THE NEUROLOGICAL ALLIANCE

OUT OF THE SHADOWS

What needs to change for people with rare neurological conditions

Judy Abel

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Rare neurological conditions, taken together, affect at least 150,000 people in England, with possibly many more cases which remain undiagnosed.[i] That is comparable with the incidence of many cancers in England.

Whilst having a rare neurological condition may seem unusual from the perspective of the NHS and society as whole, this does not mean that children and adults with rare neurological conditions should receive anything less than the treatment and care they need. For this reason, the Neurological Alliance and its members are calling for **parity of priority** for people with rare neurological conditions when it comes to their health and care. Rare neurological conditions are health conditions like any others – people with them need to be brought out of the shadows.

To address this requires tackling the historic underfunding and understaffing of NHS neurology services; for example, for every four neurologists in Germany there is only one in the UK.[ii] In addition, despite neurological conditions affecting at least one in six people in England,[iii] they were not prioritised in the 2019 NHS Long Term Plan.[iv] Increased investment, support and focus on neurological services in the NHS and in the health care workforce, would hugely benefit people with both common and rare neurological conditions. With an ageing population and the neurological effects of COVID-19 adding to those requiring the benefit of specialist neurological expertise, NHS resources and staffing must be increased to tackle this.

"Whilst having a rare neurological condition may seem unusual from the perspective of the NHS and society as whole, this does not mean children and adults with rare neurological conditions should receive anything less than the treatment and care they need."

The Alliance has consulted widely in producing this report and many common concerns have emerged from our rare condition members, which we recommend are addressed primarily by those responsible for commissioning neurology services, and in the next Rare Disease Framework, the first part of which is expected by the end of 2020. These concerns are:

1. **Awareness about rare neurological conditions must be improved in primary care.** Health Education England, professional bodies, patient organisations and the NHS more widely, must work together to develop and disseminate information and educational materials to help close the knowledge gap in primary care about rare neurological conditions and to avoid misdiagnosis. The publication of this report hopes to form part of this process.

2. **Perceptions around rare neurological conditions need to change.** Labelling something as 'rare' or 'very rare' can act as a barrier to treatment, creating the perception by clinicians that a particular condition may be so complex that it is too hard to treat; even fairly obvious interventions may not be delivered, simply because of the complex name attached to a condition.

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This needs to change. People with rare neurological conditions have the same right to treatment as any other group in society. Their equal rights are, for example, enshrined in the Convention on the Rights of Persons with Disabilities.[v]

3 **The NHS England and Improvement (NHSEI) Neuroscience Transformation review provides a unique opportunity to transform the care of people with rare neurological conditions.** Optimum clinical pathways are currently being developed for most major neurological condition groups, which, once completed, will cover nearly all of neurology. In its role as Co-chair of the National Neuroscience Advisory Group, the Neurological Alliance is facilitating consultation on the development of these pathways, which include motor neurone disease (MND) and muscular disorders; functional neurological disorder (FND); headache, migraine and facial pain; movement disorders, traumatic brain injuries, autoimmune conditions, epilepsy and multiple sclerosis (MS). These pathways will, together, cover many rare neurological conditions too.

Recommendations 3–7 below, set out the major improvements in care that are needed, which must be reflected in the specialised services commissioned and the treatments approved for people with rare neurological conditions.

4 **Speed of diagnosis must be improved.** In a recent survey of 10 of our rare condition member organisations, conducted in September 2020,[vi] four in 10 organisations that responded told us that the people they represent wait, on average, three to five years for a diagnosis, equating to thousands of people each year. During that time, people's mobility and overall health can decline and families can be under tremendous strain before getting the help and support a diagnosis brings. The Alliance is calling for average diagnosis times to be substantially reduced, through: faster referral to specialists from primary care when children and adults present with unexplained neurological symptoms; faster onward referral to specialist Neuroscience Centres when diagnosis and treatment is not possible in secondary care; and quicker access to appropriate diagnostic and/or genetic tests, supported by the development of new pathways for neurological conditions.

> *'The Alliance is calling for average diagnosis times to be substantially reduced, through faster referral to specialists from primary care.'*

5 **Personalised, coordinated care, meeting individuals' and families' needs must become a reality for people with rare neurological conditions.** Our recent survey of members in September 2020 found that many people with rare neurological conditions do not receive even the basic elements of care such as information about their condition once diagnosed or having a care plan, which would, for example, include the details of who to contact in an emergency. In the survey, six out of 10 organisations responding said that the care coordination received by the people they represent, was 'not very good'. This mirrors exactly the findings of our more extensive National Neurological Patient Experience Survey of 2018/19.[vii] Having a care coordinator, with

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a specific skillset, who could coordinate different elements of care such as outpatient, rehabilitation, mental health and social care services, would undoubtedly improve efficiency – and save valuable clinicians' time. NHS England and Improvement (NHSEI) should carry out an evaluation of how better care coordination for people with rare neurological conditions could be achieved in practice, which staff should ideally be tasked with this, and whether the voluntary sector could contribute to fulfilling such a care coordination role – if they were provided with the resources to do so.

6 **Improved access to diagnostic testing must be matched by better access to new treatments.** With the wider availability of genetic testing and the advent of the National Genomic Medicine Service in England – extremely encouraging developments – the likely advances in diagnosis this will bring should be matched by the provision of appropriate new therapies coming onstream. We urge the National Institute for Health and Care Excellence (NICE) to ensure that its forthcoming Methods Review in 2020/2021[viii] simplifies and streamlines decision-making about access to leading edge treatments, recognising the wider value of therapies that can potentially transform the lives of those with no current treatment options, and their families.

7 **The mental health care of people with rare neurological conditions must not be an afterthought.** It is widely acknowledged that people with neurological conditions are more likely to have comorbid mental health problems than those with other long-term conditions because of the complex interplay of mood and brain function; for some, the reality of living with declining function as a result of having a progressive neurological condition will also have a significant impact on their mental wellbeing. Health and care provision must, therefore, take a holistic, person-centred approach, with services commissioned that address the complexity of people's needs. The new Integrated Care Systems (ICSs) within the NHS in England should develop truly integrated commissioning models for physical and mental health care services.

'Health and care provision must take a holistic, person-centred approach, with services commissioned that address the complexity of people's needs.'

8 **The role of the voluntary sector in supporting people with rare neurological conditions and their families must be acknowledged.** Voluntary organisations are often a lifeline for people with rare neurological conditions, providing support and a listening ear when people need it most. Given the recent pressures on the voluntary sector due to COVID-19, including a reduction in the income of some rare condition charities – with a knock on effect on the research they are able to fund – their vital contribution should be recognised, acknowledged as supporting NHS service provision and, where possible, financially supported by government.

Key recommendations: the actions needed to transform care for people with rare neurological conditions

1. **Change perceptions around rare neurological conditions and their treatment,** so people with such conditions are not seen as 'different' or 'special' in any way, but simply as having the same right to healthcare as any other member of society, based on the principle of equal access for all to services and treatment.

2. **Build on initiatives to gather data on the incidence and prevalence of rare neurological conditions** through the establishment and coordination of patient registries, so the size of the rare neurological population is recognised and understood. Without understanding the scale of the need, adequate health and care services cannot be commissioned and delivered.

3. **Speed up time to diagnosis** through: increasing knowledge and awareness of rare neurological conditions in primary care; swifter onward referral to secondary and tertiary care for diagnostic and genetic testing; and increasing access to neurologists in A&E so conditions such as rare epilepsies, which often present in Emergency Care, can be picked up at the 'front door' of the NHS.

4. **Develop an easy-to-access compendium** of Rare Neurological conditions for use in primary care, which has started with the publication of this report.

5. **Continue to develop optimal pathways for neurological conditions, including rare conditions,** through the NHS Neuroscience Transformation review, setting out: clear criteria for referrals to specialists; standards of care once diagnosed – and for those who remain undiagnosed; access to multidisciplinary care including psychological support and rehabilitation; and integrated social care provision.

6. **Ensure that existing guidance,** such as the NHS RightCare Progressive Neurological Conditions Toolkit and NICE Guidance on Suspected Neurological Conditions, is updated to include reference to rare neurological conditions; includes feedback from voluntary sector organisations working with people with rare neurological conditions; and is used to inform treatment and care options, including referral to specialists.

7. **Improve the transition for young people with rare neurological conditions from paediatric to adult care services,** including ensuring all clinical notes are handed on and supporting young people with any emotional challenges they may be facing.

8. **NHS England and NICE should develop further commissioning specifications** and clinical guidance for rare neurological conditions to raise standards of care.

9. **Improve access to mental health support as a matter of urgency,** including through the integrated commissioning of physical and mental health care for people with rare neurological conditions, coordinated at the Integrated Care System (ICSs) level.

Key recommendations: the actions needed to transform care for people with rare neurological conditions

10 **Identify a named care coordinator for all people with rare neurological conditions,** who can coordinate the various elements of their care, thus saving valuable clinicians' time and improving the experience of care for people with rare neurological conditions and those closest to them. This could, for example be: a specialist nurse; another allied health professional; or a dedicated care pathway coordinator.

11 **Provide written information to people with neurological conditions at diagnosis,** as a matter of course, and signpost them to support available from voluntary organisations.

12 **Embed genomic medicine into the care pathway for people with rare, genetic neurological conditions,** with the consent of individuals and their families, with adequate time and resources allocated to genetic counselling and support being reflected in the NHS tariff, whilst also recognising that not all rare neurological conditions are genetic so the care of those with non-genetic conditions must not be deprioritised in any way.

13 **With the advent of more genetic testing, the Newborn Screening Review Process should be reformed** to reflect advances in genetic testing for rare neurological conditions that are present at or soon after birth, which could greatly improve early treatment options.

14 **Through the forthcoming NICE Methods Review, improve and streamline the way in which NICE approves new treatments,** speeding up access to a wider range of new therapies for people with rare neurological conditions, in line with practice in the rest of Europe, and to capitalise on the advances in diagnosis and genetic testing offered through the new NHS Genomic Medicine Service.

15 **Align the reformed NICE Methods process more closely with new NHS pathways of care** for treating rare conditions. Greater alignment and coordination is needed between the NICE process and the commissioning of NHS services in order that patients don't have to wait longer for access to treatments which NICE has already approved it. This is especially important when a medicine is the first treatment for a particular condition.

16 **Ensure people with rare neurological conditions are adequately represented** on appropriate decision-making bodies such as NICE and in forums such as the Rare Diseases Advisory Group (RDAG) and the Clinical Priorities Advisory Group (CPAG).

17 **Increase awareness around the health inequalities experienced by people with rare neurological conditions,** both those protected under the Equality Act 2010, for example people with a disability, ethnic minorities and older people, and those whose poor outcomes are linked to their socio-economic status.

Key recommendations: the actions needed to transform care for people with rare neurological conditions

18 **Improve social care provision,** where this is required by people with rare neurological conditions and to support families and carers; this should not be seen as an afterthought, but an integral part of their health care. Recent declines in services, particularly post COVID-19, need to be addressed urgently by the Government after years of promises about a new settlement for social care.

19 **Build the neurology workforce,** including consultants, specialist neurology nurses and allied health and care professionals, which will help improve care for all people with neurological conditions, including those with rare and very rare conditions, and support the move towards delivering more integrated care through Integrated Care Systems in England from 2021.

20 **Government should continue to support vital neurological research** in UK institutions and universities, particularly given the impact that COVID-19 has had on voluntary sector research funding. Where possible, the UK should continue to participate in European research collaborations post-Brexit, advancing the understanding of rare neurological disorders, both genetic and non-genetic, leading to further advances in treatment.

'...greater alignment and coordination is needed between the NICE process and the commissioning of NHS services in order that patients don't have to wait longer for access to treatments which NICE has already approved it. This is especially important when a medicine is the first treatment for a particular condition.'

1 Introduction

Having a rare neurological condition may seem an unusual occurrence from the perspective of the NHS and society as whole, but to the person with that rare neurological condition and their family it is their daily reality. Whether it is classed as a rare condition or not, in fact, makes little difference to them – what matters is that they, or their loved ones, receive the best possible treatment, care and support to live with, what are often, very challenging and progressive symptoms. In this report we hope to set out a new way of thinking about rare neurological conditions, which does not set apart the people who have them, but sees them as very much requiring and having a right to the same health and social care as everyone else in society. Indeed, in the context of delivering more personalised care, as set out in the NHS Long Term Plan,[ix] every individual's care should be tailored to their needs. This is our starting point.

Our National Neurological Patient Experience Survey 2018/2019 demonstrated significant gaps in care for all people with neurological conditions. In particular, we found that people with more rare neurological conditions fared considerably worse in access to care and support than those living with more prevalent conditions. This evidence led us to the development of this report.

Our Patient Experience Survey also seemed to indicate differences in access and experience of care across demographic characteristics and indicators. For example, those living in more deprived areas reported greater feelings of pain and discomfort compared to people living in less deprived areas. Women often reported longer waits to see a specialist compared to men. COVID-19 has of course shone a harsh light on some of the health and wider inequalities that persist in our society too, with the virus having a disproportionate impact on many people who already face disadvantage and discrimination. Tackling health inequalities has to be at the heart of any future initiatives that seek to transform treatment, care and support for people with rare neurological conditions.

As the UK Strategy for Rare Diseases comes to an end at the end of 2020 and a New Rare Disease Framework is being planned and having consulted with our members and clinical colleagues, the Neurological Alliance, (hereafter called the Alliance) is setting out what we think should be in the new Framework, to meet the needs and expectations of people with rare neurological conditions.

'Our National Neurological Patient Experience Survey 2018/2019 demonstrated significant gaps in care for all people with neurological conditions. In particular, we found that people with more rare neurological conditions fared considerably worse in access to care and support than those living with more prevalent conditions.'

2 About rare neurological conditions

What is a rare condition?

A rare disease is a health condition that affects a much small number of people compared with other prevalent diseases in the general population. The 2013 UK Strategy for Rare Diseases defined a rare disease as 'a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000 and which requires special, combined efforts to enable patients to be treated effectively'.[x] Symptoms can be non-specific, complex and progressive, affecting many different organ systems, often making them hard to diagnose. Latest estimates from the Orphanet database of orphan drugs and rare diseases suggest that there are more than 6,100 rare diseases (excluding groups of disorders and disorder subtypes), around 72% of which are genetic and around 70% of which begin in childhood;[xi] other estimates suggest the total number of rare diseases is higher at around 8,000.[xii]

For the purposes of this report we will referto rare neurological diseases as rare neurological conditions.

Perhaps surprisingly, different countries use slightly different definitions of what constitutes a 'rare' condition in their health systems, but there is a growing move to have a standard international definition of what constitutes a rare condition. The European Union defines a disease or condition as rare if it affects fewer than one in 2,000[xiii] people within the general population – equivalent to around 34,000 of the UK population per condition (or 28,000 people in England); however, worldwide, national definitions vary from a prevalence of five to up to 86 persons per 100 000 of the population.[xiv]

Definitions do matter, as if a condition is classified as rare it may attract fewer resources, or a different commissioning structure may apply. In England, for example, there is specialised commissioning for rare conditions and highly specialised commissioning for very rare conditions that generally affect less than 500 people in the population.

