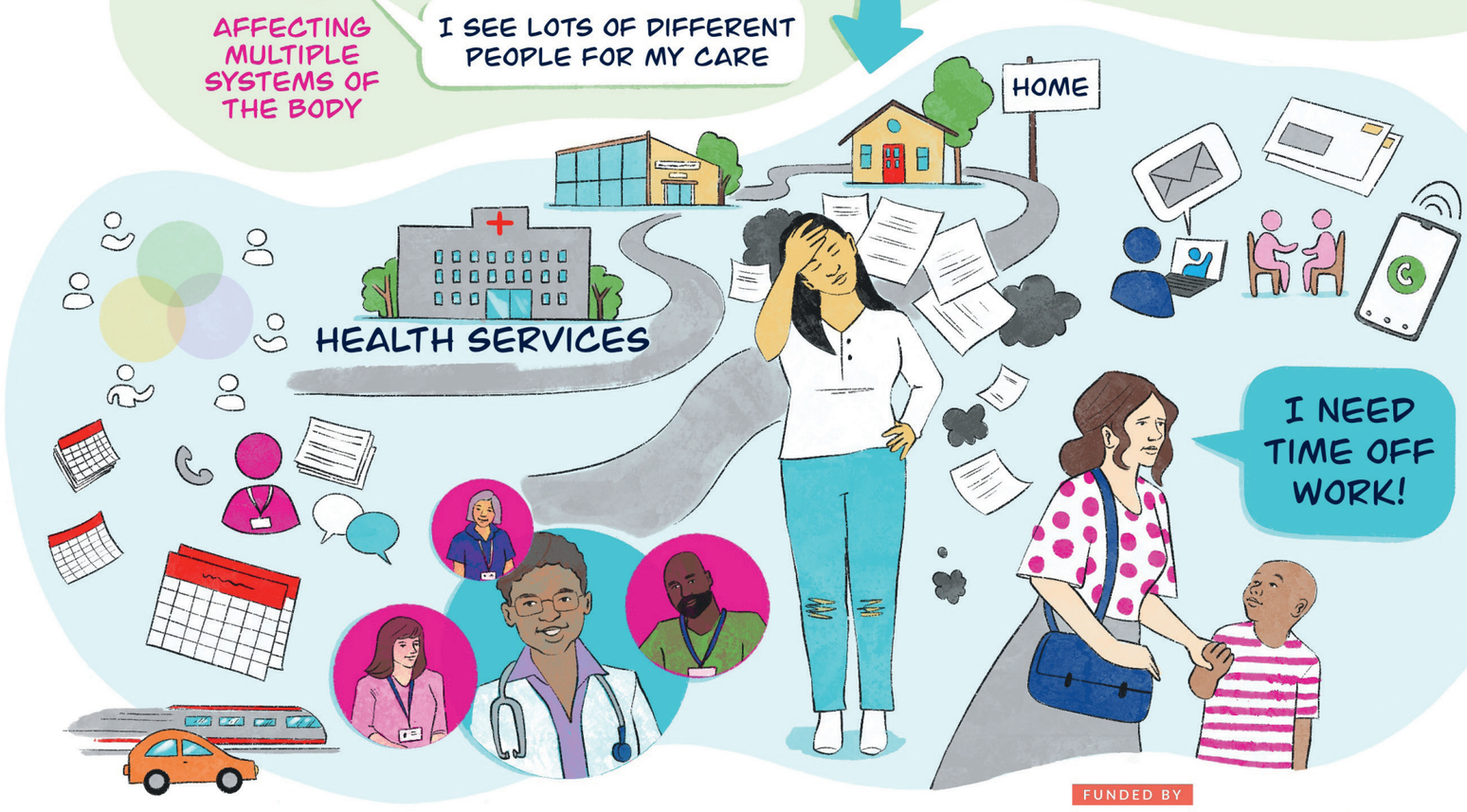
I SEE LOTS OF DIFFERENT PEOPLE FOR MY CARE

LACK OF **CARE COORDINATION**

3.5 MILLION PEOPLE

HOW?

THIS NEEDS TO BE **IMPROVED**



GENETICALLIANCE.ORG.UK/CONCORD

FUNDED BY NIHR | National Institute for Health Research

1 WHAT DOES 'COORDINATED CARE' MEAN?

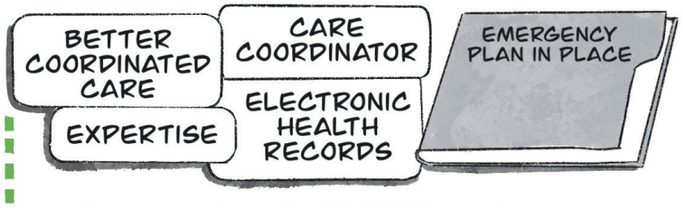
WORKING TOGETHER TO ACHIEVE SHARED OUTCOMES

2 IS CARE FOR PEOPLE WITH RARE CONDITIONS IN THE UK COORDINATED AND, IF SO, HOW?

ACCESSING CARE

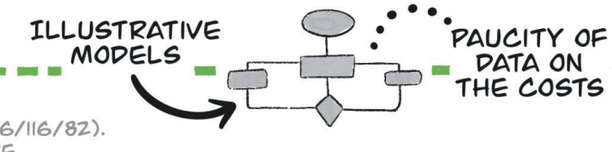
	HAVE A CARE COORDINATOR	ACCESS TO A SPECIALIST CENTRE	HAVE A CARE PLAN
% OF PATIENTS	12	32	10
% OF PARENTS/CARERS	14	33	44

3 WHAT ARE THE PREFERENCES OF PATIENTS, FAMILIES AND HEALTHCARE PROFESSIONALS IN RELATION TO HOW CARE IS COORDINATED?



4 WHAT ARE THE DIFFERENT WAYS?

5 HOW COULD CARE BE BETTER COORDINATED?



THIS PROJECT IS FUNDED BY THE NATIONAL INSTITUTE FOR HEALTH RESEARCH HEALTH SERVICES AND DELIVERY RESEARCH PROGRAMME (PROJECT NUMBER 16/116/82). THE VIEWS EXPRESSED ARE THOSE OF THE AUTHORS AND NOT NECESSARILY THOSE OF THE NHS, THE NIHR, OR THE DEPARTMENT OF HEALTH AND SOCIAL CARE.

KEZ AND HASSAN:
ALSTROM SYNDROME HIGHLY SPECIALISED MULTIDISCIPLINARY CLINICS

Alstrom syndrome is an ultra rare condition. It is a life limiting and multisystemic condition, affecting multiple parts of the body and more complications arise as a patient gets older. Most people experience multiple sensory deficits, i.e. vision problems, hearing difficulties, a poor sense of smell and poor control of bodily functions.

There is a highly specialised service for treating Alstrom syndrome, with a specialist centre at a hospital in Birmingham. Families attend an annual check up where they see a range of different specialists over a day or two and the healthcare professionals provide care as part of a multidisciplinary team (MDT).

Condition: Alstrom syndrome

A diagnosis unlocks access to coordinated care

We had a very late diagnosis. As Hassan's parents, we identified that things weren't right within the first few months around his development, we approached our midwife who then referred Hassan to an eye specialist to get his sight tested. At about the age of two we were told his vision was bad enough that he could be registered as blind. At age 15 he lost all of his vision and at age 16 he started to have blood glucose issues and difficulty with excess weight. He wasn't diagnosed with Alstrom syndrome until he was 17, which came as a bit of a shock. Initially we were only dealing with a visual impairment, not knowing that it was part of a rare condition. Before the diagnosis, the healthcare system sort of left us to deal with the vision impairment on our own with no other offer of support, but things became different when we actually got a diagnosis and understood what support was available.

For me, care coordination is about individual person centred care, which we did get after Hassan was diagnosed. All of a sudden I was in multidisciplinary team (MDT) meetings with representatives from Alstrom Syndrome UK who were endorsing the challenges and complexity of Hassan's condition. The care and compassion started coming through and that was having a positive impact on all of our wellbeing. That, to me, was light at the end of the tunnel, when people started to listen and were prepared to understand what Hassan needs. There was a feeling that it could have been a lot different if we knew about it earlier so sometimes we feel like we've missed out on 17 years of not knowing what you're dealing with.