Taking all rare conditions together, one in 17 people will be affected by a rare condition at some point in their lives. That amounts to 3.5 million people in the UK. That is approximately the same number of people affected by type 2 diabetes.[xvi] Seen in this context, rare conditions as a group are not that rare at all. In terms of rare neurological conditions, the Alliance's last Neuro Numbers report of 2019 estimated that there are at least 150,000 rare neurological cases in England.[xvii] Other estimates from the US suggest that a third of all rare conditions have at least a neurological component,[xviii] leading one to estimate that perhaps a far higher number of people in England have a rare neurological condition or symptoms.

> *"Taking all rare conditions together, one in 17 people will be affected by a rare condition at some point in their lives."*

Some rare neurological conditions will affect just a few hundred people, for example Batten disease, a neurodegenerative condition of which there are 13 different genetic types, while others will affect thousands of people, for example Huntington's (a rare,

2 About rare neurological conditions

inherited condition that causes the progressive degeneration of nerve cells in the brain); most will be genetic (around 70–80%), but not all; most will be progressive, but a minority will be intermittent; most will begin in childhood, but some conditions manifest themselves only later in life.

The more we understand about many rare neurological conditions, the more apparent it is that there are many variants between different syndromes which were previously thought of as one disease. For example it is now more accurate to talk about the epilepsies than epilepsy, although all conditions are characterised by seizures with other symptoms. The rare epilepsy conditions, in fact, make up a significant proportion of the epilepsy population, indicating that even more common conditions can have much rarer variants. Clearly then, there is significant diversity within rare neurological conditions, but, taken together, they represent a major condition group. In Appendix A, based on our Neuro Numbers 2019 report,[xix] and the results of a recent survey of our rare neurological condition members, we list around 40 rare neurological conditions, their prevalence, whether they are genetic, which groups of the population are generally affected and whether there is a genetic test available for that condition. The list is not exhaustive, but it illustrates the diversity of rare neurological conditions and why making a diagnosis can pose challenges. Delivering care and treatment clearly needs to reflect the needs of different rare neurological condition groups – a one-size-fits-all approach is not possible – but, nonetheless, there are many common approaches which, if adopted, would significantly improve diagnosis, quality of care and access to new treatments. We discuss these options later in the report.

We should also not omit to mention those people who have neurological symptoms, including epilepsy and learning difficulties, but no formal diagnosis – usually children and young adults. Those young people who have an undiagnosed genetic condition, or a 'syndrome without a name' often present a complex picture to clinicians, although they are, generally, the rarest of cases.[xx]

Finally, people with neurological symptoms coming from overseas to settle in the UK, including from less developed countries, should not be overlooked. For families from countries with limited access to medical resources, individuals affected by a rare neurological disorder can sometimes have been given an inaccurate diagnosis in their home country. Where these individuals come into contact with medical services in the UK, an assessment needs to be made of how that diagnosis was reached and, where appropriate, a second opinion from a paediatrician or specialist neurology service should be sought.

"Delivering care and treatment clearly needs to reflect the needs of different rare neurological condition groups – a one-size-fits-all approach is not possible – but, nonetheless, there are many common approaches which, if adopted, would significantly improve diagnosis, quality of care and access to new treatments."

3 A timeline of recent UK initiatives to improve treatment and care for people with rare conditions

Although not specific to rare neurological conditions, there have been many significant initiatives in recent years to improve the provision of care for people with rare conditions, including to take advantage of new opportunities presented by advances in genetic testing, genomic medicine and more general investment in the life sciences sector.

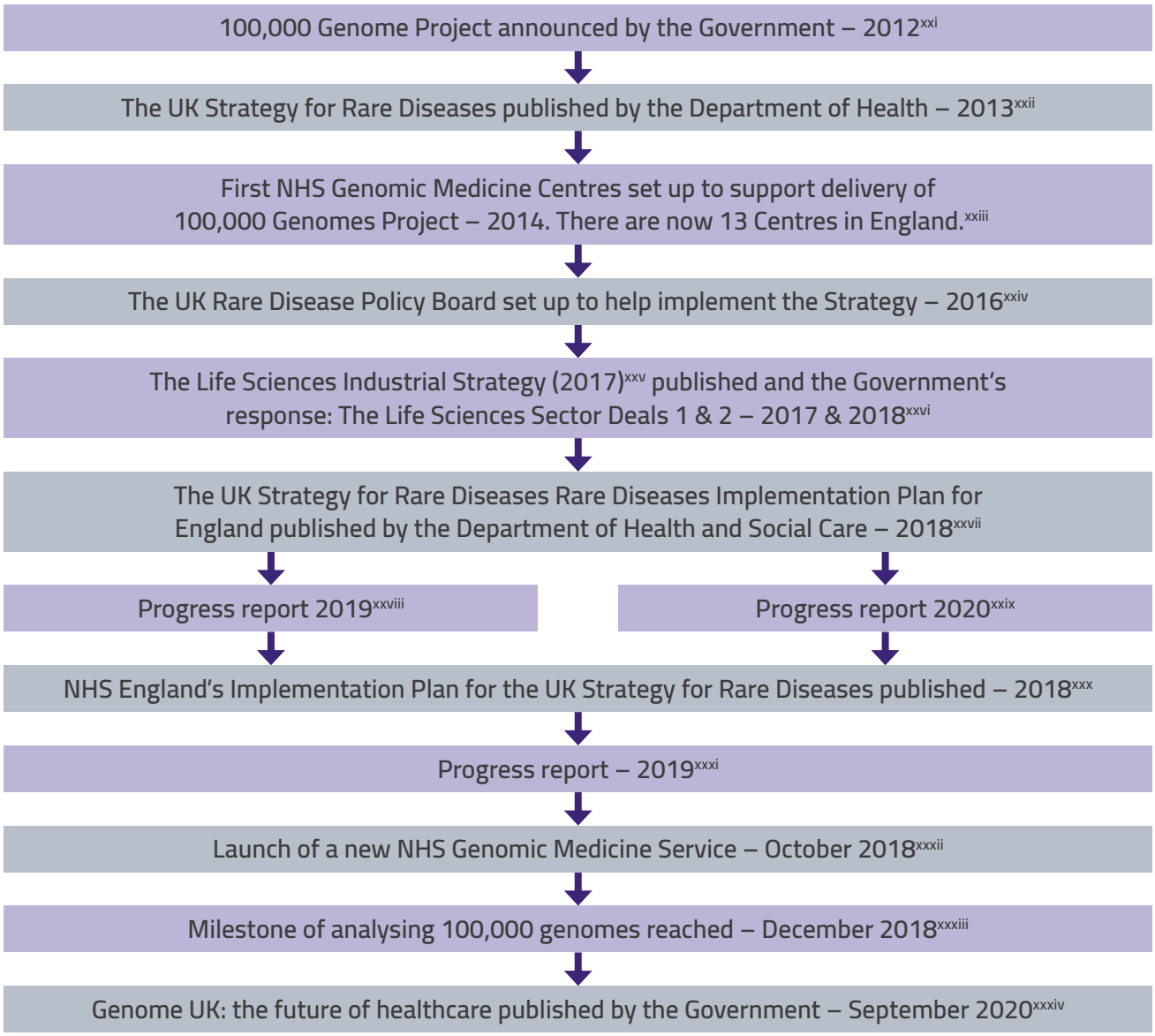

Fig. 1 – A timeline of advances in policy and practice on the treatment of rare conditions

3 A timeline of recent UK initiatives to improve treatment and care for people with rare conditions

This timeline is not exhaustive, but rather focuses on the most significant care and treatment milestones and initiatives over the past seven years since publication of the Rare Disease Strategy. We do not intend to scrutinise the history of these events, but some developments are important to note from the point of view of people with rare neurological conditions. Implementation of the Rare Disease Strategy was also split between the then new Department for Health and Social Care and the NHS, potentially leading to some confusion and duplication around their respective roles and responsibilities.

The UK Strategy for Rare Diseases of 2013 had five overarching goals:

- Empowering those affected by rare conditions
- Identifying and preventing rare conditions
- Diagnosis and early intervention
- The coordination of care
- The role of research

The Rare Disease Strategy listed 51 specific commitments under the above five headings: many were concerned with equality of access; improving patient engagement; supporting clinical expertise in specialist centres; developing new care pathways and evidence-based treatment; improved access to new treatments; and more generally promoting collaboration between the NHS, research communities, academia and industry. However, in practice, how much has really changed? Our National Neurological Patient Experience Survey 2018/2019[xxxv] found that people with rare neurological disease fared worse on almost all the key indicators of care.

The Department of Health and Social Care's Implementation Plan[xxxvi] did not follow for a further five years, an extremely lengthy delay which took much of the wind out of the Rare Disease Strategy's sails. The Implementation Plan looked back at what had been achieved in terms of achieving the five overarching Strategy goals and, belatedly, reported on measures such as: the establishment of the UK Rare Disease Policy Board – which included two patient representatives; the greater involvement of people with lived experience in research, for example through NIHR's INVOLVE project; the launch of the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS); the setting up of the Genomic Medicine Service to 'embed genomic medicine into routine care pathways'; and the introduction of the Accelerated Access Pathway in 2018 to 'get innovative treatments to patients more quickly'. These developments will be considered in more detail later in this report.

In September 2020, the Alliance carried out a short survey of 10 of our member charities representing people with different neurological conditions, to determine to what extent these goals were, and still are, relevant and how much progress has been made in delivering on these goals. The results of this survey are highlighted throughout this report.

The devolved Nations produced their own Implementation Plans and updates in response to the original Rare Disease Strategy.[xxxvii, xxxviii, xxxix]

NHS England's Implementation Plan of 2018[xl] set out NHSE's proposed actions against the 51 commitments in the Strategy in the areas for which it had a lead responsibility, in particular focussing

3 A timeline of recent UK initiatives to improve treatment and care for people with rare conditions

on facilitating earlier diagnosis and intervention, improving care coordination and promoting research. NHS England noted some of their key achievements to date:

- The development of genetic testing to underpin the new NHS Genomic Medicine Service launched in 2018 and the initiation of new genomic medicine pathways.

- An emphasis on personal care plans for patients, bringing together health and care services, with more support for patients and their families.

- The development of new criteria by NHSE to hold providers to account for the standard of care provided to people with rare conditions via a new rare conditions 'insert' in NHS contracts, requiring providers to demonstrate they have delivered on certain key elements of personalised care, for example care plans. These 'inserts' were first introduced in NHS provider contracts in 2018/2019.

- The development of Rare Disease Collaborative Networks, groups of providers with an active research interest in a particular rare/very rare disease, the idea being that such providers would be more likely to deliver better and more coordinated care to patients given their expertise, with associated improved outcomes. Only two such Networks have been established to date, although one is for CDKL5 Deficiency Disorder which causes seizures.

- Partnering with the Academy of Medical Royal Colleges to set up the Genomic Clinical Leads Group, bringing together members from all Medical Royal Colleges in two main workstreams: education and training and clinical pathways. The education and training workstream is designed to ensure that all relevant healthcare professionals have the tools and support to deliver genomic testing, while the clinical pathways workstream is developing care pathways for genomic testing that achieve maximum benefit for patients, while minimising any potential risks.

- A commitment to work with the Royal College of General Practitioners and the Nursing & Midwifery Council about opportunities for GPs and health visitors to facilitate earlier diagnosis of rare conditions.

- Exploring with NHS Digital the benefits and feasibility of inserting a 'red flag' onto the summary care record of any patient with a rare condition whose life would be at risk if they are treated incorrectly.

- Developing a consensus on data-sharing to support NHS clinical genetics and genomics services.

In 2018, NHSE said in the two remaining years of the Strategy its priorities were to:

- Raise public and service provider awareness of rare diseases.

- Strengthen the provision of information; and

- Involve patients in research and policy activities.

Unfortunately, it is not clear how much these listed achievements have actually had an impact. For example, in talking to clinicians for this report, there was not much agreement on whether the rare disease 'insert' was really making

3 A timeline of recent UK initiatives to improve treatment and care for people with rare conditions

a tangible difference in terms of performance standards by NHS providers. There still needs to be an evaluation into the insert's effectiveness. Likewise, despite efforts to improve knowledge about rare neurological conditions in primary care, this does not appear to be feeding through into much speedier diagnoses. There has been much progress in theory, but has it really resulted in improved care for people with rare neurological conditions? We examine this in the context of our most recent Patient Experience Survey in the following Section of the report.

"...despite efforts to improve knowledge about rare neurological conditions in primary care, this does appear to be feeding through into much speedier diagnoses. There has been much progress in theory, but has it really resulted in improved care for people with rare neurological conditions?"

4 Our Patient Experience Survey 2018/2019: how did people with rare neurological conditions do as opposed to people with more common conditions?

In 2018 and 2019, the Neurological Alliance conducted its third Patient Experience Survey of people with neurological conditions, to find out about their treatment and care, which received more than 10,000 responses, 1,742 from people with rare neurological conditions. The findings of the survey and our recommendations for service improvement were published in our 2019 Neuro Patience report.[xli]

When looking at the treatment and care which people with rare neurological conditions received, compared to people with more common neurological conditions such as Parkinson's and multiple sclerosis, it was clear that people with rare conditions generally fared worse on most treatment and care indicators. A technical report on the 2018/2019 PES is available with the full survey results and methodology.[xlii]

The following table presents some key extracts from the report's findings:

Patient Experience Survey Question (2018/2019)	Non-rare conditions – percentage of respondents who agreed with the statement	Rare conditions – percentage of respondents who agreed with the statement
Q. Did you/your family understand the explanation given to you at your diagnosis/when you were first told about your condition?	65.6%	58.7%
Q. When you were told you had a neurological condition, were you given written information about this?	45.0%	33.8%
Q. Information about my specialist treatment and my condition is effectively passed on to the people that care for me (such as my GP, nurse, or family/carer).	53.2%	46.5%
Q. Have you been offered a care plan to help manage your neurological condition?	30.3%	26.3%
Q. Do you see a specialist nurse for your neurological condition?	56.0%	39.2%
Q. Have you been asked about your mental wellbeing by a health or social care professional?	44.7%	33.1%
Q. Are your social care needs being met?	30.5%	27.2%

Table 1 – Extract of results from Neurological Alliance Patient Experience Survey 2018/2019 – rare v non-rare conditions

4 Our Patient Experience Survey 2018/2019: how did people with rare neurological conditions do as opposed to people with more common conditions?