How the coordinated service works for families

Since Hassan was diagnosed we have access to a MDT of healthcare professionals at a specialist centre in Birmingham. Annually we'll travel to the hospital for a 'full body MOT' that takes place over two days. It's done in a coordinated way and **it really feels like world class care.** The first day is all about getting the tests done and the second day is where you spend an hour or two with your clinicians going through your results. **Over these two days there are probably about 10-15 different healthcare professionals involved in Hassan's care.**

At the hospital there is a dedicated room for Alstrom families. In that room there is a dedicated care coordinator who manages everyone's appointments throughout the day. There is also staff from Alstrom Syndrome UK and a nurse to support the families. We feel quite special during those days because we don't have to worry about things like buying lunch or breakfast, they're provided to the room and you just go in and out to have your appointments. We stay overnight in a local hotel, which is paid for, where we get to meet up with other Alstrom families. We have these amazing clinicians who have got to know us over the years and we've built a great rapport. Hassan is very mindful that he has to manage this condition, he is always a bit anxious about the results but often for him the results are positive and that has a positive impact on Hassan too.

Coordinating with local services

Local doctors and our GP are also good at liaising with the specialist centre for things in between the annual review and the MDT communicates well with the GP to let them know the results. For example, if a local consultant wanted to send Hassan for a liver biopsy they will liaise with the MDT at the specialist centre to understand what tests he's already had and if there's any information that could be used from those.

Care coordination goes beyond healthcare

As parents you're involved in special educational needs meetings to put educational care plans in place. Before Hassan's diagnosis there was no support when we tried to explain Hassan's needs and we were often faced with many challenges for what he needed. When Hassan was diagnosed he was still in full time education and **his MDT were able to endorse the challenges of Hassan's condition, what his needs were and how those needs could be met.** Things started changing for Hassan. Going from no care package to getting 20 hours a week of care has put such a positive spin on his life: it helps him go to the gym which is really important to help him manage his condition, it also allows him to go out and meet his friends, giving him more of a social life. Hassan is a keen and enthusiastic person and his ambition is to be as independent as possible. Having a coordinated care approach and a MDT enabled Hassan to have this education and care package in place.

Condition: Alstrom syndrome

Location: Birmingham

Prof Timothy Barrett, Leonard Parsons Professor of Paediatrics, University of Birmingham; and Honorary Consultant Paediatric Endocrinology and Diabetes, Birmingham Women's and Children's Hospital.

A three person approach to support coordination

For the Alstrom service, we have a tripartite model between a children's diabetes Specialist Nurse, a Transition Coordinator, and a Family Support Worker from Alstrom Syndrome UK, each carrying a distinct but overlapping role.

The specialist nurse will coordinate the medical aspects of care and also liaise with schools to educate teachers about the medical aspects of the condition and potential safety issues. For example, if a child has diabetes and the community services are really worried about what to do if a child has low blood sugar in school, the specialist nurse can step in and educate teachers to help them feel safe to intervene if needed.

The Transition Coordinator coordinates many things such as supporting individuals moving into senior school or sixth form and moving onto employment afterwards. They help families claim any benefits or personal independence payments (PIP) they are entitled to. They play a key role in understanding what having the condition means to individuals and their families, what individuals understand about their medicines, what they understand about internet safety, what they understand about banking or accessing bank accounts, what they understand about relationships etc. The Transition Coordinator also supports families getting an educational healthcare plan (EHCP) at school. This coordinator role also pulls together the support between the end of children's services and the beginning of adult services, which can be a process that takes a few years and at the point of transfer, there is a face-to-face handover between the paediatric Multi Disciplinary Team (MDT) and the adult MDT. The Transition Coordinator role is particularly important for families where English is not their first language.

We also work in partnership with the patient charity, Alstrom Syndrome UK, who provide a Family Support Worker who coordinates travel and accommodation for the patients and their families to come to the clinic. They also support families to receive specific items of equipment for the home e.g. exercise bikes and they also overlap a little bit with helping families claim benefits. Between clinics they provide emotional support to families, and connect families to each-other through family days and conferences.

Coordination beyond healthcare

The real value of this coordinated approach is that we're able to stretch beyond the healthcare system, particularly reaching into education settings which is very important for Alstrom syndrome, as it's a condition that involves sensory deficits (hearing, vision loss). Schools are sometimes unsure as to the educational needs of children with Alstrom syndrome but the coordinators can step in, accessing the healthcare professional team, to ensure that the condition is explained properly and therefore the child's needs are understood as part of that.

Care coordination also ensures that the healthcare professionals are coordinated themselves. The Transition Coordinator helps facilitate and arrange MDT meetings with each clinic so that everyone sits round and discusses their interaction with a patient and their recommendations. This ensures that information is shared and consistent recommendations are made. **For example, if a child has chronic renal failure and also has headaches, one doctor may want to prescribe a painkiller such as a non-steroidal anti-inflammatory drug like Ibuprofen for the headaches but this isn't suitable due to the renal failure.** By healthcare professionals coming together to discuss their decisions, they can avoid prescribing mistakes that may result in further complications. At these MDT meetings there are about seven to eight healthcare professionals across eight different specialties and we put together a management plan to support the local team care for the patient before the next visit in 12 months time.

Care coordination has evolved over time

This model of coordination facilitates communication between different healthcare professionals, providers and the community has evolved over the years. It was set up in 2008 as a partnership between the children's hospital, adult hospital and the patient support group, which was very good. **When the service started, the main coordination was delivered through the specialist nurse but as time went on they took on more responsibility, particularly with emotional wellbeing and PIP support to families.** In about 2014 we made a case to commissioners for the Transition Coordinator and over time have increased their hours.

This model has been extended to other rare condition services such as Bardet-Biedl and Wolfram syndromes, also delivered at the same centre, but other conditions that I treat don't have the same coordinated service. For example, we have a specialist clinic for Prader-Willi syndrome that consists of me, a specialist nurse and sometimes a spinal surgeon but no coordinator or other specialties are involved. I think the patients really lose out because they have to have separate appointments for psychology or dietetics etc, they don't have dedicated support to help them develop an EHCP and don't have enough resources to send nurses into schools to educate staff about how to deal with the condition. **The majority of the rare conditions I treat do not benefit from care coordination and patients suffer as a result because of delays more than anything else,** in getting things done, in providing the optimum treatment or advice for educational services.

A one-stop-shop model

The Alstrom clinics operate six to seven times a year, delivering a one-stop-shop model where patients get all their tests and assessments done over a two day period. **The MDT service really is a gold standard.** There is still a lot of room for improvement, in an ideal world every person would have a named care coordinator they could contact, however with our three person approach it does mean there is almost always someone available. We'd also like to add other services into our MDTs but this does have to be balanced with how much you can fit into a single visit and not over-tiring families. We'd also like to include local healthcare providers in our MDT meetings via Zoom to have a truly joined up service with the local support network. Unfortunately communication between the MDT and local services relies on posting letters, perhaps a future answer would be for patients to be able to hold their own records through the NHS app that they could share when being transferred between local providers.

Another thing we'd like to improve on is our home health monitoring, such as posting blood testing kits for families to do at home. We are currently able to offer this to selected patients who are unable to access their hospital or GP practice for a blood test.

Coordinating mental health and wellbeing services

As part of the MDT service we have a clinical psychologist to see patients and their families. Although they can't do anything therapeutic in a one off annual consultation, they can identify issues and signpost to local services which is very helpful. The Transition Coordinator, as part of their initial assessment, also looks at the emotional health and wellbeing of families and is also able to identify issues and signpost to local support. However, problems arise when local support services like CAMHS have an 18 month waiting list because those services are very stretched.

Replicating this service for other rare conditions

I don't think we can replicate this model for every rare condition but having national disease specific centres might be valuable for the ultra-rare or multisystemic conditions where lots of doctors are required. For other rare conditions there may be some benefit in having designated regional rare disease hubs that can look after children or adults with more common rare conditions and have a range of specialties at their disposal e.g the SWAN (Syndromes Without a Name) clinics we have been running in our children's diabetes centre for the last 5 years. This could be a more efficient way forward for the larger numbers of children and adults with rare conditions.

TEDDY AND SOPHIE:
THEIR EXPERIENCE OF UNCOORDINATED CARE

Condition: deletion on chromosome 4q

Diagnosis

Teddy was diagnosed with a chromosome deletion when he was 14 months old. We first noticed that something was different about Teddy when he was not giving a social smile, one of the first developmental milestones for a baby. The GP showed concern but was originally unsure about where to refer us. They were putting in queries with departments but because it was not a standard practice, the process was taking a long time and the doctors did not seem confident. The uncertainty was very unsettling so I decided to take this issue privately. At such a crucial developmental age for Teddy, his doctors not knowing what to do was very worrying.

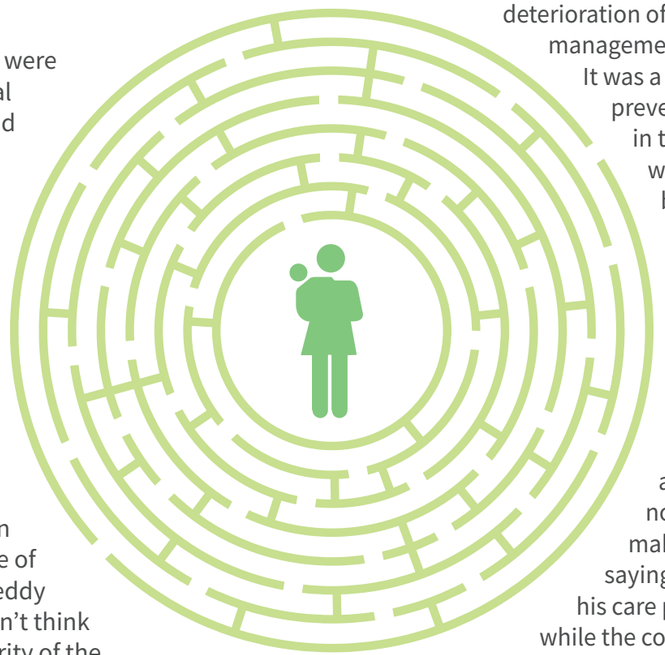
After seeing a developmental consultant privately, we were referred to Great Ormond Street Hospital (GOSH) for genetic testing when he was ten months old. We received his results and an official diagnosis when he was 14 months old. The delivery of diagnosis felt almost negligent; the geneticist was smiling while giving us life-changing news. We were chucked out onto the street without any information on next steps for our son's care. I am also outstounded that we weren't given any emotional support after the delivery of such life-altering news. It left us feeling desperate; like no one knew about Teddy's condition and that, because of the rarity, there was no help available.

We returned to the developmental consultant after receiving the diagnosis but only for a few months as she focussed on infants under 18 months. At this point we were transferred to the care of the community paediatrician but they were failing to make essential referrals. Teddy's needs were changing and so we needed someone who could refer us quickly.

Fighting for the right care

Teddy's condition affects several parts of his body. We see several departments across several hospitals for physiotherapy, speech therapy, respiratory, endocrinology, ENT, neurodisability, eyes, orthopaedics… Access to services are based on evolving needs at this point in Teddy's care and whether we are considered likely candidates for that service.

Very few of these services were accessed in a conventional or straightforward way and pretty much everything has been a battle. Teddy has narrow airways and there were multiple incidents where he had trouble breathing at home. We called ambulances but the response was so poor. I inquired with the GP and the community paediatrician about it with no avail.I even had an out of hours GP accuse me of making up the fact that Teddy had a chest infection. I don't think they understood the severity of the situation for a child with Teddy's condition. I feared that this could be what killed Teddy. By coincidence, I mentioned it to Teddy's endocrinologist at GOSH who helped to set up access to the respiratory department, through a referral by the GP. **Access to essential services such as these shouldn't be left to chance.**



Once accessing the service, it has also been difficult because often the doctors are not experienced in treating someone with this rare condition. In the example of physiotherapy, Teddy has hypermobility and hypotonia. He was not walking at two and a half years old. He was being seen only once a month by junior physiotherapists. It was not enough to address his symptoms and the lack of action was very disconcerting. In response, we again went privately and took Teddy on a three week intensive physiotherapy course. He participated in two of these courses and made great progress. By the end of the second course, he was able to walk. To maintain this progress we returned to the NHS physiotherapist. However, Teddy was placed with a junior physiotherapist in training who prescribed exercises which were damaging to Teddy's condition. This could have been detrimental to Teddy's condition and the complete lack of awareness about the condition was dangerous.

Teddy then presented with a limp and was diagnosed at GOSH with Perthes disease, a deterioration of the bone, which requires management through physiotherapy. It was a time sensitive treatment to prevent the bone from healing in the wrong position. GOSH would be communicating between the orthopaedic and physiotherapy departments and also would need to speak to the community physiotherapist. I continually followed up with the community physiotherapist and after a month there was still no progress on getting an appointment because they had not had contact with GOSH. To make matters worse, GOSH were saying that Teddy needed to finish his care plan to prevent regression while the community physios were seeing improvement and so wanted to stop his care plan. **The lack of communication between services threatens to have a detrimental impact on my son's health.**

Being my child's advocate

Having to act as the advocate, I had to be quite forceful for people to take me seriously and take action. At this time in particular, an advocate would have been really helpful to make sure that the different hospitals were communicating with each other as well as making sure that the appointments were being booked quickly. Someone who had a level of authority that could make the doctors accountable for the service they are providing.

There is certainly a role for care coordination beyond health services as well. I have had to go to tribunal to secure support for Teddy at school. Teddy receives speech therapy at school for speech apraxia, as specified in his Education and Healthcare Plan, but the school does not have training on the type of therapy that has been prescribed. Instead of getting access to another therapist, Teddy has been put under supervision. As a result, the agreed approach is not being implemented but nothing is being done about it. The school has been implementing the wrong approach for three years and is wasting money without benefit to Teddy. **I am dedicating my energy on fighting for services rather than focussing on supporting Teddy and my family and putting my own mental and physical health at risk as a result.**

What care coordination means to me

For our family, care coordination would ensure that Teddy is getting all of the care that he needs and help us to access those services without putting that burden onto me and my family.

A care coordinator with specialist knowledge and understanding of the impact of Teddy's condition could help to ensure that we have access to the correct services. They could ensure that doctors were equipped with the correct information and Teddy's medical history before an appointment. They could also help to improve communication between departments, hospitals and services to prevent delays in treatment.

We're already stressed and concerned about Teddy. The extra load of having to battle for access to services has taken an enormous toll on my mental health and well being. I had to start doing a bit less and just give up a bit on a few things, which isn't how it should be. It is very stressful trying to find a balance in the family of making sure that Teddy can get to his appointments, while also looking after our other child, managing work and everything else about living your life. It is extremely triggering, worrying how this will impact Teddy in the future.

RARE DISEASE SPECIALIST SERVICES

EAST OF SCOTLAND GENETICS SERVICES

The East of Scotland Clinical Genetic Service provides a unique service for supporting people living with rare conditions in Scotland. Employing two full-time genetic clinical nurse specialists, the aim of the service is to ensure that patients with rare conditions are supported on a long-term basis and that they are provided with the best information about their condition and its management.

Location: East of Scotland Genetics Service

Kirsten is a Band 7 genetic specialist nurse. Her role is aimed at those with complex (and sometimes multisystem) conditions such as the muscular dystrophies, ataxias and neurofibromatosis providing nurse-led clinics which are likened to a yearly 'MOT'. Each patient and professionals involved in their care will receive either a care plan or clinic summary after each encounter. Care coordination is a vital part of this role for those who need regular screening investigations but don't necessarily require specialist clinical input (this may include cardiac, respiratory or blood tests). In addition they provide support during the transition journey from child to adult services to ensure the process is as smooth and supportive as possible.

Another essential part of this role is multidisciplinary working involving preparing, facilitating and disseminating information for multidisciplinary team meetings and ensuring patients are always provided with feedback about their condition and their management plan.

Grace is a Band 6 genetic nurse, caring for people with a condition such as Lynch Syndrome or Familial Adenomatous Polyposis, or family history giving them an increased risk of bowel cancer. This involves organising screening and offering appointments at a nurse-led clinic to provide support, information, and lifestyle advice. It also involves working with other departments to ensure timely screening for the highest risk patients and to discuss results with Pathology, Endoscopist or Gastroenterology MDT when necessary. Other patients seen at her nurse led clinic include those with a moderate risk family history of bowel or breast cancer for risk assessment and referral for screening, alpha-1 antitrypsin deficiency, haemochromatosis and a follow up appointment for patients recently found to have Lynch Syndrome to give them the opportunity to clarify information and ask any additional questions. Grace is also a member of the Hereditary Haemorrhagic Telangiectasia (HHT) multi-disciplinary team (clinical genetics and ENT), coordinating clinics, maintaining the database, being a point of contact for patients and assessing patients pre-clinic regarding quality of life and epistaxis to determine specific issues to be addressed.

Supporting people with a wide range of rare conditions

Our roles are unique in Scotland and involve coordinating care for people with a wide range of conditions, which is significantly different from that of other clinical nurse specialists whose role is usually single condition specific.

We see many different conditions in our roles so it is not possible to be an expert in each rare condition, but what we do have is an understanding of how challenging it can be to manage your care when you have lots of people involved and a condition that many healthcare professionals may not know a lot about.

Many of the people we work with see many different health professionals, in different healthcare settings. They may have had to attend lots of appointments, repeat their story over and over again, spend time chasing up appointments and have ended up worried and anxious when they have no one to contact to ask questions about their condition or their care.