Overall, the Alliance's Patient Experience Survey 2018/2019 found that people with rare neurological conditions were:

- Less likely to understand the explanation about their condition given to them at the time of diagnosis.
- Less likely to be given written information about their condition at the time of their diagnosis or to be signposted to further information.
- Less likely to have information about them passed effectively between professionals.
- Less likely to be offered a care plan.
- Less likely to see a specialist nurse.
- Less likely to be asked about their mental health and wellbeing or to say their mental health needs were being met.
- More likely to report a worse experience of social care, in terms of delays and their overall experience of social care.

"When looking at the treatment and care which people with rare neurological conditions received, compared to people with more common neurological conditions such as Parkinson's and multiple sclerosis, it was clear that people with rare conditions generally fared worse on most treatment and care indicators."

5 What are the key challenges to delivering high quality care for people with rare neurological conditions?

Some of the current systemic challenges to providing high quality treatment for rare and very rare neurological condition include the following key factors:

- There has been a lack of adequate data on the incidence and prevalence of rare neurological conditions, leading to the potential under-commissioning of specialist services.
- There is still a lack of knowledge about rare neurological conditions in primary care, especially where symptoms are less easily defined, for example in conditions such as dystonia (a neurological movement disorder in which a person's muscles contract uncontrollably, not technically rare) or ataxia (a group of rare neurological disorders that affect balance, coordination and speech).

"For some people with a rare neurological condition, obtaining a diagnosis is particularly difficult given their unexplained symptoms, which may adversely affect the management of their health and their wider care."

- The UK has traditionally had an overstretched neurology workforce compared to many other European countries, meaning adults with rare neurological conditions may experience long waiting times to see a specialist, to have appropriate diagnostic tests and to receive a diagnosis and appropriate care and treatment.
- For some people with a rare neurological condition, obtaining a diagnosis is particularly difficult given their unexplained symptoms, which may adversely affect the management of their health and their wider care. Some people never receive a diagnosis.
- At diagnosis, too often there is a lack of high-quality, tailored information provided to people with rare neurological conditions and their families to help them understand their condition and potential treatment choices.
- The mental health needs of people with rare neurological conditions can often be overlooked due to the complexity of their presentation and symptoms, depriving them of the emotional support they need.
- The rehabilitation needs of some people with progressive rare neurological conditions is not always adequately prioritised along the entire treatment pathway leading them to lose mobility more rapidly.
- There can be a lack of coordination and information-sharing between primary, secondary and tertiary care providers and, consequently, of joined-up care planning.
- People with rare neurological conditions and their families may have long distances to travel to specialist centres for their treatment. For this reason, in some situations, it may be appropriate to continue using the

5 What are the key challenges to delivering high quality care for people with rare neurological conditions?

remote digital appointments made necessary by COVID-19, although this should not automatically become the norm.

- There is a lack of knowledge about genetic testing and genomic sequencing amongst many clinicians, which can hamper progress towards diagnosis and possible treatment; over time this situation should improve through the activities of the Genomic Education Programme, in addition to work underway to develop a Genomic Competency Framework for genetic testing to assist clinicians.[xliii]

- There has traditionally been a lack of therapies/interventions for most rare neurological conditions, although, again, this is starting to change through the availability of genetic testing and new, targeted therapies, the number of which is likely to grow rapidly in the future.

- The cost of advanced therapies benefitting relatively small population groups has not been spread across the NHS, leading to the relatively limited approval of new therapies for rare conditions by bodies such as NICE - and therefore to the limited availability of potentially life-changing new drugs for people with rare neurological conditions. A lack of funding for new NHS services to deliver such treatments should also not become another barrier to access.

- The research efforts of small charities funding research into single rare neurological conditions has, in many instances, been badly affected by COVID-19.

- There can be a lack of awareness amongst health care providers, and wider society as a whole, including social care providers, employers and financial assessors, of the effects that rare neurological conditions can have on a person's education, family, social relationships, ability to work and financial security.

"There can be a lack of awareness amongst health care providers, and wider society as a whole, including social care providers, employers and financial assessors, of the effects that rare neurological conditions can have on a person's education, family, social relationships, ability to work and financial security."

6 The age of onset of symptoms of rare neurological conditions

Just as there are many different types of rare neurological conditions, the age of onset of rare neurological conditions also varies significantly.

Diagnosis soon after or around birth

Some rare neurological disorders are 'congenital,' meaning they are present at birth. Others are 'acquired,' i.e. developed after birth. Those with an unknown cause are termed 'idiopathic.' In some cases, a rare neurological condition will, sadly, be evident in the first months of life because an infant is struggling with basic motor skills and/or cognition. This is clearly deeply distressing

> *'In some cases, a rare neurological condition will, sadly, be evident in the first months of life because an infant is struggling with basic motor skills and/or cognition. This is deeply distressing for new parents...'*

for new parents, who may come to learn early on that their child will not be able to live an independent life or they may have a life-limiting condition, such as spinal muscular atrophy, a rare, genetically inherited neuromuscular condition, causing progressive muscle weakness and loss of movement due to muscle wasting (atrophy). The most common form is 5q SMA, which includes SMA types 1, 2, 3 and 4. For neonatal neurological conditions, detection and early intervention can help to prevent or lessen serious illness in children; early diagnosis and ongoing support are absolutely key. In such cases, it is critical that neonatal services and paediatric services work together to ensure a seamless transition between the two.

To date, screening for rare neurological conditions has not had a role in picking up rare neurological conditions in infancy: there are currently no neurological conditions routinely screened for at birth in the UK[xliv] and, as a whole, the UK screens for far fewer conditions at birth than many other high-income countries (nine conditions in England compared to around 20 or more in much of Europe and up to 50 in the US).[xlv] With the advent of more genetic testing, Alliance members have told us that there should be reform of the Newborn Screening Review Process to reflect advances in genetic testing for rare neurological conditions that are present at or soon after birth, which could greatly improve early treatment options. We were also told by Spinal Muscular Atrophy UK, for example, that too often, parents are not listened to when they have concerns about their infants, even though early drug treatment would be likely to maximise the potential benefits of such treatment.

Ongoing family support, high-quality information for parents, developmental follow-up, physiotherapy and aid with motor problems, feeding interventions, long-term ventilation, control of seizures and multidisciplinary support will, in many cases, be vital services,[xlvi] which should be commissioned by the NHS.

Diagnosis in childhood

Some very rare neurological conditions, such as Dravet syndrome (a rare, drug-resistant form of epilepsy that begins in their first year of life) or Batten disease (a life-limiting genetic disease of the nervous system whose normal age of onset is between five and 10 years), are more likely to become apparent later in childhood. Hereditary rare neurological disorders are, in fact, much more likely to have their onset in childhood than in adulthood.[xlvii] For

6 The age of onset of symptoms of rare neurological conditions

example, approximately 11 to 17 children and young people are diagnosed with Batten disease in the UK each year.[xlviii] A much higher number, around 18,000 children, are known to have rare forms of drug-resistant epilepsy, a matter of deep concern to parents who currently see only limited treatment options for their children.[xlix] The challenges faced by young people with rare neurological conditions can be significant: in the case of Dravet syndrome, children will not only experience difficult-to-control seizures, but this can be compounded by varying degrees of learning disability, autism, mobility problems, speech difficulties and feeding problems.[l] In the case of children who develop ataxia, they will experience impaired coordination of movement and balance and a lack of muscle control during voluntary activity. Children with Friedrich's ataxia (a genetic form of ataxia which causes difficulty with walking, a loss of sensation in the arms and legs and impaired speech), will generally be confined to a wheelchair within 10 to 20 years after the appearance of their first symptoms.[li] This can have a significant emotional impact on the young person and their family.

The right treatment and support can make all the difference; for example paediatric myasthenia gravis (a very rare neurological autoimmune condition which causes muscles, especially in the eyes, mouth, throat and limbs, to weaken after activity), if managed well, can be in remission within 6–16 months. Often the condition is not picked up and managed well, however, leaving many children needlessly spending years with myasthenic weakness.[lii]

Where parents may be aware that their child is developing new and concerning neurological symptoms, their GP should urgently refer them for an immediate assessment by a paediatric neurologist.

The transition to adult services

The transition to adult neurology services for young people can be difficult and our members tell us it is not always as seamless as it should be. Indeed, this was recognised as a priority in the Rare Disease Strategy itself. There needs to be a good 'handover' from paediatric to adult services for young people with rare neurological conditions, including the most basic of things such as the transfer of clinical notes and diagnostic test results.

In their 2014 report *Patient experiences of transition between care providers,*[liii] Rare Disease UK commented on aspects of the transition from Children and Young People's services to Adult Services which were less than satisfactory, including:

- Patients and families feeling disconnected from the transition process.
- Age-appropriate services not always being available to patients.
- Medical professionals receiving insufficient training in adolescent care and medicine.
- Transition coming as a shock to young people and being too sudden for them.
- Parents feeling anxious and unsupported during the transition process.
- A person's individual circumstances not always being considered during transition, i.e. not taking a person-centred approach.

The report concluded that good communication and coordination are essential for successful

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transition to adult services. Services also need to anticipate change, such as a young person needing a wheelchair, to avoid a long wait for such support, which can happen at present.

How far the situation has improved since 2014 is unclear, although in a more recent report from 2019,[liv] Rare Disease UK still reported significant challenges: 'Young people report that they feel 'in limbo' between adult and child services and as though they do not fit in either place. Many feel they would benefit from dedicated teenage or young adult services that would offer a phased entry into adult services.' They also report that young people should be more empowered to make decisions about their own care, a recommendation the Alliance would fully endorse.

'Services also need to anticipate change, such as a young person needing a wheelchair, to avoid a long wait for such support, which can happen at present.'

Diagnosis in adults

While around 70% of rare genetic disorders appear in childhood, some rare genetic conditions and non-genetic conditions develop in adulthood. For example, spinal muscular atrophy type 4 (the least severe form of SMA) generally develops in adulthood,[lv] while symptoms of multiple system atrophy (MSA) (a progressive neurological disorder caused by degeneration of nerve cells resulting in problems with movement, balance and autonomic functions of the body) and progressive supranuclear palsy (a rare and progressive condition that can cause problems with balance, movement, vision, speech and swallowing), usually start somewhere between the ages of 50 and 60, and post-60 respectively, although MSA can begin at any time after 30.[lvi, lvii] Other conditions such as ataxia can develop in both childhood and adulthood.

Post-diagnosis

Following diagnosis of a rare neurological condition, clinical management is normally shared between a regional or national Neuroscience Centre (a hub) and a local hospital (spoke), with routine health care still provided in primary care, although some people with rare neurological conditions are not referred on from secondary care to a specialist centre, which can cause delays in diagnosis and receiving full, multidisciplinary care.

Those with no diagnosis

There is also a group of, mostly young, people who live without a diagnosis, even if they experience diverse neurological symptoms, where genetic testing has failed to identify the cause, sometimes referred to as having a 'syndrome without a name'. This can be really hard for families who have no answer to the question as to why their child has concerning symptoms such as seizures or has physical and/or learning disabilities.

Approximately 6,000 children born in the UK every year with a genetic condition are likely to remain undiagnosed,[lviii] some of them with rare neurological conditions such as Livvy, whose story is told on the next page. People without a diagnosis still need good care and family support and medical professionals need to communicate regularly with patients and their families about what care and support is available to them.

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Livvy's story: Living with an undiagnosed rare neurological condition

Livvy, aged 19, has a rare, undiagnosed, condition. She lives in Brighton with her parents and three siblings. Livvy's mother tells us her story.

Olivia (Livvy) was my first baby. Born on a winter's day with rare London snow, she was perfect. She met all her milestones, crawling and babbling on cue. She was smiley, communicative, a joy. Her severe reflux may have been an early sign, but it was dismissed by the GP as normal. We first had that gut-wrenching sense that she wasn't developing like her peers when she was around nine months.

It was as if Livvy had shut down. I look at the photos of her 'glazed' expression now, but at the time we put that down to an ear infection.

Months followed, an eerie time, knowing something was wrong, but not knowing what. At first, we thought it was glue ear, but grommets did not switch Livvy's communication back on. She wasn't waving, clapping, interacting. A visit to a paediatrician, who told us 'there is something seriously wrong with your daughter', sank us into despair. Months of tests followed for genetic conditions, all resulting in no explanation and a diagnosis of severe autism and severe learning difficulties.

This was a world we became comfortable in. Livvy developed epilepsy aged five, but medication kept her head drops under control and this wasn't unusual in children with autism. But at the age of nine, Livvy's condition deteriorated quite dramatically. We took her and her brother and baby sister to Disneyland in Paris and she stopped walking and eating. Her paediatrician felt it was an 'autistic' shut down, but shortly after we returned, she started having dramatic head drops where she would fall to the floor. I remember holding her all day

Continued on next page

6 The age of onset of symptoms of rare neurological conditions

Livvy's story (continued from previous page)

on the sofa in tears after one drop resulted in bruising to her entire face. The morning after a 24-hour electroencephalogram (EEG) at the Evelina Hospital, the neurophysiologist told me she was 'locked in a world of seizures'. Livvy was having almost continuous epileptic activity including tonics in her sleep, long absences and head drops.

At the age of 19, Livvy has a severe type of medicine-resistant epilepsy, with other complicating symptoms such as scoliosis and autism, but no specific diagnosis, and she is on a range of medication including Epidiolex, the cannabis-based drug. She has daily seizures now, including tonic-clonic seizures, (where a person loses consciousness, their muscles stiffen and jerking movements are seen), severe scoliosis, has lost her swallow and is now fed via a gastro jejunostomy (a stoma). She can walk a little bit and we are clinging on to this skill.

Eighteen years on from that first paediatrician's appointment, Livvy has been tested for a myriad of conditions that cause her relentless epilepsy, but we still don't have an answer as to why. We know it is probably 'de novo' as her three siblings are unaffected. We are waiting for the results of the 100,000 Genomes Project tests, which we joined in early 2017, but maybe her condition hasn't been discovered yet?

I left my career as a journalist to work in the voluntary sector and now run a rare disease charity, The Batten Disease Family Association. I am struck every day by the courage, strength and camaraderie the community is able to offer to each other, despite the devastating challenges they face. A diagnosis won't change Livvy – she is the incredible person she is – but maybe it would give us (and her) a sense of connection and a better understanding of how to give her the very best quality of life we can.

7 Problems with getting a diagnosis – what are the underlying reasons?

The NHS Implementation Plan did not pull its punches when it came to identifying the problem of delayed diagnoses for people with rare conditions.

'On average, patients with a rare disease will consult with five doctors; receive three misdiagnoses; and wait four years before receiving their definitive diagnosis. Delays in diagnosis mean that opportunities for timely interventions can be missed; conversely, patients may be given inappropriate or harmful treatments if they have been given the wrong diagnosis. Timely diagnosis also allows for the making of reproductive choices by couples when a disease is known to be heritable.'

THE NHS IMPLEMENTATION PLAN, 2018

'Getting a correct diagnosis reduces the time that patients and families have to spend searching and visiting a multiplicity of healthcare professionals. Improving diagnosis means patients and families can be signposted to services that are expert in the care of their disease. Other agencies such as education and social services can be reluctant to commit to specific interventions without a diagnosis.'