Not knowing what their condition means for them, as well as being unsure of what to expect from their care, can lead to people feeling frustrated and anxious.

This is where we come in

We are the named contact, reachable by phone and email and on hand to support the person and their family. For some this means just being a point of contact should they have questions, for others this can involve developing and managing the person's care plan, managing communication between specialties and ensuring tests or referrals are actioned. We are here to provide holistic care and we actively work with people to understand their needs and signpost them to appropriate sources of information and support.

We are not here to take control of a person's care, but rather we are here to support them to control their own care.

Person centred care coordination

Sometimes, we meet people with rare conditions we haven't heard of, but this doesn't stop us from offering support. Whilst we always make sure we prepare for appointments by researching the conditions and finding available information, we also ensure that we are led by the person sitting in front of us. It is important to spend time in our initial appointments taking time to hear directly from the person about how they view their condition and how it affects them and their family. The greatest wealth of information always comes from the person.

We are always honest about our expertise and will tell people that we are not experts in their rare condition. It's important that we make that clear but also explain how we can help and support them to find the answers that they need. **We find that people value having a person on their side, a named contact who they can rely on when they need support.**

One of the things that we think works well, and is valued by the people we work with, is building a care plan with our patients. These plans have all the key information about their care coordinated and communicated in a single document. We are

able to include signposting information on these care plans so people can also access information related to their non-clinical needs. We've recently been working on developing new approaches to sharing information, including developing QR codes to link people with relevant information.

Delivering positive outcomes for people with rare conditions

All of this contributes to positive outcomes for our patients. On the one level, there is the management of their clinical care, making sure appointments, referrals and tests are arranged, carried out and acted on in a timely manner which can make life a bit easier for our patients, especially when we can bring things together and reduce the number of appointments needed.

Importantly, having regular contact means we can act to prevent people from reaching crisis. For example, when we know a person has a rare condition which may lead to cardiac problems or perhaps diabetes, we can take proactive steps to arrange regular tests and monitor their health and offer preventative advice. Ultimately, this can not only improve patients' outcomes, but can also save the health service resources by avoiding patients reaching crisis and ensuring they can live better with their condition.

Beyond the direct benefit to our patients, we also think our roles provide significant benefit to our clinical colleagues and NHS Scotland. By acting as a point of contact, and essentially being able to triage a patient's needs, we can reduce consultant time. We save time and can speed things up for our patients by initiating tests and referrals, ensuring our patients are getting the right care at the right time. We are also on hand to support our colleagues in the genetic service, able to provide follow up care for patients, freeing up consultant time and also being a point of information on rare conditions for the service.

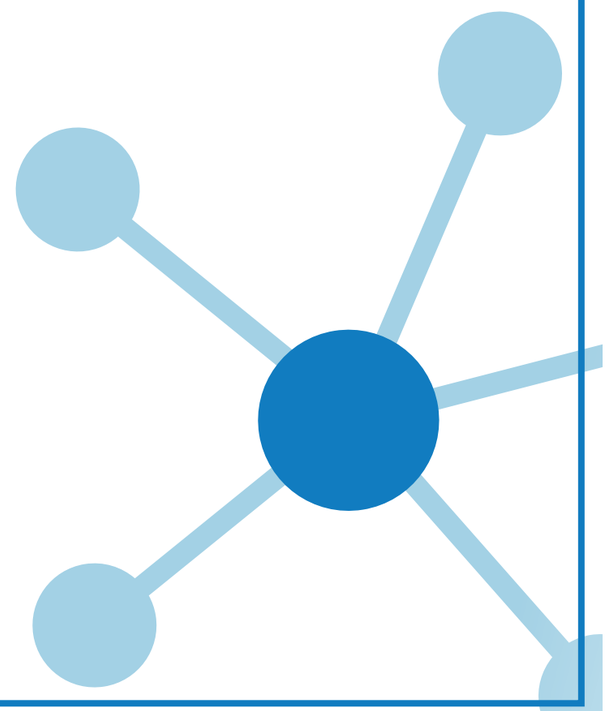
Of course there are some challenges. A source of frustration is the different IT systems. When patients are seeing services in different areas, sometimes we cannot see what has been done. This can result in having to make many phone calls to fill in the gaps. This takes time away from seeing patients. Better connectivity would mean greater efficiency and allow us more time spent face to face with patients.

We thoroughly enjoy our roles and value seeing a variety of different patients with different rare conditions. This allows us to use our skills and to be challenged. It is rewarding to be able to help people who otherwise would be left without good quality information and support.

A model of coordinated care for people living with rare conditions in Scotland

Our roles work well because we work within a collaborative team, within a relatively small geographical area. At the moment we are providing a unique model of care in Scotland, which benefits people living in the East of Scotland. It would be wonderful if we could replicate this model in other parts of Scotland so people with rare conditions could have equitable access to this level of coordinated care. Of course there would be some operational differences depending on regional and population variations but we feel the principle of our roles, in that we coordinate care across a wide range of conditions, could certainly be replicated.

It won't ever be possible to have a clinical nurse specialist for every rare condition, but our roles prove that it is possible to provide a high standard of care and support to people regardless of what rare condition they have.



WALES
SWAN CLINIC

Location: Wales

The Syndromes Without a Name (SWAN) clinic pilot was funded by the Welsh Government for 2 years to establish an evidence base for commissioning an all Wales SWAN service. The clinic sees paediatrics and adults, both virtually and in person and takes a multidisciplinary approach. Care coordinators were recruited to support patients and their families and coordinate care.

We know life experiences can be very difficult for patient's living with a rare condition and in particular where we have not yet got that diagnosis. The patient journey crosses every part of the health service, which makes coordination difficult and includes a significant amount of help from social care and education, particularly for children.

Importance of coordination generally for rare condition patients

Individual professionals within the system must understand the needs of rare condition patients. There are simple things that can be done like combining the cardiology visit with the inherited metabolic disease visit. The patient is desperate to get really good care but that one journey is far better than two or three. It's important that physicians themselves designing the clinics are aware of the needs of patients to coordinate these aspects.

The other way is outreach clinics. That is more difficult to coordinate with different specialists. It's important to have someone who has detailed knowledge of the condition. There may be aspects of that care where the local physician can assist the patient in receiving care locally if you are all agreed. We have to be flexible and use local resources where appropriate but driven by someone with specialist knowledge.

It's about navigating the system; what is available to patients, organisations such as yourselves that are very much involved in research and the knowledge of specialist conditions, but also organisations that can help patients in the social and education setting are key.

One of the barriers to good care coordination is electronic health systems and the fact they do not always communicate between different systems. We need to get each system talking to each other especially for complex patients who have lots of agencies who need to work together. In Wales, we can use the Welsh Clinical Portal which provides a good way of seeing Health-care Professional interactions across the Welsh Health system. For all four nations we need to find better ways of transferring data and investigation results for rare diseases patients.

The role of the clinical nurse specialist in coordination of care and research

A key aspect of the SWAN Clinic and I know many patients find this important as they track across the system, is the role of the clinical nurse specialist (CNS). We have been lucky to secure CNS's in the SWAN clinic, both in adult and paediatric functions. The area that is important, particularly for children and young adults, is the role of care coordination across education and health. The CNS role in the paediatric arm of the SWAN clinic will liaise with schools where necessary who want to better understand a condition and the needs of that patient irrespective of whether we diagnose them.

Mental health, particularly with young people, can be supported. We would act as a patient voice – we need to work with the local lead clinician and local mental health services to get services in place. It's about trying to help get the support systems that may assist in improving mental health. Directing patients to organisations such as Genetic Alliance UK who also can assist in that process is very important.

Being involved in research is important for rare diseases patients. We've got colleagues working as part of the SWAN clinic, at the forefront of research. This will be part of the assessment of the clinic – finding new variants and writing up in scientific literature. They need reporting so that others can learn. Research is very much on the agenda and will be part of the outcomes we develop.

The SWAN service – how the process works

The referral comes from a secondary care consultant and we have a multidisciplinary team meeting that looks at the case; assessing the validity of that referral. The team talk about the case and decide whether to do more tests and invite the patient to clinic. The CNS can act as a sounding board and assist the patient.

The CNS was recently updating me about a patient who found it difficult travelling to lots of different appointments. Coming to the one SWAN clinic where it could be coordinated through the CNS and followed up was really useful. The patient was grateful that someone was listening, recognising that we may not get a final diagnosis, but trying to coordinate the pathway. It's absolutely key – the CNS role helps the patient and will be a care navigator through the service.

The specialist must be informed so that we're open and transparent. It's important to recognise that being referred to SWAN will still require local services to be involved and to support with coordination. The patient will remain under the care of the referring clinician which makes care coordination an active two way process but the nurse specialist can follow up with support such as making local physiotherapy appointments or sorting out a new wheelchair. This is an all Wales service so there is the option for the patient to be seen virtually if the patient felt that was valuable to them or to attend an outreach clinic locally. We're hoping to use those opportunities to see how we can provide a better experience for patients and their carers.

We have recruited adult and paediatric geneticists, clinicians, CNS's/ clinical care coordinators as well as clinical fellows.

We send a copy of the letter to the patient with the outcomes and they would be able to take that to their GP. The GP could come back to the nurse coordinator for help with any outstanding questions. Advise may be given to the referring specialist to make recommendations about further referrals. We may offer the family genetic counselling because there are reproductive issues which impact the rest of the family.

We would not be facilitating ongoing active clinical care on a regular basis because it's a pilot at the moment, but we would be advising that could be done by the local physician. Sometimes we cannot completely close the loop. That's going to be an important learning point from the pilot; if we think there's a chance of something coming up later, they stay on the books for further investigation. We will be learning about the criteria for leaving a patient on the list.

We now have full staff in place, we need to decide the learning from the clinic but we are also linking and learning from colleagues across the globe in Perth and Harvard who have set up similar clinics. We have recently agreed a full suite of PROMs and PREMs to be carried out with patients utilising the clinic. Those will be absolutely key in learning, does it actually make a difference to their life? And two, if it does, how could we improve it?

Summary

The key difference in my view, between poor and good care coordination generally is the sharing of knowledge between the patient and the professionals involved in their care and support. The patient should understand their diagnostic journey and be aware of all the steps taking place in the care pathway; what the diagnosis is, what the likely prognosis is, what help do I need and who am I being referred to. When care coordination is poor, the patient is left in a very difficult and unsatisfactory situation sometimes leaving them feeling alone and neglected.

KATIE AND HER EXPERIENCE OF TRANSITION FROM PAEDIATRIC TO ADULT SERVICES

Condition: Ehlers-Danlos syndrome, chronic intestinal pseudo-obstruction

Living with Ehlers-Danlos syndrome and chronic intestinal pseudo-obstruction

I was diagnosed with Ehlers-Danlos Syndrome (EDS) when I was 11 and diagnosed with chronic intestinal obstruction when I was 14.

The EDS affects my joints so they dislocate a lot and I have a lot of chronic pain and chronic fatigue so I use a wheelchair. My bladder and bowel are affected by the pseudo-obstruction and my heart and blood pressure are also affected. The pseudo-obstruction has a large impact on my life as I am not able to eat or drink anything; I receive nutrition through intravenous tubes that go directly into my bloodstream. I also have an ileostomy and use a catheter. It is difficult because it is very unpredictable. I can be fine one day and the next I can be hospitalised.

When I was diagnosed, **I was at the age where I could speak for myself but they always looked to my parents for answers.** It often felt like I was missed out of the conversation. I felt that doctors were taught to speak to very young children or adults and there was little training on the inbetween.

Poor transition experience caused me to avoid hospitals

When I reached 16, I realised that I hadn't started my transition process so I raised this with my doctor. They had forgotten to start the process. **I felt really forgotten about in the whole experience.** There were several things that the paediatrics unit was supposed to do to prepare for my transition to adult care that they didn't do. They were supposed to write a transition letter to the adult team summarising my medical background. I was supposed to go and see the adult ward. I should have had joint clinics with adults and paediatrics.

There was an official 'transition day' planned where I would have an appointment in the adult clinic and I would be handed over. But I became really ill about three days before. I ended up in A&E and no one took responsibility for me. The whole time was taken up with them arguing about whether I should go to the children's or adult ward but no one was focusing on me or my health. While they were arguing, my condition had deteriorated and I had to go to the paediatric intensive care unit.

After that, it was strange because I was put on the paediatric ward, but I was only being seen by adult doctors. It was very confusing because I had spent a lot of time on this ward and now everyone was refusing to see me. Instead I was being seen by people I had never seen before despite the adult team having none of my history and no communication had been made with my paediatric team.

A drastic change in my care

Once I was discharged, I was then under adult care. It was very overwhelming and confusing as there were so many changes I had not been prepared for. Once I entered adult care, it was very clear that the doctors would not speak to my mum and would only speak with me. It was a drastic difference. It was very difficult as I was still at school and the hospital would call me in the middle of class. It took some time to establish with the doctors that my mum would still have some input in my care. In particular, when I am very ill, I cannot speak on the phone to a doctor so I need my mum to step in.

My first experience on an adult ward, my parents weren't originally allowed to stay. I was placed on a bay where I was exposed to adults who were severely ill and it was traumatising. **It was such a bad experience and it has affected how I look at being admitted to hospital, I avoid it at all costs.** I was also still in full time education when I moved to adult care but I had no input from the hospital school as that is located in the paediatric ward. As a result, I had to drop one of my a-levels. These two experiences have had a huge impact on my perspective of adult care and have given me a really negative mindset when looking at hospitals.

Care coordination in paediatrics was generally better than in adult care. I had a lead paediatrician who oversaw all of my care and communicated with my specialists. Since transitioning, I have found that adult care is much more specialised and I struggle knowing who to go to for what. I think a lot of adult teams seem to think that the GP should take the lead on coordinating care for an individual. But, when you are medically complex and have a rare condition and your GP doesn't know a lot about it then , it may be better to have a specialist in that role.

Challenges also came as different trusts have different ages for transition. I was under my specialist paediatrician until I was 18, with local physiotherapy I transitioned at 18 but community nursing was when I was 16. The different ages for transition made it even more difficult for teams to communicate with one another. There was also some resistance as adult teams didn't like talking to paediatric teams as they didn't understand that I was still classed as a child under other services.

What should care coordination consider?

At the age of transition, it is important to consider all the other changes that are happening in a young person's life. **A lot of the time it is forgotten that you have so much more going on in your life than just your rare condition.** For me, it was important that the transition process was tailored to my experiences and my needs. Therefore, making the transition a gradual process could help to let the person adjust more seamlessly. It is also important to check-in with the individual, making sure that they are comfortable with the rate of change.

For example, I was referred to University College Hospital London and there, they have a separate department for adolescents. I was given the opportunity to speak to the doctor alone, without my parents. Having that opportunity at a younger age was really important as the responsibility of care shifted from my mum to myself. UCLH also has a young adults department which covers 19-25 year olds. That gradual transition is really nice and less overwhelming.

For me, communication was something that massively failed; between myself and the doctors; between doctors in different hospitals and departments; and between health care and social care providers. If there was better communication between these groups, the whole experience would have been much better.

AILSA AND HER EXPERIENCE OF UNCOORDINATED CARE

Condition: Cavernoma

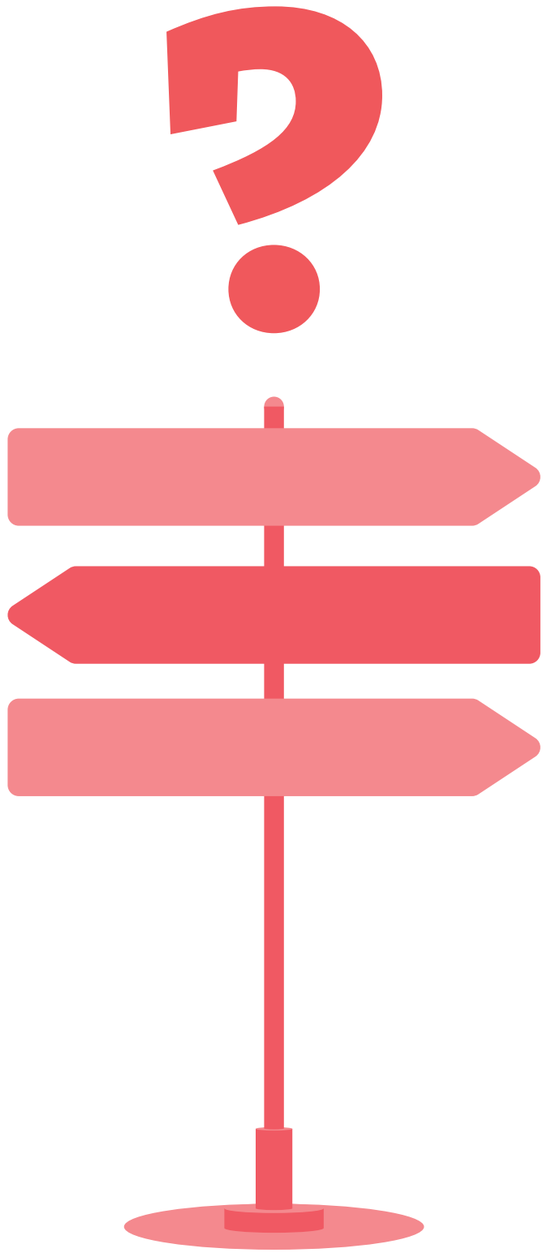
I was diagnosed with a cavernoma that bled onto the right thalamus in my brain seven years ago. I'm able to talk about it now because I've had time to process everything that went on. **At the time, you don't think about care coordination, you rely on others to guide you through when you're ill.**