THE NHS IMPLEMENTATION PLAN, 2018

7 Problems with getting a diagnosis – what are the underlying reasons?

In 2004, a survey of eight rare conditions revealed that 25% of patients said there was a gap of between five and 30 years between first symptoms appearing and diagnosis,[lix] a long journey the NHS later referred to as the Diagnostic Odyssey[lx] for people with rare conditions – with all the frustration, upset, exhaustion and potential deterioration in health this can entail. Although the extreme end of that wait for a diagnosis has, thankfully, come down, there is still an unacceptable delay for some people with rare neurological conditions in receiving a diagnosis.

In 2008, research quoted in the Rare Disease Strategy[lxi] identified five aspects of diagnosis that are particularly difficult for GPs, which may occur together, compounding the problem of making an initial diagnosis:

- Atypical presentations
- Non-specific presentations
- Very rare conditions
- Comorbidity (more than one disease present)
- Perceptual features that could be missed

Rare neurological conditions may sometimes present with a collection of seemingly unconnected, non-specific, symptoms and to a GP who has not seen many rare conditions in their career, it is perhaps understandable – especially in the current climate of downward pressure on referrals[lxii] – that the message to parents and adults may sometimes be 'go away and come back if it the symptoms don't improve.' People presenting with complex/unusual symptoms may also be referred to specialists other than neurologists. The PSPA (Progressive Supranuclear Palsy Association) reported to the Alliance in our recent survey of September 2020, that misdiagnosis is common for people with this condition and referral to specialists other than neurologists is very common. This leads to variations in care across the country. Parkinson's UK have also told us that some healthcare professionals don't immediately think a condition may be Parkinson's due to the age profile of the condition as most people are diagnosed when they are over 60. This, again, results in people with the condition taking a long time to be diagnosed and, even then, the care offered may sometimes be inappropriate as people are directed towards services for older people.

Some symptoms will be more obvious, of course, such as loss of gross motor skills, so it is clear a child needs to see a specialist, but with other conditions such as late onset ataxia, for example, the symptoms are less easy to pin down. Underpinning the situation is an acknowledged lack of knowledge and training amongst GPs about neurological conditions, especially rare neurological conditions. There have been attempts to address this in recent years, including through e-learning resources made available by the Royal College of General Practitioners,[lxiii] but the neurology learning tools do not tend to focus on rare neurological conditions – this needs to be addressed.

One of the key objectives of this report is to spread awareness in primary care about rare neurological conditions – which, as mentioned above, affect many thousands of people in England today. They are not rare taken as a whole and their rare status needs to be demystified.

The NHS itself suggested in its Implementation Plan of 2018 that GPs could potentially be provided with a series of prompts to help them diagnose a rare condition, for example via an online algorithm. Whatever the mechanism, GP should have access to some form checklist of symptoms often seen in rare neurological conditions such as:

7 Problems with getting a diagnosis – what are the underlying reasons?

- Loss of motor skills
- Uncoordinated movements
- Seizures
- Visual disturbance
- Respiratory problems
- Fatigue
- Learning difficulties

Appendix A to this report could act as a further short information aid for GPs.

In our recent survey of 10 charities representing people with rare neurological conditions, carried out in September 2020, four in 10 organisations said most of their beneficiaries with rare neurological conditions waited an average of three to five years before receiving a diagnosis and seven in 10 said little progress had been made in speeding up diagnosis and early intervention.

Even once a person has been referred for a specialist opinion, the lack of established pathways to achieve a diagnosis, including patchy access to genetic testing, means the difficulties do not always end in primary care. For example, we were told by Ataxia UK that, while around one third of ataxia patients attend specialist ataxia clinics where they receive good or excellent care from neurologists with expertise in the condition, two thirds of people with ataxia are more likely to be seen by general neurologists, the result being that they tend to receive less expert care. The same applies to people with epilepsy, who are often referred to generalists with no experience of genetic/complex epilepsies, underlining the lack of specialist service provision for this group,[lxv] an issue which needs to be addressed, given that rare epilepsies make up a significant proportion of the whole epilepsy population.

An Association of British Neurologists (ABN) Specialist Advisory Group identified the following roadblocks to better treatment for people with rare epilepsies:[lxvi]

- The lack of knowledge of rare conditions amongst referring doctors and some neurologists.
- Shortages in the specialist neurology workforce.
- Inadequate commissioning structures for specialised services.
- A need to level up services for patients, i.e. variation in access to services.
- A lack of treatment guidelines.
- Underfunding of neurology services.
- Loss of access to trials or orphan medications and/or medication with restricted indications.

‘Getting a diagnosis was something that really played with my emotions, causing great anxiety and taking up an enormous amount of time.’

Rhys, with superficial siderosis (a disabling neurological condition resulting from chronic bleeding around the surface of the brain and spinal cord), Rare Disease UK website[lxiv]

7 Problems with getting a diagnosis – what are the underlying reasons?

The first three of these factors were also mentioned by specialists working in acute neurology services.

It is widely felt, including by patient groups, that there needs to be more involvement of neurologists at the 'front door' of the NHS. We were, for example, told by the ABN:

> *"From an acute neurology perspective, the advantage of having neurologists nearer to acute presentations is that this may increase the likelihood of a rare neurological disease being recognised and diagnosed sooner."* [lxvii]

It should also not be forgotten that there are long waiting lists to see a neurologist, made worse by the COVID-19 pandemic. By July 2020, there was already an anticipated backlog of 227,000 neurology and 58,000 neurosurgery appointments,[lxviii] and with a second wave of the pandemic this Autumn, this situation is likely to deteriorate further.

There is, however, some hope for improvement on the horizon once people are referred to regional Neuroscience Centres. The advent of more genetic testing for rare neurological conditions and whole genome sequencing, mean that new opportunities are becoming available to more rapidly diagnose and potentially treat, rare genetic neurological conditions.

Sequencing of an individual's genome is also increasingly utilised as a diagnostic tool for children and adults with unrecognised signs and symptoms and to support diagnosis of a rare disease; around 25% of patients sequenced through the 100,000 Genomes Project received a diagnosis for the first time through the programme. In Appendix A of this report we set out some of the rare neurological conditions for which there is now a genetic test, a list that is growing all the time. The challenge is now to make high quality diagnostic testing accessible to all who consent to it, through common, clinically agreed systems or pathways which become 'business as usual.' Traditionally, NHS Trusts have been reluctant to fund genetic testing because of the cost, but increased central NHS funding for genetic testing should improve this situation.

Finally, protocols need to be in place to identify patients with no diagnosis and the standards of care they can expect, ensuring that a lack of diagnosis does not create a barrier to treating their symptoms.

7 Problems with getting a diagnosis – what are the underlying reasons?

Bradley's Dad's story: progressive supranuclear palsy (PSP)

We knew something was wrong with my Dad John's health when he began to have balance problems during the summer of 2016. He was falling backwards a lot and quickly progressed to using walking aids from 2017 onwards, in order to still get about.

At first Dad used a walker, but after 12 months, and still being unsteady on his feet, he started using a wheelchair in 2018, after a bad fall resulted in multiple bone breaks and a stay in hospital. As his symptoms progressed, Dad lost the strength in his arms and well as the ability to eat standard meals. Mum was his main carer and he largely stayed at home. But he was a very proud man, so refused to have a PEG fitted, and since he had problems eating, he lost around four stones during his illness.

When Dad first started to be poorly, my parents were living in Essex. Unfortunately, due to the rareness of PSP, the healthcare professionals really struggled to diagnose what was wrong. Eventually, following multiple investigations, they discharged Dad, saying there wasn't any more they could do to help. Needing an answer, my parents paid to see a private neurologist who suspected multiple system atrophy.

My parents then moved to Devon to be closer to my sister, and Dad's healthcare really improved there. He had regular physio appointments and they tried a couple of different medications. Unfortunately, nothing seemed to help, and Dad's symptoms just got worse.

It was in Devon, doctors started to mention progressive supranuclear palsy (PSP), but Dad's diagnosis wasn't fully confirmed until he passed away from a pulmonary embolism in May 2020, after they had performed a post-mortem.

Our thanks to PSPA for allowing us to use this case study.[lxix]

8 Specialised commissioning for rare neurological conditions – how does it work?

People with rare neurological conditions may require support in many forms, including therapies, rehabilitation and mental health support. In terms of actual therapeutic interventions, only 5% of all rare genetic conditions currently have a suitable treatment option available, which will be based on an individual's genetic signature and other clinical characteristics. However, this situation is beginning to change rapidly as set out in Section 13 below.

Specialised NHS services support people with rare and complex conditions, including rare neurological conditions. Specialised commissioning for neurology funds all specialised and non-specialised in-patient activity at designated Neuroscience Centres, as well as out-patients seen at those centres referred in by consultants. There are 24 Neuroscience Centres in England. Some highly specialised services, including those for very rare neurological conditions, are only provided at a very small number of national centres across the country, making access difficult for people who live a long way away. You can find a list of the 24 Neuroscience Centres in Appendix C.

Within the 146 specialised services commissioned by the NHS,[lxx] including rare surgical procedures, there is a further subset of services classified as 'highly specialised' – around 70 services in total. Each highly specialised service is provided to a much smaller number of patients compared to specialised services, usually no more than 500 patients a year. Nine highly specialised neurological treatments are nationally commissioned in this way, including treatments for a rare form of ataxia, for neurofibromatosis (a genetic disorder that causes tumours to form on nerve tissue, including in the brain and spinal cord) and for rare neuromuscular disorders. The conditions and treating centres for those very rare neurological conditions are set out in Appendix B.

The Getting it Right First Time (GIRFT) neurology programme has unearthed significant variation in how neurology services are commissioned across the country.[lxxi] For example, the cost of neurology services varies from £4.80 per head to £17.10 per person.

"Nine highly specialised neurological treatments are nationally commissioned in this way, including treatments for a rare form of ataxia, for neurofibromatosis (a genetic disorder that causes tumours to form on nerve tissue) and for rare neuromuscular disorders."

The highest spend is in the region based at the National Hospital for Neurology and Neurosurgery, a highly specialised centre, which has a high spend on in-patient elective activity. For most regions, the main variation arises from the differing spend on non-elective neurology services, which varies between £0.90 per head and £5.10 per head.

8 Specialised commissioning for rare neurological conditions – how does it work?

Neuroscience Centres typically operate on a hub and spoke model, with referrals coming from the surrounding District General Hospitals (DGHs), who may provide ongoing care through specialist clinics. For example, the specialist Walton Centre in Liverpool has been operating a hub and spoke neurology model for over 10 years with the Centre acting as the main hub, supporting spokes or satellite sites in local acute hospitals. Neurologists from The Walton Centre work in the satellites for typically four days a week (normally two days per consultant), holding outpatient clinics and undertaking ward consultations. All have sub-specialisms and for the rest of the week are based at The Walton Centre.[lxxii] Typically, more care for rare neurological conditions is delivered by the hub.

The Neuroscience Service Specification

In 2013/2014, the Interim Specialised Neurology (Adult) Service Specification[lxxiii] was published to outline standards of care for people with complex single need or multiple neurological conditions delivered via specialist services, using multidisciplinary teams and with some disease-specific protocols and specified pathways. The Neurology Service Specification applies to both more common and rare neurological condition groups:

- Inflammatory disorders of the nervous system
- Epilepsy and related disorders
- Movement disorders
- Neuromuscular conditions
- Inherited neuromuscular disorders

Acquired disorders include:

- Stroke
- Motor neurone disease
- Neurogenetic diseases
- Neuro-oncological diseases
- Specialist clinic for cognitive disorders

8 Specialised commissioning for rare neurological conditions – how does it work?

The aims of the Specialised (Adult) Neurology Service Specification were to:

- Improve specific clinical outcomes.
- Improve the quality of Neurology services and the patient experience.
- Reduce the number of unscheduled admissions and re-admissions to hospital with neurological conditions.
- Reduce hospital lengths of stay.
- Improve referral and signposting to appropriate services for patients, including voluntary agencies and community groups.
- Improve transitional care.
- Increase patient choice.
- Reduce inequalities in health between those with neurological condition, both across the regions and with other parts of the UK.
- Facilitate future research into these conditions, through 'academic neurology', research led by specialist nurses and other health professionals, and research into best ways of providing services.
- Facilitate training of future consultant neurologists in these conditions, and of specialist nurses and allied health professionals essential for the care of patients.

Its objectives were to:

- Provide improved quality of life and patient experience for patients and their carers.
- Provide a service accessible for individuals and their families, ensuring standardisation of care across the specialist centres.
- Offer the best services within the resources available.
- Provide high quality care for all individuals with smooth transitional care for young adults and effective support for families.
- Provide care as close to home as possible, with regular reviews by local service providers, with specialist centres acting as coordinators to ensure appropriate care plans are in place for communication and whole system management of patients.
- Specialised services will be available within each NHS region to support local provision where appropriate.
- Meet national standards and audit services to document that the best possible outcomes and patient experience are delivered.
- Ensure a patient-centred approach to the delivery of services.
- Improve access to services in primary care through referral to more integrated services, particularly for disadvantaged groups and geographical areas.
- Encourage a range of approaches for case- management.
- Encourage greater participation in self-care through appropriate support mechanisms.
- Improve the availability of clinical and non-clinical information.
- Use innovative technologies to enable those living remote from neurology centres to receive the same care as those living close by.
- Enable advancement of future care through research and enhanced training.

8 Specialised commissioning for rare neurological conditions – how does it work?

The Neuroscience Service Specification is in the process of being revised. This report hopes to tackle the weak links in the chain that prevent the best possible care from being delivered, which, as is clear from our Patient Experience Survey 2019, is still not happening consistently. More generally, there is an NHS review underway of how Service Specifications are developed,[lxxiv] while new pathways are in the process of being finalised for specific neurological conditions which we will discuss in Section 11 below.

The Neurosciences Clinical Reference Group (CRG) covers specialised neuroscience services and specialised adult neurosurgery. Membership includes clinicians and two patient and public voice representatives, including one from the Neurological Alliance. The treatment recommendations issued by the CRG are used as a basis for regional commissioning of specialist neurology services and/or treatments, some for rare conditions. It is diverse in format – some for procedures, some for therapies – and activated through different NHS mechanisms, painting a somewhat confusing picture. The specialist neurological treatments recommended by the CRG are listed in Appendix D of this report.

The Rare Diseases Advisory Group (RDAG) is responsible for making recommendations to NHS England (and the devolved administrations of NHS Scotland, NHS Wales and NHS Northern Ireland) on the development of services for people with rare conditions and on highly specialised services. In turn, RDAG receives recommendations from Clinical Reference Groups (CRGs) set up by NHS England.

In our recent member survey, some organisations told us that developing new service specifications for rare neurological conditions which do not have them, or potentially groups of neurological conditions, for example all spinal cord inflammatory conditions, could be helpful. Developing service specifications, even for conditions with small population numbers, could improve precision, helping to raise quality standards and outcomes.

> *"The approval of treatments, the commissioning of neurology services and the mechanisms used to recommend specific treatments…need to be streamlined, simplified, and made more transparent and easier to understand."*

The commissioning and service provision landscape for rare neurological conditions is complicated which, in turn, reinforces the perception that rare neurological conditions represent an area of medicine that is complex, both from the viewpoint of clinicians, the voluntary sector and people with neurological conditions and their families. The approval of treatments, the commissioning of neurology services and the mechanisms used to recommend specific treatments, e.g. policies routinely commissioned or policy statements, need to be streamlined, simplified and made more transparent and easier to understand.

9 Delivering integrated, personalised and coordinated care – the essential elements

The NHS Long Term Plan of 2018 sets out a commitment to deliver personalised care for people using NHS services. Never could the need to deliver personalised, holistic care be more important than for people with rare neurological conditions, each with their own symptoms, personal and family circumstances and journey to diagnosis, and with sometimes only a very small number of people living with a particular condition.

i. Integrated commissioning of care – seeing the bigger picture

The importance of delivering integrated care is often cited, but what does it really mean in practice, and how can it be achieved for people with rare neurological conditions? At its heart it is about joining up the key elements of care and treatment so that the care pathway is not confusing or fragmented, ideally underpinned by joined-up commissioning of services, including mental health support and rehabilitation. The Integrated Commissioning for Better Outcomes Framework[lxxv] is a practical tool for NHS Commissioners and local government to support improving outcomes through integrated commissioning. Different commissioning bodies, care settings and NHS and local authority organisations, need to work together collaboratively and effectively to provide the right care, in the right place, at the right time for the people that need it. Even with the advent of Integrated Care Systems (ICSs), integrated commissioning of services is not yet widespread on the ground in England and integrated commissioning of specialised services with other elements of care, such as mental health and social care, may present particular challenges, especially as, in practice, social care is often provided by the private sector.

Integrated commissioning for people with rare neurological conditions relies, at its heart, on an accurate appraisal of the services people need, gained through good data collection and data sharing, which we discuss in Section 12. Without knowledge of the incidence and prevalence of different rare neurological conditions, or knowledge of the outcomes and experiences associated with good care, NHS Commissioners simply cannot adequately commission the specialised services required. It is of course critical that the experiences and needs of people with rare neurological conditions are central to commissioning and service improvement – that means a commitment to, and resourcing of, patient and public involvement through ICSs and other NHS structures. Too often, this just doesn't happen.

ii. Delivering personalised care – respecting individuals and their families

The key elements of personalised care include:

- Providing individuals and/or their families with information about their health condition and care and treatment options, particularly at the point of diagnosis when they may be struggling to understand the implications of their diagnosis for their lives and what they can expect in terms of service provision.
- Communication that is accessible – i.e. in a format that corresponds to an individual's needs and which is two-way, to enable the individual to express their needs and preferences.
- Shared decision-making on an individual level about care and treatment choices, as well as opportunities for individuals/families to participate in planning and making decisions about their service provision (co-production).

9 Delivering integrated, personalised and coordinated care – the essential elements

- Care planning, to enable people/their families to record their priorities and preferences for treatment and care, to facilitate self-management where this is possible, to enable future scenario planning, and generally to help people to be, and feel, in control.

- Care that is well coordinated between the different professionals and agencies involved in delivering an individual's care.

National Voices, a coalition of health and care charities, recently published a series of I-Statements that reflect the most common wishes and expectations people have about their condition when they were first diagnosed, as part of their *What We Need Now* report.[lxxvi] These wishes included being listened to, knowing what to expect from treatment and being supported while waiting for treatment.

In our recent survey of 10 charities representing people with rare neurological conditions carried out in September 2020, half of organisations reported that their beneficiaries do not receive adequate written or other information from the NHS at the time of diagnosis.

iii. Providing the right information at the right time – helping people to understand their choices

The Rare Disease Strategy 2013 states: 'For a patient to make informed choices they must have access to reliable, correct information about their condition,' yet our Patient Experience Survey 2018/19 found that only 34% of people with rare neurological conditions were provided with written information about their care.

Best practice also indicates that every person with a rare neurological condition and/or their families should have a written care plan, co-produced by them with their specialist treating clinician, where possible, setting out clearly the treatment they can expect, including:

- The timing of any regular reviews.

- What therapies they are being prescribed (if any).

- What other allied health services will be involved in providing care and where, e.g. psychological support, rehabilitation or speech and language therapy.

- Sources of information and support, including the voluntary sector.

- Who to contact in an emergency, e.g. a specialist neurology nurse.

- The name of the person coordinating their care, who might also be their specialist nurse or another health professional.

This information should be available in both written form and digitally, i.e. so it can be downloaded onto a smart phone or laptop. This might seem like an unnecessary investment in valuable time and resources, but evidence suggests that people/families who are better supported to manage their health, through tools such as care plans, are less likely to require unscheduled NHS care.[lxxvii] Improving the quality of health information available to individuals and their families, as mentioned, also allows people with rare neurological conditions to feel more in control of their care, although as some conditions progress, this becomes more challenging and people may need to be supported in the decisions they are able to make.

9 Delivering integrated, personalised and coordinated care – the essential elements

iv. Good care planning and coordination – essential for high-quality care

The Rare Disease Strategy 2013 clearly summarises the care planning challenge:

> *"Following diagnosis, a patient should have an evidence-based care plan that identifies the anticipated course of their condition and sets out the responsibilities of specialist, general and primary care services in care management. Good communication between patients, their families and professionals is essential, to ensure that the primary care plan is agreed and the care team has information and appropriate specialist support. The ultimate aim will be to ensure that the agreed care plan is delivered effectively."*

Good care planning, in addition to having a written care plan, helps people with rare neurological conditions and their families to negotiate the many different transitions and boundaries involved in their care and support, necessitating strong communication between services and good record sharing. These transitions/boundaries include:

- From children and young people's services to adult and older people's services, as mentioned above.
- Between primary, secondary and tertiary care.
- Between specialist care and other care providers, e.g. rehabilitation and mental health services.
- Between health care, social care and voluntary sector support.
- Between different medical disciplines, where a person with a rare neurological condition has other long-term conditions (comorbidities).
- Between NHS providers, the Genomic Medicine Service, the Genetic Testing Service, researchers and academics.

In our recent survey of charities representing people with rare neurological conditions carried out in September 2020, nine out of 10 organisations said that the care coordination received by their beneficiaries was not very good or not coordinated at all, while six in 10 said co-produced care plans were not widely used.

Delivering joined-up care is greatly helped by having a named care coordinator; this could be a specialist nurse, or even someone in a dedicated care coordinator role, for example, an allied healthcare professional who has responsibility for ensuring that a person, once diagnosed, is linked to all the health and care services provided

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by their multidisciplinary team, in addition to social care provision by local authorities or other agencies. In their 2013 report, *Rare Disease Care Coordination: Delivering Value, Improving Services,*[lxxviii] Rare Disease UK found that having a professional in post who can fulfil the care coordinator role helps to improve the quality of care and patients' experience of care by:

- Helping patients receive timely access to the specialist knowledge and care they require.
- Meeting patients' information needs.
- Providing emotional and practical support to patients and their families.
- Providing a continuing point of contact.

There is strong evidence that having dedicated care coordinators represents good value for money for service providers, through saving consultants' and GPs' time and helping to prevent unplanned hospital admissions.[lxxix] The South West London Neurology Network is scoping having care coordinators for rarer neurological conditions which could be a model followed by other regional providers.

In this context, The Alliance supports the concept of the new rare disease 'insert' in NHS England's contracts with care providers delivering specialised services to patients with rare conditions, which became operational in 2018/19. The focus of this insert is on improving care coordination around three separate criteria:

- Ensuring that there is a person responsible for coordinating the care of any patient with a rare condition.
- Giving every patient with a rare condition an 'alert card' which includes information about: the patient's rare condition; any particular aspects of the treatment of that rare condition that need to be taken into account in providing care to that patient; and details of how to contact an individual specialist involved in that person's care.
- Ensuring that every paediatric patient with a rare condition has an active transition to an appropriate adult service, even if that adult service is not the commissioning responsibility of NHSE.

Unfortunately, it does not appear from speaking with clinicians in preparing this report, that these rare disease 'inserts' in provider contracts have made any tangible difference.

NHSE is also exploring with the Royal College of Emergency Medicine how rare condition alert cards could be clinically recognised in A&E departments,[lxxx] something supported by Rare Disease UK.[lxxxi] The Alliance would fully endorse this measure so clinicians in A&E Departments are aware that someone presenting for care has a rare neurological condition. This could save much anxiety and inappropriate treatment and will be particularly important when new genetic therapies become available for patients. Having more neurologists in acute care would also help rare conditions to be picked up at this stage.

For example, most families will present at A&E when it comes to first seizures in infants and children; with a prolonged seizure, clinicians in A&E need to be aware that this it is not just a febrile convulsion: this should be reflected in appropriate NICE guidance.

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v. Engaging people with lived experience

Engaging people with their care and giving them more control is a key theme of the NHS Long Term Plan. It states: 'since individuals' values and preferences differ, ensuring choice and sharing control can meaningfully improve care outcomes.'[lxxxii] It is important that people with rare neurological conditions and their families also feel this sense of empowerment, engagement and shared control over their care and choices. The first of the 51 commitments in the Rare Disease Strategy was 'To strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners.'[lxxxiii]

In our recent survey of charities representing people with rare neurological conditions carried out in September 2020, five in 10 organisations reported that their beneficiaries felt they had only poor control over their care and treatment.

When it comes to the specialised commissioning of services, the 2017 'Framework for patient and public participation in specialised commissioning',[lxxxiv] sets out how patient and public members are included in their governance structures, for example through stakeholder forums such as Clinical Reference Groups (CRGs), the Rare Diseases Advisory Group (RDAG) and the Clinical Priorities Advisory Group (CPAG), which all have lay member/patient and public voice (PPV) partner representation. Appendix A of the above Framework contains a useful organogram of all the decision-making bodies involved in specialised commissioning and the number of PPV representatives in each case.

We cannot cover in this report all the specialised commissioning mechanisms and how patients with lived experience can be involved in these forums – the very complexity and multiplicity of organisations suggests this is also difficult

'It is important that people with rare neurological conditions and their families also feel this sense of empowerment, engagement and shared control over their care and choices.'

for lay people to navigate. However, rare neurological conditions, which may account for up to one in three of all rare conditions, should be better represented at the top decision-making table. Although challenging, it would also be a positive step forward if more people with lived experience could be present in such forums, as well as the voluntary sector groups representing them, which could be facilitated by remote attendance at such meetings. Rare Disease UK's Patient Empowerment Group (PEG) [lxxxv] is a collective of patient representatives who have helped to monitor and take forward the implementation of the UK Strategy for Rare Diseases, with several members from the rare neurological condition community represented.

The Rare Disease Strategy said: 'Patient groups have an important role in helping patients and their families feel less isolated.' However, the COVID-19 pandemic has left many voluntary sector organisations, including neurological charities, badly affected;[lxxxvi] nonetheless, they are still trying to provide vital support to

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their beneficiaries, some of whom may be experiencing isolation and loneliness as a result of restrictions placed on them by the pandemic, or a loss of access to services for the same reason. This vital work must continue and Government should be actively putting in place measures to financially support voluntary sector organisations, which take so much pressure off the NHS and which are such valuable sources of condition-specific knowledge and expertise.

Finally, the development of digital tools for people with rare conditions, including rare neurological conditions, should be explored, to help with self-management – where this is possible for the person with a rare neurological condition – and to provide support, supplemented by assistive technology where necessary. Such platforms may also be a suitable way of collecting and understanding people's experiences of care and support, and their health outcomes. Digital decision-making support tools were recently cited by NICE in their response to the Independent Medicines and Medical Devices Safety Review, as a way of ensuring better patient engagement in treatment plans and medication decisions. This could be important for some people with rare neurological conditions too.[lxxxvii]

Whilst apps might not be a suitable tool for everyone with a rare neurological condition, self-management apps could be made available to people with rare neurological conditions via the NHS Digital Apps Library. There may be apps that are specific to individual rare conditions or generics apps, for example, for parents or children who have a rare condition. Apps could also be helpful in providing information for clinicians who do not normally treat people with rare conditions, for example in primary care, something advocated in the NHS Rare Disease Implementation Plan.[lxxxviii]

'The Rare Disease Strategy said: 'Patient groups have an important role in helping patients and their families feel less isolated.' However, the COVID-19 pandemic has left many voluntary sector organisations, including neurological charities, badly affected; nonetheless, they are still trying to provide vital support to their beneficiaries, some of whom may be experiencing isolation and loneliness as a result of restrictions placed on them by the pandemic, or a loss of access to services for the same reason.'

10 Mental health support – an integral part of providing holistic health care

Living with a rare neurological condition can be extremely challenging. Children, young people and adults risk being misunderstood by society at large because of their symptoms, for example difficulty in communicating and movement disorders, and their prospects are uncertain or they may, in the worst-case scenario, be life-limiting.

Until very recently, unlike conditions such as most cancers, there were very few therapies available for people with rare neurological conditions, denying people with such conditions and their families the hope of recovery, with all that means for their psychological well-being. Coping with this immense psychological challenge can require specialist mental health support. The Neurological Alliance recognises the great resilience of people living with rare neurological conditions, and that of their families.

As mentioned in Section 4, the Alliance's Patient Experience Survey 2018/2019 revealed that the mental health of people with rare conditions is generally worse than that of people living with more common conditions, which is, itself, not always good. For example, when people with rare neurological conditions in our survey were asked whether their mental health had been discussed with them by a health or social care professional, whereas this was the case for 44.7% of respondents with more common neurological conditions, it was only the case for a third (33.1%) of people with rare neurological conditions Likewise, when asked whether they felt their mental wellbeing needs were being met, only 31.5% of people with a rare neurological condition agreed with this statement, while the figure was 37.6% for more common conditions – quite frankly, very low percentages for *all* people with a neurological condition.

Responses to mental health questions in our Patient Experience Survey 2018/2019	Non-rare conditions – percentage of respondents who agreed with the statement	Rare neurological conditions – percentage of respondents who agreed with the statement
Q. Have you been asked about your mental wellbeing by a health or social care professional?	44.7%	33.1%
Q. To what extent do you feel your mental wellbeing needs are being met?	37.6%	31.5%

Table 2 – Extract of results from Neurological Alliance Patient Experience Survey 2018/2019 related to mental health: non-rare vs rare conditions

10 Mental health support – an integral part of providing holistic health care

If less than a third of people with a rare neurological condition feel that their mental health needs are being properly addressed, this would indicate a serious failing in NHS health care provision, if these findings reflect the overall national picture.