What living with cavernoma looks like for me

Living with cavernoma has two stages, there's the acute stage immediately after a brain bleed (although this can last a few years) and then the chronic stage. Immediately after a brain bleed, there's a huge combination of physical and cognitive symptoms, some visible and some invisible. For me, I lost function in my left hand side including my arm and hand, I couldn't walk properly unaided, I had extreme fatigue and I was struggling to process things; I struggled with word finding, maintaining attention and being in noisy environments. I had strange sensations in my brain, my eyes would glaze over and I couldn't communicate properly for a period of time. I had to learn how to walk again, practise my language, have physiotherapy and occupational therapy etc. Now, seven years later no one can 'see' the cognitive symptoms but I struggle with them the most, such as information processing, fatigue and I might be having absent seizures. This has had a knock on effect on my work, my finances and my own mental health and emotional wellbeing. I find that in daily life I can't do the things I used to and I lack the support to navigate that.

Poor care coordination leads to a slow recovery

After diagnosis, I was given six weeks with a neuropsychologist where they assessed me, presented me with a bell chart and said you're 'below average' but then had no follow up. My brain was my strength and it became my weakness. It took me another three years, through my work with the Cavernoma Alliance UK, to realise that I could request a referral to a neuropsychologist and occupational therapist to help with my difficulties. I was 25 and I didn't know about these options or how the system works to get this help.



This had a huge impact on how slow my recovery has been; **if I had this support from the outset I wouldn't have lost so much confidence and I wouldn't have felt 'stuck' for so long as a young adult.** It makes you feel incredibly alone, especially if you've never heard of cavernoma or know anyone else with it.

I was also told that I should probably get some physiotherapy but this wasn't followed up. I had to go back to my GP and ask for it. The first available appointment offered to me was eight months later. I couldn't wait that long so I spent my savings on a private neurophysiotherapist. **When I did attend the NHS physiotherapy appointment, they didn't have any information about my condition or my diagnosis and therefore didn't know how best to help me.**

I was responsible for coordinating my own care and communicating information between all of the different doctors I was seeing. At one point I was being seen across seven different specialties in different hospitals. I couldn't drive so my mum had to take me to each appointment. If there was a dedicated person to help coordinate my appointments it would have been helpful.

What care coordination means to me

For me, **care coordination means treating someone as a whole individual,** communicating across different specialties and making shared decisions about a person's care with the individual, not approaching it as a linear process in silos. This isn't just for cavernoma, this is applicable to everyone with complex and rare conditions. **The benefits of having effective care coordination could mean that individuals access appropriate care faster therefore they recover more quickly and can re-enter the world sooner after an acute episode of cavernoma.** It would ease the emotional and mental burden of having to constantly explain yourself over and over; when you're struggling to cope this can be an added exhaustion. With all of this repetition it's easy to lose details along the way, if there was a centralised way of capturing and sharing this information, perhaps a live summary document that can be transferred along with you, things are less likely to be missed.

BLESSING:
CARE COORDINATION FOR SICKLE CELL DISEASE VERSUS LYMPHOMA

Condition: sickle cell disease and lymphoma

My diagnoses with sickle cell and lymphoma

My parents didn't know that I had sickle cell for the first few years of my life. They were quite worried because I was always uncontrollably upset – probably because I was having a crisis and they just did not know what was happening. They ended up having to go to multiple hospitals before they were actually able to understand what my issue was, by then I was four. My family had to manage it the best way they knew how and, at the time, they did not have a lot of information. My parents have done their best to be strong. My mum was very hands-on in caring for me so whenever I had a crisis she was very much there.

My diagnosis with lymphoma started with back to back sickle crises at work. I was in hospital every two weeks for about six to eight weeks. Guy's didn't understand why I was being affected so much by my sickle cell and blood tests came back ok. Then I had pain from gallstones and my mum told me to call the ambulance, which took me to Lewisham hospital, where they kept me for seven weeks, but they still weren't able to understand what was wrong with me. About a week after I was discharged, Lewisham hospital called to say that my test results were abnormal and that I might have tuberculosis or lymphoma. Then a couple of weeks later Guy's called telling me to come into the hospital immediately. The type of lymphoma I had presents itself at a late stage, so I had to be treated immediately. They told me that I was at a late stage of my cancer and I had to be treated immediately. They gave me a 30% chance of surviving. It was a rare form of lymphoma and they had to consult with a US doctor to agree on the treatment. The cancer was completely unrelated to the sickle cell even though they are both blood conditions. I'm not sure if they found the cancer faster because of the sickle cell or not, I'm not sure why Guy's hospital missed it when I first got seriously ill.

To find a bone marrow donor they wanted to go through all my family members. They tested my mum, my dad, and then they tried my younger siblings. They started with my eldest sister. She was a match, but that bone marrow donation triggered a sickle cell crisis for her. She must have had a really mild form that they didn't spot. It's really upsetting that my treatment caused a crisis for my sister. But the positive is that I am now five years post cancer treatment and I have not had a crisis in all that time. Before that, I was in hospital three to four times a year because of how severely my sickle cell was affecting me.

What care coordination means to me

For my sickle cell, I have always been looked after by two doctors and two nurses that I have known from a young age. From that perspective, it makes a massive difference because the care can be a little bit more personal. They are able to identify me as a human and not just a number within the system. I've had good care coordination in individual organisations but not across the board. For example, I could go to Guy's and then go to my GP tomorrow and they will know nothing about what I've just spoken about with Guy's. It is very repetitive. It's the same for A&E, they ask what my issue is, and then at the ward you have to explain it over and over again. I think why am I doing this? This is a hospital I come to frequently. **So care coordination is a beautiful ideology, it's just not the truth for my sickle cell.**

I have had a care coordinator but that was for my cancer

She was called Karen and she was absolutely amazing. She knew every piece of information that was necessary at the time and she did her utmost to keep me in the loop with everything. She was the first person in the NHS that gave me her email address, and I still appreciate that communication to this day. She's also the reason why I was able to still be paid whilst I was off work with my cancer. I was so worried about my financial situation and she was just amazing. She explained the benefit system and helped get my application processed quickly. That made me feel like I was able to manage the whole cancer situation.

I think the contrast is genuinely amazing. It's a real shame. Karen is actually how I knew that there was a sickle cell social worker. Just like when I was younger my mum only found out that I was actually entitled to disability benefits through the sickle cell grapevine. And because my mum only got very basic support to apply, nothing came of it and she continued to struggle financially. I also didn't know about free transport until my cancer. There were times where I had hospital appointments, and I couldn't get to the hospital. My parents couldn't take me, I didn't have a car. I obviously couldn't drive and I didn't know that getting transport from the hospital was available until Karen told me.

So there's a lot of things that could have helped my lifestyle if they just cared enough to put in that little extra mile.

The difference care coordination would make

Sickle cell is an unseen disability that people do not understand. I think **better coordinated care would relieve a lot of this frustration that we feel when you're having to explain yourself multiple times.** That will take away pain from myself because sometimes sickle cell is worse when you get worked up, the stress is a trigger and having to constantly explain yourself and feeling like you're not being understood is a trigger point in itself. It gets to a point where sometimes you just want to give up because you just think, what's the point if no one is ever going to be able to understand me.

Coordinated care would make me feel at ease. Now I'm not at ease because I know if I go to Croydon University Hospital for example, I know that I have to be almost on top of my senses in order to be able to explain what care I need. I've been to A&E and I've had to wait nearly five hours just to get painkillers, where I thought I would pass out because of how severe the pain was. And I know if there was communication from Guy's, to Croydon or from my GP to Croydon, or they were able to access my records easily, I know for a fact that the situation would have been different but it's never been like that.

It would also have brought a little bit more understanding of the disease especially for my family, because my mum and dad are immigrants who have not always fully understood sickle cell. If things had been streamlined maybe they would have understood the disease a little bit more themselves. Better understanding would have made them feel safe because there's times where they felt worried because of all my appointments.