In our recent survey of 10 charities representing people with rare neurological conditions carried out in September 2020, **all 10 organisations** reported that the mental health needs of their beneficiaries were 'not being met very well' or were 'not being met at all'.

Rare Disease UK published a report on mental health in people with rare conditions in 2018,[lxxxix] whose main findings were that:

- Living with a rare condition can have a huge impact, including causing anxiety, stress, low mood, emotional exhaustion, and even suicidal thoughts.
- Many of the drivers of poor mental health reflect issues that are specific to managing a condition that is rare, with patients/carers facing challenges at many points during their journey from the onset of symptoms.
- Patients and carers can experience not being taken seriously by healthcare professionals, and sometimes even being misdiagnosed with psychiatric illness, when trying to access support for their physical condition.

Clearly, mental health care and support cannot simply be an add-on extra for people with rare neurological conditions. However, too often people with rare neurological conditions are passed from pillar to post for mental health support, because they are not deemed to be the medical team's responsibility. Neuropsychology or neuropsychiatry services must be commissioned to meet the overall care needs of the rare neurological community and fully integrated with the specialised commissioning of neurology services. Neurological disorders are conditions like any other, although particularly challenging where they are progressive, and adequate mental health support must be made available to the people that need it; this cannot simply be left to chance or good intentions.

The Alliance fully supports Rare Disease UK's three key recommendations on improving mental health care for people with rare conditions:[xc]

- Healthcare professionals should be provided with the skills, knowledge and capacity to demonstrate awareness of the emotional challenges of living with a rare condition and handle discussions about mental health sensitively.
- Patients and carers should be routinely signposted to sources of mental health support.
- Rare condition services should include an assessment of individuals' mental health needs, extended to family and carers.

The National Neuroscience Advisory Group (NNAG), a multidisciplinary group of organisations committed to transforming neurological treatment and care, recently set out its own recommendations for transforming mental health support for people with neurological conditions in a report published in July

10 Mental health support – an integral part of providing holistic health care

2020.[xci] This includes specific recommendations on the need for all condition-specific and general neurological clinical guidelines to include comprehensive and explicit guidance to address the mental health needs of people with a neurological condition. It also supports developing the concept of a 'brain workforce' to address the ongoing competencies, curriculum, training and recruitment requirements of the breadth of health and social care professionals involved in the care and support of people with neurological conditions and comorbid mental health needs.

Sam's story: Huntington's disease

Sam is 54 years old and has Huntington's disease, a rare condition affecting approximately one in 10,000 people. Sam's story is one highlighting the lack of awareness and knowledge of Huntington's, the poor availability of care services and poor coordination of physical and mental health care services.

Sam was living at home with his wife, but after three suicide attempts and short term stays on psychiatric wards that followed, without any follow up from mental health services, he moved to a rehabilitation unit for people with mental health needs. This placement failed due to the lack of experience and training of staff in Huntington's. He was then moved to a Specialist High Dependency Unit, but this was not appropriate as it was too far from his home and his family play an important role in his care and support. He lived at two further nursing homes, but both stays failed, resulting in him being sectioned.

Following the section, he was placed in two psychiatric wards before being transferred back to the home from where the section had happened with no change to his medication or care plan. He was then readmitted to a psychiatric ward. It was agreed that he needed to move to a specialist ward for a full assessment. It took six months for this move to happen. Following the assessment, he was discharged to yet another home. From here, there were three further transfers to hospital and 19 separate incidents raised, including a safeguarding issue against the home. Sadly, following a further hospital admission for pneumonia, Sam has been diagnosed with lung cancer. He is now in a new home where he can be treated palliatively.

Throughout Sam's five previous placements and five stays on psychiatric wards the story has been the same. Failures have happened due to inappropriate placements, poor communication with other professionals and his family and a lack of understanding of Huntington's disease.[xcii]

11 New optimum clinical pathways being developed for neurological and rare neurological conditions

Clearly, knowing what care ought to be funded and delivered, based on a comprehensive assessment of the needs of people with suspected and confirmed rare neurological conditions is critical. The development of optimum clinical pathways is therefore of the utmost importance.

As part of both the NHSE Neuroscience Transformation Programme and the work of NNAG, eight optimum clinical pathways are being developed which encompass most common and rare neurological disorders:

- MND and muscular disorders
- Functional Neurological Disorder (FND)
- Headache, migraine and facial pain
- Neuro autoimmune conditions
- Movement disorders
- Traumatic brain injury
- Epilepsy
- MS

On the next page we summarise the key elements of health and social care which need to be commissioned, although each of the above pathways will provide detailed and condition-specific guidance once published.

‘Clearly, knowing what care ought to be funded and delivered, based on a comprehensive assessment of the needs of people with suspected and confirmed rare neurological conditions is critical. The development of optimum clinical pathways is therefore of the utmost importance.’

11 New optimum clinical pathways being developed for neurological and rare neurological conditions

Care should be delivered close to home for people with rare neurological conditions, where this is possible. For example, in paediatric services, outreach clinics held in child development centres in the community can enable families to be seen in one place at one time by the local paediatrician, the regional neurologist and other therapists from both local and regional services. In paediatric care this model has been successful for young people with muscular dystrophy (a group of progressive muscle-weakening and wasting conditions)[xciii] and it could work for other rare conditions, where the symptoms and anticipated challenges are similar.

The use of outreach clinics could also be used more widely in adult services, as envisaged in the Neuroscience Service Specification, which spreads expertise and learning from specialist centres into secondary care, although the organisation of this is challenging and it has not happened widely on the ground. This could be supplemented by virtual outpatient appointments where there is no clinical contraindication to this, to improve access to services for people in remote geographical areas and in traditionally hard to reach groups.

People with rare neurological conditions, in particular progressive conditions, should have access to a specialist nurse, who can be particularly important where anticipatory care is vital to avoid a crisis or hospital admission.

Individual clinicians with a special interest, working as part of a multidisciplinary team, can be very important for spreading best practice on the management of rare or very rare conditions. For example, at UCL/the Royal Free Hospitals in London, a multidisciplinary clinic for patients with Wilson disease (a rare movement disorder) was set up more than a decade ago. All patients are seen by a neurologist, a hepatologist and a clinical biochemist/consultant in metabolic disorders at every appointment, with access provided to liaison psychiatry services, reflecting the need to monitor symptoms and possible complications closely. The Clinic then helped to set up a UK Wilson Disease Network which meets annually and which established criteria for Wilson disease centres of excellence, for example advising the NHS on prescribing guidelines for specific therapies. There are now several multi-disciplinary Wilson disease clinics running across the UK. The Network has also helped to enhance research related to the condition. This is potentially a very helpful model for the treatment of other rare neurological disorders.

‘People with rare neurological conditions, in particular progressive conditions, should have access to a specialist nurse, who can be particularly important where anticipatory care is vital to avoid a crisis or hospital admission.’

Finally, in terms of regular outpatient follow-up, the announcement by NHSE in August 2020 on Implementing Phase 3 of the NHS Recovery post-COVID-19 placed a heavy reliance on Patient Initiated Follow Up (PIFU) for outpatient appointments.[xciv] This requires the right information and support to ensure that people know when to trigger a follow-up: this isn’t always in place and subtle changes in a neurological condition (for example to cognition) are not always evident to the person involved.

PIFU pathways are therefore not appropriate for many people with rare neurological conditions and are not a straightforward solution to efficiency pressures.

12 Securing the right data to support the commissioning of specialist services

Integrated commissioning and care delivery rely on high quality data on the size of the rare neurological condition population, including the prevalence and incidence of different rare conditions. Unfortunately, this is lacking in many cases. In 2015, the Genetic Alliance reported that 'the NHS does not record health information to a sufficient granularity for individual rare diseases to be visible in the data. It is not currently possible to know how many UK citizens are affected by almost any rare or genetic condition.'[xcv] Yet it is only with good granular data that the care and treatment needs of the rare neurological condition population – including neurosurgery, inpatient care and therapy, outpatient appointments, rehabilitation, speech therapy, mental health support and/or social care – can be commissioned and delivered. A lack of data also affects the ability of external stakeholders to advocate for improvements on behalf of their beneficiaries.

However, since the publication of the Rare Disease Strategy, there have been some notable improvements in data collection, in particular, the establishment of the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS).[xcvi] Before the advent of NCARDRS, less than half of the country collected data on congenital anomalies, but from 2017 onwards, Public Health England established national coverage of congenital anomaly registration, which has further expanded to collect data on other rare conditions. By 2018, NCARDRS was collecting data from 567 NHS providers across the UK,[xcvii] supporting research, service commissioning and service evaluation, and providing an information resource for clinicians, to support their clinical practice, and for people with rare conditions and their families/carers.

Individuals can also ask to have their details removed from NCARDRS at any time.

The Multiple System Atrophy (MSA) Trust have told the Alliance that their condition-specific registry now includes over 200 people and they will shortly be using this registry to find out more about the demographics of those on the Registry – a real success story.

> *'...it is only with good granular data that the care and treatment needs of the rare neurological condition population – including neurosurgery, inpatient care and therapy, outpatient appointments, rehabilitation, speech therapy, mental health support and/or social care – can be commissioned and delivered.'*

In 2018, NCARDRS started exploring the feasibility of a patient portal to enable patients to self-register on the service, but it is not clear whether this is operational yet. Improving secure access to patient registries for people with all rare neurological conditions could be very helpful and, in future, include the ability of people with rare neurological conditions to report their outcome measures, which would assist clinicians to better understand the rare neurological conditions landscape and people's needs and experiences.

12 Securing the right data to support the commissioning of specialist services

The Neuro Intelligence Collaborative, in its forthcoming Neurological Intelligence Strategy, should include in its remit the development of specific initiatives to support data gathering and sharing around rare neurological conditions; with the demise of Public Health England, it is very important that this work continues.

Finally, consistent coding of rare conditions is vital. SNOMED CT is a clinical terming language that enables the consistent identification and transmission of clinical information across many clinical settings. The NHS set a target that SNOMED CT would be utilised by all secondary care settings by April 2020. SNOMED CT is expected to be advantageous to both Specialised Services Commissioning and the identification/ reporting of rare diseases as it has the capability for greater clinical granularity than is possible using the existing OPCS Classification of Interventions and Procedures and International Classification of Disease (ICD) clinical codes. The NHS is also looking at using SNOMED and point of care recording to minimise manual data entries and to allow for better sharing of key information to facilitate care coordination.[xcviii]

13 Recent advances in the diagnosis of rare genetic conditions in the NHS in England

Within the NHS there have been significant advances in the infrastructure around genetic and genomic testing over the past five years. When the Rare Disease Strategy was published in 2013 there were 3,200 rare diseases for which the causative gene was known, but new technology such as next generation sequencing (NGS) and falling sequencing costs, mean achieving a DNA-based diagnosis is becoming a reality. The NHS infrastructure is being put in place to have the capacity to deliver more routine genetic testing, which should, in turn, help to improve rates of diagnosis and subsequent care. Genomic medicine is increasingly moving towards the 'business as usual' phase. This should benefit people with rare neurological conditions.

The NHS Long Term Plan of 2019 also made the clear commitment that all seriously ill children who are likely to have a rare genetic disorder, children with cancer, and adults suffering from certain rare conditions or specific cancers, would be offered whole genome sequencing via the new NHS Genomic Medicine Service from 2019.[xcix]

'The NHS infrastructure is being put in place to have the capacity to deliver more routine genetic testing, which should, in turn, help to improve rates of diagnosis and subsequent care. Genomic medicine is increasingly moving towards the 'business as usual' phase. This should benefit people with rare neurological conditions.'

Integrating genomics into NHS clinical pathways really took a leap forward in October 2018, when NHS England launched the NHS Genomic Medicine Service (GMS) to 'embed genomics in NHS care and provide consistent and equitable care'.[c] The service has six key elements:

1. A national Genomic Testing Service delivered through a network of genomic laboratory hubs (GLHs).
2. A National Genomic Test Directory defining the testing available in the NHS in England and the technology to deliver it.
3. National Whole Genome Sequencing provision and a supporting informatics infrastructure developed in partnership with Genomics England.
4. An integrated clinical genetics service that includes genomic counselling for rare and inherited diseases and cancer.
5. Regional infrastructure (GMS Alliances) built on the 100,000 Genomes Project infrastructure, to support the systematic embedding of genomic medicine.
6. A national implementation, coordination and oversight function in NHS England and NHS Improvement (the Genomics Unit).

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The 2018/19 National Genomic Test Directory was also published in October 2018 on NHS England's website and the 2020/21 Version is now available.[ci] It specifies which genomic tests are commissioned by the NHS in England, the technology through which they are available, and the beneficiaries who will be eligible to access a test. This represents another major step forward and the Directory includes tests for conditions such as Huntington's, SMA type 1, hereditary ataxia and many other rare, genetic neurological conditions. In total there are now 532 genetic tests available. The Test Directory will be updated annually, supported by the latest scientific evidence. It is critical that clinicians avail themselves of this knowledge so they can refer people with rare genetic neurological conditions for appropriate genetic testing, which will, hopefully, speed up diagnosis and may open the door to any new genetic therapies where they are available.

"The Genomic Test Directory includes tests for conditions such as Huntington's, SMA type 1, hereditary ataxia and many other rare, genetic neurological conditions."

GMS Alliances together form a new infrastructure being put in place by NHS England and NHS Improvement, to oversee the embedding of genomic medicine within seven newly designated regions: Eastern, Central, North East, North West, Southern, South West and Thames. Each GMS Alliance will be a provider partnership and will establish a network with NHS providers and organisations across their geography. This will involve engagement with Primary Care Networks, Cancer Alliances, ICSs, Academic Health Science Networks and academia.

The GMS Alliances are not just new geographical designations: they will be accountable for achieving demonstrable improvements across their whole geography in the following key areas:

- Equitable patient access to standardised, end-to-end care pathways, including genomic testing, clinical genetics and genomic counselling services.
- Equitable patient access to personalised treatments and medicines optimisation driven by genomic and diagnostic characterisation.
- Systematic consideration of eligibility for clinical trials for patients who would potentially benefit.
- Active participation in the nationally coordinated approach to genomic research and discovery across the country.[cii]

There is also going to be a National Pharmacy Genomics Collaborative network[ciii] to deliver education and training opportunities to support the pharmacy workforce, to realise the benefits of genomics for medicines optimisation and personalisation of treatment interventions, which should benefit people with rare, genetic, neurological conditions where there are therapies for them.

Taking us right up to the present, at the end of September 2020, the Government published *Genome UK: the future of healthcare,*[civ] a new strategy focussing on three key areas:

13 Recent advances in the diagnosis of rare genetic conditions in the NHS in England

- Diagnosis and personalised medicine
  - The NHS England Genomics Medicine Service will now offer whole genome sequencing (WGS) as part of routine care.
  - There will be a new emphasis on pharmacogenomics which will help in the delivery of tailored drug treatments.
- Prevention
  - Early life screening could be expanded, following research into how this might be implemented and an assessment of the risks and ethical issues.
  - Targeted screening, using genomics to improve population health.
- Research
  - Using data to support innovation, while ensuring the responsible use of this data and ensuring diversity and equity of access to research.

The Genome UK report indicates how genomic medicine could be transformed in England, but it must not be forgotten that not all rare conditions, including rare neurological conditions, are genetic, so the new NHS genomic services will not help the approximately 20–30% of people whose rare condition is not due to a genetic mutation, other than to potentially rule this out; for example progressive supranuclear palsy (PSP) is not, generally, a genetic condition. People with non-genetic rare neurological conditions must not be left behind as genomic medicine becomes more mainstream. In addition, whilst having a diagnosis can, undoubtedly, be life-changing if it unlocks access to a new therapeutic treatment, for many, it will not do so. For this majority, all the necessary elements of care must still be made available, including routine care, rehabilitation, psychological support and social care.

Whilst the NHS Genomic Medicine Service is still in its infancy, clinicians have told the Alliance[cv] that centrally organised laboratories and networks of clinicians in tertiary hospitals, in addition to the mainstreaming of genetic testing into specialties, is starting to improve access to genetic testing and diagnosis for affected individuals. The publication of the Genomic Test Directory is also proving helpful for clinicians, as is digital access to the testing laboratories, and portals for accessing genetic test results.

> *‘People with non-genetic rare neurological conditions must not be left behind as genomic medicine becomes more mainstream.’*

However, a note of caution is also needed: genetic screening should, generally, only be carried out at the request of specialised clinical teams in regional neurology centres to avoid excessive testing; most mutations are, in fact not disease-producing so some people may become alarmed without reasonable cause. More widespread testing may also potentially

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overload highly specialised services with uncalled-for test results and lead people being referred to specialist services who do not have a rare condition at all, placing additional demand on already stretched clinical neurology services. Striking a balance will become important.

There are other considerations that will also need to be addressed: genetic test results will need to be carefully explained to patients with adequate time taken to do so; not everyone with genetic variants will go on to develop a condition, as in the case in Huntington's, for example, where each child has a 50% chance of inheriting the mutated gene and developing the condition. Such factors will need to be clearly explained and neurologists will need to be able to access geneticists and genetic counsellors to support those they are treating. It will take time, training, additional staffing – in particular geneticists – and increased resources to bed in these new systems and see them fulfil their potential to improve patient care. Education and training for health professionals will be key.

Informed consent for genomic data collection and handling

The nature of genomic data requires that specific considerations should be thought through for all people having genetic tests and or their families/ advocates. Individuals and families should have to provide informed consent for their genomic data being processed and stored. This is because, in addition to being personal and unique to every individual, genomic data may, for example:[cvi]

- Be stored and used indefinitely.
- Be used to inform individuals about their susceptibility to a broad range of conditions (some of which are unexpected given their personal or family history).
- Carry with it risks that are uncertain or unclear.
- Be reinterpreted and change in relevance over time.
- Raise privacy concerns, in part because of the risk of re-identification.
- Be relevant for family members and reproductive decision-making.

In designing an effective informed consent process for genomics research and/or treatment, researchers and clinicians must consider the information participants and their families will need in order to understand the risks and potential benefits to them of having genetic testing and/or participating in research studies so they are, in all instances, providing their genuine, informed consent. The ethical issues around using genomics in screening to improve population heath must also be understood and agreed by wider society. The Government's new Genome UK report states a key theme of developing new genomic services will be 'engagement and dialogue with the public, patients and our healthcare workforce, placing the patient and the diverse UK population at the heart of this journey.'[cvii] This applies equally to people with rare neurological conditions and their families.

Providing specialist training for neurologists

The DHSC Implementation Plan of 2018 set out the ambition that, by 2020, every clinician should have access to high-quality data and information about rare diseases, including their prevalence and trends, in a clear, accessible and useful format.[cviii] This simply hasn't happened and needs to be picked up in the next Rare Disease Framework.

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However, clinicians will need more than information, they will need training. As rapid advances in technology and understanding mean that genomic testing becomes much more integral to the practice of neurologists, they, in turn, will need to become familiar with the testing process, the implications for people with rare neurological conditions and their families, and how genomics can benefit neurology patients. The integration of neurological and genomic expertise together is essential for making best use of the new technology coming onstream.

Health Education England's Genomic Education Programme has some very useful resources for neurologists about rare genetic neurological conditions.[cix] For example, in September 2020, the Programme posted an article about new genomic research which sheds light on the cause of two life-limiting neurodegenerative conditions, including motor neurone disease, and providing targets for potential therapies and treatments.[cx]

In addition, the Genomic Education Programme provides videos and other learning materials for neurologists online.[cxi] Where there is existing good practice, health systems should look at how this can be replicated through knowledge sharing within the NHS, potentially similar to the Improving Access to Psychological Therapies (IAPT) Webinar series run by NHSEI's Mental Health Team.

Useful resources for clinicians and voluntary sector organisations working with people with rare conditions

Orphanet – a global information portal for rare conditions and orphan drugs which includes:

- An inventory, classification and encyclopaedia of rare genetic conditions
- An inventory of orphan drugs
- A directory of Expert Centres in rare conditions by country, including the UK
- A directory of medical laboratories providing diagnostic tests

UK Pharmascan – a database of information on new medicines, indications, and formulations in the pharmaceutical pipeline.

14 Access to new therapeutic treatments: realising the potential of personalised medicine

We have talked about what personalised care means in terms of people receiving holistic tailored to their needs, but what does personalised medicine mean in terms of accessing personalised therapeutic interventions?

As already mentioned, until recently, only around 5% of rare neurological conditions had a therapy: diagnosis only led to a therapeutic intervention being available in very few cases. However, that situation is, potentially, set to change with the new NHS Genomic Medicine Service as discussed in Section 13.

This is the ambition set out in the NHS's most recent guidance for hospital pharmacists of July 2020:

> *"Personalised medicine will provide opportunities to improve how we treat disease. Based on comprehensive genomic and diagnostic characterisation, different subtypes of patients with a given condition can be identified, and treatment can be tailored to the underlying cause. The involvement of pharmacists and the broader pharmacy workforce will be critical to establishing the integral link between the use and optimisation of medicines and the expression of genomic variants."* [cxii]

Personalised medicine for subtypes of patients includes treatments that fall into the following broad categories, most currently suited for treating rare cancers:

- Gene therapies and advanced therapeutic medicinal products (ATMPs), e.g. CAR-T cell therapy to treat cancer, but also therapies for neurological and autoimmune disorders.[cxiii]
- Targeted treatment where access is based on a genomic test result.
- Histology-independent or tumour-agnostic products, a new class of cancer therapies for tumours expressing a genomic alteration, regardless of where in the body they originate.
- Pharmacogenomic test guided therapy, e.g. fluoropyrimidines for cancer.

However, even once a rare neurological condition has been diagnosed, through genetic or other diagnostic tests, and even if there is a therapy for that condition, personalised, targeted treatments will only become a reality where a therapy and the infrastructure required to deliver the treatment is both approved and funded, which we discuss further in Section 15 on the next page.

15 The funding of treatments for rare conditions: addressing – and overcoming – the barriers

The NHS has a legal responsibility to fund new medicines and devices recommended by the National Institute for Health and Care Excellence (NICE), but in many cases, NICE has been slow to recommend new treatments, particularly for rare conditions where the number of therapies approved has been very limited. Likewise the NHS has sometimes been slow to fund them even once approved, as new NHS services are sometimes needed to deliver them on the ground. Data from the Alliance's Patient Experience Survey 2018/2019 show that respondents with rare conditions were less likely to be receiving treatments for their condition than people with more common conditions, although this is perhaps to be expected as there are fewer treatments available for people with rare conditions.

Currently, for the approval of treatments for more common conditions, and for some rare conditions, NICE Technology Appraisals are carried out, making recommendations on the use of new and existing medicines and treatments within the NHS, following one of the three following routes:[cxiv]

- A single technology appraisal (STA) which covers a single technology for a single indication.
- A fast track appraisal (FTA) which also covers a single technology for a single indication, but with a shorter process time to speed up access to the most cost-effective new treatments.
- A multiple technology appraisal (MTA) which normally covers more than one technology, or one technology for more than one indication.

For very rare conditions on the other hand, a system for the approval for Highly Specialised Technologies (HSTs) for was set up in 2013 by NICE, to decide on access, and funding in principle, for new therapies for people with very rare conditions.[cxv]

> *'Data from the Alliance's Patient Experience Survey 2018/2019 show that respondents with rare conditions were less likely to be receiving treatments for their condition than people with more common conditions...'*

By 2017, following a public consultation, NICE introduced changes to its methods for the evaluation of highly specialised technologies (HST). Under the revised methodology, products that are evaluated through the HST program are assessed against a sliding scale, so the more additional quality-adjusted life years (QALYs) a treatment offers, the greater the funding it will attract, i.e. taking a cost-effectiveness approach (even though some subjective elements still remain in the decision-making process). However, only a very limited number of technologies/ therapies are reviewed through the HST review process each year and only seven pieces of guidance were published in the first five years of the HST programme, what the Specialised Health Care Alliance termed 'a glacial pace'.[cxvi]

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In addition, the current £100,000 – £300,000 spending limit per QALY is arbitrary[cxvii] because it does not take into account the quality of life value of treatments to patients and their families. To date, guidance has only been published on 12 treatments for very rare conditions under the HST scheme, including Ataluren for treating Duchenne muscular dystrophy with a mutation in the dystrophin gene (in people aged 5 and older who can walk).[cxviii]

But what about the children and adults with the rare neurological conditions themselves and their families? They can be caught in the middle of procedural and complex decision-making processes, sometimes as their condition, or that of a loved one, progressively deteriorates. It is to be hoped that a review of the HST process will lead to changes in line with what now happens in Scotland, where a system based around granting conditional access to all new ultra-orphan medicines was approved in 2018.[cxix] Surely this is the only compassionate thing to do.

In 2016, the independent Accelerated Access Review,[cxx] set out a vision for the UK of getting the best technologies and treatments from the lab to the clinic more quickly and cheaply; the Government responded in 2017 with its own report[cxxi] and, in 2018, the Accelerated Access Collaborative was set up,[cxxii] a partnership of government bodies, industry, patient groups and NHS organisations, working to streamline the adoption of new innovations in healthcare, including for people with rare conditions.

NICE is currently undertaking a review of its Methods for approving new treatments,[cxxiii] with a six-week consultation ending on 18 December 2020, but this will not initially cover the HST review process: a separate review will be undertaken of this in 2021. Reform is clearly needed as NICE's Methods and Processes have not kept pace with innovation in rare conditions. For example, in our recent survey of rare neurological members[cxxiv] two organisations commented:

'The NICE appraisal process took far too long (Autumn 2016 – July 2019) depriving people of access to a drug that was being prescribed in the US and later in almost all the rest of Europe during this time – England was one of the last to approve access.'

SPINAL MUSCULAR ATROPHY UK

'The NICE process takes literally years of upset and our families don't have years; two children died waiting to access Brineura.'

BATTEN DISEASE FAMILY ASSOCIATION

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Regrettably, as the Genetic Alliance commented in response to the Government's reply to the Accelerated Access Review,[cxxv] the therapy approvals landscape is complex and there are 15 different routes to approve a licensed medicine in the UK for use in the NHS for a rare genetic condition, eight applying in England; if anything the process has become more complex and drawn out. As mentioned, approval currently relies on a cost-benefit analysis being carried out, but as the Genetic Alliance points out:

> *"Health technology appraisal methodologies consistently fail to take all of the impacts of a rare condition, and consequently the value of the treatment, into account. Part of the problem is the scale of unmet health need. Rare conditions devastate families for generations. Unless we quantify this devastation and consider it to be a cost to us, these treatments will never be considered cost neutral."* [cxxvi]

In our recent survey of 10 charities representing people with rare neurological conditions, carried out in September 2020, we had some very mixed comments about improved access to major new treatments over the past five years:

- Subcutaneous immunoglobulin, funded by the NHS, has helped chronic patients to take control of their treatment (Guillain-Barré & Associated Inflammatory Neuropathies, GAIN).
- Whilst some new medicines are available…a fight for supply often prevents our clients from accessing the best quality medicines (Narcolepsy UK).
- There are no current treatments for progressive supranuclear palsy (PSP) or corticobasal degeneration (CBD) (PSPA).
- Brineura ERT has been funded by the NHS since last September after a very difficult process (Batten Disease Family Association).
- There are numerous treatments on the horizon…advances in research are very much welcomed, but there are no treatments available yet to make a change to people's lives (Huntington's Disease Association).
- Nusinersen/Nusinersen (Spinraza) is effective and funded for many by the NHS, but funding is limited by a Managed Access Agreement which excludes certain groups, in particular many people with SMA type 3. The paediatric roll out of Nusinersen (Spinraza) has been quite good, but the treatment roll out for adults hasn't progressed at all well (Spinal Muscular Atrophy UK).

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Given that diagnostic testing is improving and genetic sequencing is getting less expensive, the likelihood is that a rapidly increasing number of rare neurological conditions will be identified over time. This is crucial to improving the quality of life for people living with undiagnosed conditions and inaccurate (best fit) diagnoses. However, will this technological advance be matched by the development of associated new orphan drugs for people with rare neurological conditions (therapies developed to treat medical conditions so rare they could not be produced without government assistance)? It seems likely as there are currently hundreds of new orphan drugs in the pipeline worldwide, although strained national healthcare budgets could lead to a downturn in new treatments being brought to the market. The key question is to what extent will new therapies be funded by the NHS in England? Put bluntly, will diagnoses accelerate, while the funding for new, approved therapies lags behind? That would be a tragedy for people with rare neurological conditions and represent a poor return on the investment in the NHS Genomic Medicine Service.

To put this in context, a recent study found that while more than half of centrally authorised Orphan Medical Products (OMPs) for rare diseases had marketing approval in the five countries considered in that study, access to patients was then restricted by different national reimbursement policies, including in the UK.[cxxvii] The Association of the British Pharmaceutical Industry (ABPI) likewise points out that, despite the UK's leading position as an innovator in pharmaceuticals, the UK is slow at adopting new treatments. For every 100 patients that get a new medicine in its first year of launch in the EU – including France and Germany – just 21 patients in the UK get access.[cxxviii] That could be due to the different funding model for healthcare in the UK as opposed to much of the rest of Europe, but it is nonetheless concerning.

The Genetic Alliance in its report *Action for Access* published in October 2019,[cxxix] summarised the situation concisely:

> *'Health technology assessments (HTA) decide whether or not medicines will be made available on the NHS. But the path between licence and HTA decision is governed by a system that is difficult to navigate. We have identified five systemic problems: fragmentation, inflexibility, challenges with capturing the value of treatments, delays, and a lack of transparency. But behind these systemic problems are two fundamental problems: uncertainty and money.'*

The same report looked at the situation in Germany for rare condition therapies where those with an orphan designation and a marketing authorisation from the European Medicines Agency (EMA) are simply considered – by definition – to bring added benefit to patients. This is logical as the orphan designation shows that a therapy meets an unmet health need, and the market authorisation indicates that it is an effective treatment. On this basis, the treatment is delivered

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to patients as soon marketing authorisation is granted, providing the revenue for the company remains below a certain threshold. That reflects the situation in Scotland, as referred to above.