Mental health and contact with my school are two gaps that I've missed. My hospitals have never asked about my mental health. They never made a statement to let me know that this is something that could affect your education and that there might be times where you're going to feel this type of way when you're at school.

If my care had been coordinated and touched on all these points my life could have been more manageable.

DONNA: NHS TUBEROUS SCLEROSIS CLINICS NETWORK

Condition: tuberous sclerosis complex

Our journey with tuberous sclerosis complex

During the 'Beast from the East', we heard noises in the middle of the night and thought it was the storm, but it turned out to be our six-year-old son having had a seizure. My husband ran and brought him into our bedroom. We thought he'd had a stroke, because his face had dropped, he was dribbling and his eyes were rolling back but a seizure was not something that we'd even contemplated. We actually rang 111, not 999, because we never believed that our child could need an ambulance. Due to the snow it was not possible for an ambulance to get to us so my husband ended up driving us to the Bristol Children's Hospital and that's when we were told he had had a seizure. 'Everyone is allowed one seizure but if he has more then call an ambulance immediately' we were told.

Three nights later Wills had joined us in the night because he couldn't get back to sleep, and right in front of our eyes we saw him have a full-blown tonic clonic seizure. We called 999 and things escalated from there. That was March 2018, and on the first of May 2018 he was diagnosed with tuberous sclerosis complex (TSC). We had never heard of it before. The MRI scan showed that Wills was riddled with tubers (benign tumours) in his brain. Everyone with TSC is affected differently. For Wills it's mainly in his brain. He has small tubers in his kidneys which are monitored, and some on his skin, but his eyes, lungs and heart are fine.

I never thought I would ring 999 so many times. It has affected the whole family. My older son is now 13 and he won't sleep at other people's houses because he can't face waking up in the morning not knowing if we were there or not and if an ambulance had been called.

Surgery for seizures

In September 2018 we were brought in for an epilepsy surgery programme where Wills spent a week in a private room being monitored with lots of wires on his head (an EEG) to understand whether surgery would be possible. After the investigations it was clear surgery wasn't possible because the seizures were firing from all over the brain. Wills' seizures very quickly escalated and required lots of medical interventions constantly which resulted in us spending 2019 in hospital. We would be discharged one day and back in an ambulance the next. Wills was in hospital during Covid times, where only one parent at a time was allowed in hospital, even in intensive care where he ended up.

During the Covid period Wills' condition progressed. He was on a concoction of medication and had tried many more with no positive results, so he is now classed as medicine intolerant. Wills was admitted to hospital due to a medication side effect on his liver, and during this stay in September 2021 the clinicians realised how the seizures that we had been managing at home had worsened. His breathing was stopping and he was having so many different styles of seizures including 'drop attacks'. This is where the signal spreads so quickly from one side of the brain to the other so you lose all the use of your limbs and you drop head first, like a bowling ball, onto the floor. He was having to wear a hard helmet, and all of a sudden, we had a 10-year-old child that's back to using a beaker for safety reasons. There were days where he would need to be in his wheelchair because he could have 40+ drop seizures in a day.

To solve this, Dr Amin collaborated with colleagues and an expert from the USA to organise a procedure called a corpus callosotomy, where they cut some of the connections between the left and the right side in half to stop the seizure signals travelling from one side to the other. The recovery was horrible. Wills had to learn to talk and walk again. He was very sick and attached to tubes for weeks. The recovery in hospital ended up being three months, and his brother couldn't visit because of the Covid rules still in place It's a long time to go as a separated family. Touch wood, he has not had a drop seizure since, and he's gone a year without wearing a hard helmet on his head, which is massive for an 11 year old.

Since the operation all of his seizures happen in the night. They are tonic-clonic and tonic seizures that can have a really big impact, and knock him out for the next day. We've had to get a downstairs extension and set up the house so that we can care for him properly. We've added a sliding door between our bedroom and his bedroom so at night we can sleep with the door open and keep an eye on him.

Healthcare coordination

Sam's (Dr Amin) secretary plays a huge part. She's the middle person between the parent and the consultant. I feel like I've got a good relationship with both her and Sam. We can go from an email or telephone call to being in hospital having tests the same day.

The epilepsy surgery nurses are an amazing team of three who work in the Bristol Children's Hospital. They are on hand via email or telephone whenever we need them. We can send videos to ask about a particular seizure or to say things have changed. Anything at all like asking for advice on a risk assessment for a school trip and understanding more about how other families have tackled challenges.

Wills has a team of community nurses. They are brilliant and they will do as much of Wills' care as they can in our house, which just means it's all a bit nicer and easier. Wills has a portacath in his chest for bloods, and emergency seizure medication, which needs to be flushed every four weeks.

Wills has a GP who is very lovely and happy to help when he can, however the reality of it means we very rarely use him as Wills condition is rare and specialised, so we go straight to his consultant, Sam Amin.

The epilepsy nurses and Sam are based on a neurology ward at Bristol Children's Hospital. They can exchange information easily via email or in person. The lifetime care team is based outside the hospital. They can access his health records. As a parent we have to act as a project manager and bring all his care together. For example, one of the bloods that William has done, can't go to the Bristol Children's Hospital. It must go in an envelope in the post to London, but the nurses that come and do the bloods don't have the paperwork that goes with it or the envelope. That has to come from Sam's secretary. I coordinate this to make sure that we've already got them at home with the correct bottle.

Mental health

Mentally, it's a big change for me and for him, and for us as a family. Wills doesn't speak about it much; he's a stereotypical boy, who doesn't really want to talk, but he definitely struggles on some occasions. He'll definitely say on random occasions, for example, brushing his teeth, 'I hate my life. Why do I have to have seizures?' We try and have a conversation with him about it, but he's moved on and closes up. It's tough and sad to hear your child talk this way.

Outside of health

Outside of health, there is a massive gap in coordination and help. As a parent, you feel very lost. This world we found ourselves in with a child who now has a lifetime medical condition is really quite difficult. There's no place to go to tell you what's available, if your child needs this, what you should/could be entitled to. It's all hearsay. It's only from being in hospital and hearing from other parents that we found out about some things. I do feel there is a gap in the market for a job role that could connect all the sections together and sit with families to help them, or even a simple website.

Wills moved to a senior school in September 2022. It's an amazing school that meets his needs, but the amount of fighting and paperwork that a parent has to do is callosal. I had never heard of an EHCP and found myself working with Wills' learning mentor to complete one. We were up until midnight on many occasions working on it. At this point I was in hospital as Wills had once again been admitted. The help available was very limited and without having a truly kind and caring learning mentor to help me (which was not her role), I'd have been totally alone.

A growing relationship

The care that we have has definitely evolved. Sam and I understand each other well. We trust each other and we both know the level of care that Wills requires. It has taken years to build up this relationship. Sam's passion for TSC is apparent to anybody who knows him. You can see that he truly cares for all his patients.

Condition: tuberous sclerosis complex

Location: UK-wide

Sam Amim, Consultant Paediatric Neurologist, University Hospitals Bristol.

The impact of TSC

Tuberous sclerosis complex (TSC) is a condition involving multiple organs including the lungs, kidneys, skin, bowel, pancreas, liver, and eyes. The genes in the two types, TSC1 or TSC 2 cause benign tumours to grow in these organs causing a huge impact in a wide variety of ways. Some of the most common impacts of TSC include seizures, developmental delay, skin abnormalities, and intellectual disabilities. The severity of TSC on individuals can vary, with some people experiencing mild symptoms, while others may have more severe symptoms that affect their quality of life.

Taking the central nervous system as an example, many people with TSC have epilepsy. Two-thirds of these patients have refractory, difficult to control epilepsy. Several different medications may be trialled before one which addresses their needs adequately is found. During this process multiple hospital admissions might be necessary, as in Wills's case. That is one example of one issue (epilepsy) on one system (the central nervous system) that might be affected by TSC. More widely in the central nervous system someone with TSC might be affected by behavioural problems, attention deficit hyperactivity disorder, autism spectrum disorder or learning disability. All of those have impacts on school and wider life.

The impact of the condition is not only on people with TSC, carers, parents and relatives are also impacted. Our collaboration of clinicians have contributed to a study that demonstrates that quality of life for parents and carers is poorer compared with the general population and also compared with parents and carers for some other chronic conditions like diabetes, asthma, bowel disease, and cancer.

Care coordination and TSC

With such a complex and variable condition, which affects individuals in so many ways, it is crucial that their care is really carefully managed and monitored. Without this it can become fragmented and disorganised. For each area of impact of TSC, patients require multiple professionals and experts. One patient might need to see at least 15 or 20 experts regularly, and throughout their lives. A care coordinator is vital: an individual who can oversee their clinical management, with expertise in all these disciplines to be able to coordinate their management between these experts.

Where there is poor coordination of care, things can go wrong. This results in greater hospital admissions. For example, if we do not ensure that an individual is receiving the most appropriate antiseizure medications, their seizures may become unnecessarily damaging or more frequent. A consequence of this might be status epilepticus. This is a serious deterioration of health that could have been slowed or prevented with better care coordination. The consequences of this can be as serious as death. There is a psychological impact too. Without good coordination, families feel the need to be closely involved with all planning and care decisions. This can be exhausting and is a heavy burden to carry. When there is a system in place, they can concentrate less on monitoring and managing care, and that takes away some of the stress and strain.

With respect to coordination with organisations outside of the health service, the South West of England service is fortunate to have an epilepsy and TS nurse who is able to support patients, liaise with schools to complete education, health and care plans (EHCPs) and help with disability living allowance (DLA) applications. This role is not part of every team providing care for people with TSC in the UK, so some clinicians do not have access to this support. Without this role a clinician will not necessarily have time to liaise with other authorities. This leaves patients and families without crucial support to deal with the condition, particularly because of the multiple comorbidities with the learning issues, cognitive and behavioural problems.

The other facet of the care coordination service in South West of England is the administrative coordination, where a secretarial role makes sure that patients see all the relevant health care professional, and works to set up those clinics, making sure all necessary information is shared between clinicians.

Funded services would be more equitable

In 2018 our collaboration of clinicians published guidelines for management of TSC patients. There is not currently funding for all centres in the UK to follow these guidelines. The funding and workforce arrangements for TS services around the UK are quite varied and often the result of pragmatic and opportunistic choices by the lead clinicians. One service has managed to create a multidisciplinary team around a nephrologist with a personal interest in the condition, benefiting from their drive and expertise to create an excellent service. In another service funding was secured from a pharmaceutical company, but this is time-limited. In the South West, we have had a long-standing interest and we have managed to find enough resources to be able to fund our service with care coordination.

A new forum within the British Paediatric Neurology Association is for all neurocutaneous (neurological conditions affecting skin) disorders, including TSC. Part of the aim of this group is 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. Multi-organ conditions like TSC require multiple clinicians. The best standard of care is not possible with just one discipline such as neurologists focused on the condition, it is necessary to have nephrologists, cardiologists, dermatologists, neuropsychologists, ophthalmologists and more.

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 delivery equitable high quality care, including the recruitment 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. This would reduce the reliance on charities which currently fund some of the nurse posts, and reduce fragmentation.

Improvements we could make

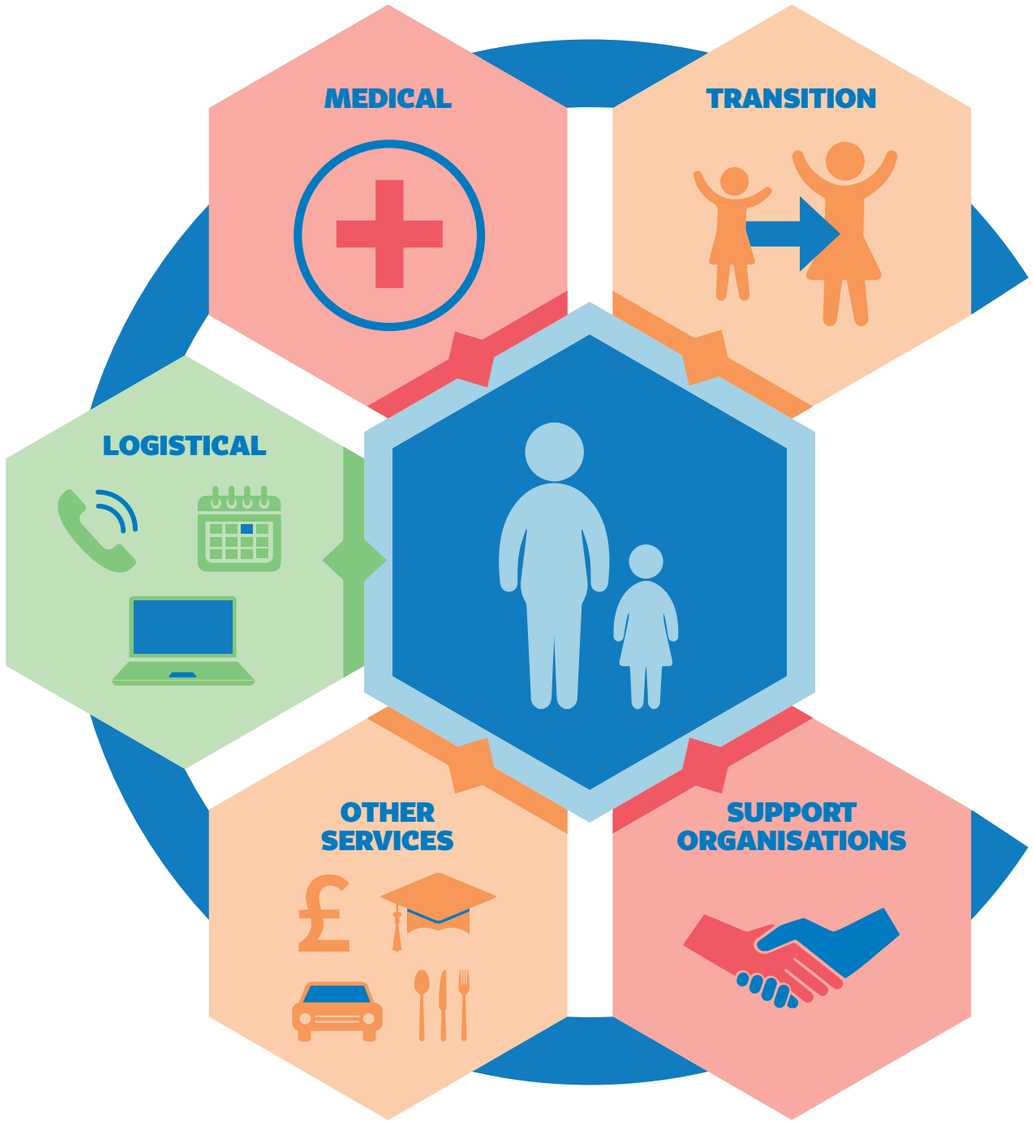
Predictable and adequate funding brings its own benefits. An example is in the magnetic resonance imaging (MRI) backlog nationally which currently means a 12 month wait for a scan for a child or someone with learning disabilities who would require a general anaesthetic. TSC treatment guidelines call for an annual scan. Predictable, national, equitable funding would allow investment in imaging resources. These gaps in care have permanent negative consequences on patients, without sufficiently regular scans it is likely that significant comorbidities, such as brain lesions and eye problems are not caught early enough.

Compatibility and access to health records is a major NHS issue affecting all forms of care. However the treatment geography for rare conditions such as TSC are large, and there can be multiple record systems within the remit of any one clinic. This creates a challenge for coordinators who need to plan ahead to ensure all scan results, reports and correspondence are available for the clinician in time for each appointment.

Mental health is another broad issue that poses challenges more widely than TSC. The guidelines make it very clear that people with TSC should have access to child and adolescent mental health services (CAMHS), but there is not sufficient capacity to offer this currently. This is another issue where there is inequity in care provision based on local arrangements. The South West of England service benefits from good collaboration with a local neuropsychology unit but this is not available in all parts of the country.

BREAKING NEWS

In 2023 the NHS has approved an application for the TSC Clinics Network to be recognised as an official Rare Disease Collaborative Network (RDCN). This means that the network and its clinics will have a stronger role and capability in improving understanding and awareness of TSC in the NHS, standardising clinical and care pathways, and maintaining best practice amongst TSC specialists and non-specialists.



OUR ASK AND NEXT STEPS

This report demonstrates the value of care coordination and the impact of its absence. It also shows that care coordination is happening now, in a variety of forms and settings in the NHS. It is possible. We want to see the delivery of care coordination grow to a much broader range of rare conditions, in a sustainable way.

Three years remain in the timescale of the UK Rare Diseases Framework. Genetic Alliance UK and its members call on delivery partners of the UK Rare Diseases Framework in all four nations of the UK to make progress on implementing Priority 3: Better Coordination of Care.

Future actions should deliver care coordination that:

- **Coordinates complex medical care**
- **Delivers well-organised logistical support**
- **Assists an effective transition from children's services to adult services**
- **Bridges the gap between healthcare and other services, such as education and benefits**
- **Integrates support from rare condition charities and support groups**

To discuss further, please contact:
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GENETIC ALLIANCE UK