It is very expensive for biotech companies to conduct clinical trials, develop new therapies and go through national approval processes. If the bar is set too high for market entry, such companies may decide not to seek entry to national markets if the barriers are too restrictive. This will inevitably affect people with rare conditions. The risks post-Brexit are also significant, depending on the nature of any deal agreed between the UK and the EU.

> *'Denying people with rare conditions access to treatment goes against all arguments of equal access to care.'*

The Neurological Alliance supports the recommendations in the Genetic Alliance's Action for Access report on improving accessibility, through introducing simplified decision-making processes and treatment pricing, which would mean, *'patients with rare diseases will no longer have to wait on average 26 months to be able to access medicines from the point they become available in Europe, and there will no longer be patients whose condition progresses beyond the point of treatment during an HTA decision-making process in the UK.'*[cxxx]

It is to be hoped that the NICE Methods Review will address these issues, in addition to the HST Review, which is likely to be rolled into the second NICE Methods Review consultation in 2021. The implementation of the results of the NICE Methods Review is set for October 2021 and it is essential that it builds in extra flexibility around approving treatments for rare conditions, so they do not fall between the gaps in the current HTA and HST approval processes. To do this, the HTA process needs to acknowledge both the severity of different conditions and where approval should sit. Also, the current criteria for approving an HST may be that it is only delivered at a small number of specialist centres, even if a local hospital could just as easily provide the same therapy. These anomalies must be ironed out.

It is to be hoped that the new £500m Innovative Medicines Fund announced by the Government prior to the 2019 General Election,[cxxxi] to replace the Cancer Drugs Fund at the end of 2020, (and covering treatments for a much wider range of conditions including rare disorders), may represent an opportunity for cutting-edge medicines to benefit patients more quickly, bypassing NICE's normal approval processes where uncertainty about the cost-effectiveness of a medicine is too great. However, it is yet another mechanism for drugs approval, when many say the system needs simplifying and unifying instead.

Ultimately, there needs to be a greater acceptance that medicines for treating rare conditions are generally going to be more expensive than those for treating common conditions, because of the smaller numbers of patients involved, but that should not, de facto, be a reason not to fund them. Denying people with rare conditions access to treatment goes against all arguments of equal access to care. Indeed, maybe the time has come to spread the cost of life-changing treatments for rare neurological conditions more equitably across the NHS and society, taking a more patient-centre approach? After all – and rightly so – we do not see this emphasis on cost-efficiency in other parts of healthcare, such as specialist surgery.

16 The vital role of new research and access to clinical trials for developing new treatments

In our recent survey of charities representing people with rare neurological conditions carried out in September 2020, while four in 10 said research since 2013 had made a big or moderate difference to the people they represent, four in 10 responded that there had been little or no research into the conditions they represent.

Clinical trials remain vital to advance genomic knowledge, including for rare neurological conditions. A powerful recent example is the treatment of spinal muscular atrophy (SMA) – a fatal childhood condition which can mean children with type 1 SMA may not live beyond the age of three. The severity relates to the presence of a duplicate backup SMN2 gene, but a new genetic medicine regulating the SMN2 gene, nusinersen (Spinraza), is making a life-changing difference for families. Clinical trials revealed that nusinersen (Spinraza) led to a significant improvement in children's motor function, allowing them to achieve, or maintain, physical milestones that they would never have reached without treatment, and to live much longer considering the typical course of the condition[cxxxii] as most children with SMA type 1 do not live beyond the age of two. Treatment effects in SMA type 2 and 3 have also been astounding: when treated early, children who could barely maintain a sitting position learned to stand and walk unsupported. However, such discoveries can only be validated through a rigorous clinical trials process.

The NIHR BioResource for Rare Diseases was established to identify the genetic causes of rare conditions, to improve rates of diagnosis, and to promote research that develops and validates treatments which improve care for patients and their families. It also provides opportunities for rare condition patients and families to become involved in research if they wish to do so.[cxxxiii]

The BioResource's Rare Disease Research Projects are listed in alphabetical order on their website in three groups:

- **Open for recruitment;**
- **Coming soon; and**
- **Closed to recruitment.**

Clinicians should be aware of these ongoing research projects and opportunities – which currently include trials on multiple system atrophy and ataxia.[cxxxiv]

It is vital that specialist neurology/genetic clinics are linked in with research institutions so that people suitable for inclusion in trials can be identified and recruited, as happened very successfully with The 100,000 Genomes project. Having a research coordinator/genetic nurse, together with holding good local databases of potentially qualifying patients, would go a long way towards ensuring that people with rare neurological conditions have access to clinical trials and the best possible treatment. The

"Clinical trials remain vital to advance genomic knowledge, including for rare neurological conditions."

16 The vital role of new research and access to clinical trials for developing new treatments

NHS Implementation Plan also referred to the development of Rare Disease Collaborative Networks, groups of providers who have a demonstrable research-active interest in a rare/very rare disease, with the aim of improving patient outcomes. Hopefully, more such Networks will be set up for research into rare neurological conditions, which would advance the outlook for people with these conditions.

Finally, as a Member of the European Union until 2020, the UK has been involved with the European Reference Network (ERN) on Rare Neurological Diseases[cxxxv] and the ERN on Rare and Complex Epilepsies (EpiCARE).[cxxxvi] ERNs are virtual networks across Europe involving specialist healthcare providers, whose aim is to tackle complex or rare medical conditions through cooperation and the sharing of knowledge and resources. Post-Brexit, UK neurology research centres will no longer be able to lead individual networks, but, it is to be hoped that they can retain their working links with these vital European research forums; this could be dependent on the post-Brexit arrangements agreed between the UK and the EU.

In our recent survey of charities representing people with rare neurological conditions carried out in September 2020, 5 in 10 organisations responded saying their research funding had been significantly or very significantly affected by Covid-19.

This is very worrying given the highly specialised, condition-specific research carried out by these of these organisations. Such charities must be supported by Government and/or other research funders so this vital medical research can continue.

17 Providing the right social care and financial support should not be an afterthought

While some people with rare neurological conditions may have a treatment available for their condition and they should have access to the mental health support and rehabilitation they need – social care, financial support and home adaptations are, in so many ways, just as important, although all too often overlooked.

In our recent survey of charities representing people with rare neurological conditions carried out in September 2020, one member organisation told us: 'In terms of social care, we are seeing packages and access reduced and the better support that was achieved for so many via the Independent Living Fund is disappearing fast.'

Social care provision has also been compromised by COVID-19.[cxxxvii] Providing adequate social care for all people with neurological conditions is a challenge that must be tackled.

Access to home adaptations and electric wheelchairs are also vitally important for those with progressive rare neurological conditions, as well as financial support for both individuals and carers. In the case of motor neurone disease (MND), there have been instances where home adaptations have come too late for people, who have passed away before the necessary home improvements could be installed.[cxxxviii] This is completely unacceptable. The time taken to have home adaptations completed and the cost are a worry to many people with MND.[cxxxix] This may, potentially, be the case for people with other progressive neurological conditions too.

Those assessing people for benefits should also have a greater understanding of neurological conditions the Alliance has been repeatedly told. Holistic care means looking after the whole person, and that includes their social care, their welfare, their housing, and supporting families and carers.

'In the case of motor neurone disease (MND), there have been instances where home adaptations have come too late for people, who have passed away before the necessary home improvements could be installed. This is completely unacceptable.'

At the end of October2020, the Alliance, alongside more than 40 other charities, co-signed a letter from Lord Sharkey to the Secretary of State for Health and Social Care, Rt. Hon. Matt Hancock, asking for a new package of social care measures for people of all ages with rare and less common conditions.[cxl] We also support the Care and Support Alliance in its current campaign for long-term reform of the social care system.

High-quality social care is vitally important for supporting people with rare and other neurological conditions and a new plan for social care is urgently needed that allows people with rare neurological conditions to live safely and in dignity in their own homes and in residential care if they should need it. This would also help the families who provide so much vital support. There must be no more delay by Government in sorting out social care reform.

18 Conclusions: looking to the future

Although there was a feeling in the rare disease community that the Rare Disease Strategy had not lived up to expectations in terms of delivering on its 51 commitments, especially given the long delay between publication of the Strategy in 2013 and the Implementation Frameworks in 2018, nonetheless the overall aims of the Strategy were both sound and significant and sight should not be lost of the progress that has been made in genetic testing, genome sequencing and the development of personalised medicines that could potentially make a real difference to people with rare neurological conditions.

Now is the time to build on the previous Strategy with a new Framework that captures the goals of the 2013–2020 Strategy to deliver real and lasting change for people with rare conditions.

At the beginning of this report we make recommendations on the 20 key policy measures that should to be adopted to improve care for people with rare neurological conditions, which we would hope to see reflected in the new Rare Diseases Framework. This is what members told us in September 2020 were their key priorities for the new Rare Disease Framework.

Neurological Alliance rare condition members' top priorities for what they want to see in the new Rare Disease Framework

- Improving knowledge of rare neurological conditions in primary care.
- Speedier referral to specialists, including from secondary care to regional centres.
- Having better access to appropriate tests and investigations and speedier diagnoses.
- Ensuring accurate and relevant health information is provided to people with rare neurological conditions and their families.
- A duty on clinicians to provide advice on where to find further support, e.g. from voluntary sector organisations.
- Better coordination of care along the whole care pathways, from primary care through to specialist care, including access to clinical trials.
- Emotional and psychological support to be integrated into care along the whole care pathway, with clear guidance on who is providing these services, so people do not 'fall between the gaps'.
- Improved access to rehabilitation for all those that need it to ensure their motor skills and independence are maximised.
- Faster and easier access to new, funded treatments.
- Better social care support to reverse recent declines in provision.
- Ensuring that assessments for benefits are conducted by people who are briefed on neurological conditions, including the 'hidden disabilities' they may have.
- Reform of the Newborn Screening Review Process to reflect advances in genetic testing and treatment, allowing for more infant screening for rare neurological conditions.

18 Conclusions: looking to the future

The new Rare Disease Framework must continue to emphasise speeding up diagnosis; delivering personalised care that meets people's needs, including improved care coordination; offering mental health support to all who need it; providing strong social care support in the community; and treatment close to home where possible. Neurologists – and bodies approving new treatments such as NICE – must also capitalise on the new opportunities offered by the NHS Genomic Medicine Service. However, improved diagnosis must be matched by approved access to new treatments, in order that people with rare neurological conditions do not have to wait years for therapies already available in other countries.

Above all, rare neurological conditions need to be brought out of the shadows and treated like any other health condition – complex yes, but not too complex to manage and treat well. People with rare neurological conditions are entitled to equal access to treatment and therapies and parity of priority in the health and care system in England.

Nothing less is acceptable.

'Above all, rare neurological conditions need to be brought out of the shadows and treated like any other health condition – complex yes, but not too complex to manage and treat well. People with rare neurological conditions are entitled to equal access to treatment and therapies and parity of priority in the health and care system in England. Nothing less is acceptable.'

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Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Acoustic neuroma	An acoustic neuroma is a type of benign brain tumour growing on the nerve used in hearing and balance, which can cause problems such as hearing loss and unsteadiness. It is also known as a vestibular schwannoma.	Progressive	Childhood and adulthood	27,800	No	-	British Acoustic Neuroma Association www.bana-uk.com
Ataxias (a broad class of disorders, the most common inherited type being Friedreich's ataxia)	Ataxia is a term for a group of disorders that affect coordination, balance and speech. For example, Friedreich's ataxia is an inherited condition that causes progressive damage to the nervous system, while ataxia telangiectasia causes degeneration in the part of the brain that controls motor movements and speech (the first signs are unsteady walking and slurred speech, usually occurring during the first five years of life).	Progressive	Childhood and adulthood	15,000*[4]	Some forms are, others not	Yes, for some genetic types of ataxia	Ataxia UK www.ataxia.org.uk

1 All definitions are take from either the US National Institute of Neurological Disorders and Stroke website, The NHS Health A–Z website, or the Patient Groups listed above.
2 Neurological Alliance, Neuro Numbers 2019. Available here: www.neural.org.uk/wp-content/uploads/2019/07/neuro-numbers-2019.pdf. Accessed October 2020.
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4 All figures marked with a * are taken from the Neurological Alliance's Survey of Rare Neurological Charities, carried out in September 2020.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Batten disease (there are 13 different genetic variants)	Batten disease is the name for a broad class of rare, fatal, inherited disorders of the nervous system also known as neuronal ceroid lipofuscinoses, or NCLs.	Progressive	Childhood, very rarely in adults	103	Yes	Yes, genetic tests for some variants are available	Batten Disease Family Association www.bdfa-uk.org.uk
Brown Séquard syndrome	Brown Séquard syndrome (BSS) is characterised by a lesion in the spinal cord which results in weakness or paralysis on one side of the body and a loss of sensation on the opposite side.	Sudden onset – sometimes related to injury	Childhood or adulthood	Not available	No	–	No dedicated support group. Further information available from the Brain & Spine Foundation: www.brainandspine.org.uk
Cerebral palsy	Cerebral palsy is the name for a group of lifelong conditions that affect movement and coordination caused by a neurological problem that develops before, during or soon after birth.	Stable with changing needs	Neonatal, infancy	25,273	No	–	Action Cerebral Palsy www.actioncp.org

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Charcot-Marie-Tooth disease	Charcot-Marie-Tooth disease (CMT) is a group of inherited conditions that causes damage to the peripheral nerves. It is also known as hereditary motor and sensory neuropathy (HMSN) or peroneal muscular atrophy (PMA).	Progressive	Childhood or early adulthood	19,376	Yes	No	Charcot-Marie-Tooth UK www.cmt.org.uk
Childhood multiple sclerosis (MS)	Multiple sclerosis (MS) is a condition that affects the brain and spinal cord (the central nervous system), causing a wide range of potential symptoms, including problems with vision, arm or leg movement, sensation or balance. Childhood MS is very rare.	Progressive	Under the age of 18	250[5]	No	–	MS Trust www.mstrust.org.uk/a-z/childhood-ms MS Society www.mssociety.org.uk

5 Source: MS Trust website. Available here: www.mstrust.org.uk/a-z/childhood-ms. Accessed October 2020.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Chronic inflammatory demyelinating polyneuropathy (CIDP)	Chronic inflammatory demyelinating polyneuropathy (CIDP) is a neurological disorder characterised by progressive weakness and impaired sensory function in the legs and arms.	Progressive	Childhood and adulthood	3,329	No	–	Guillain-Barré and Associated Inflammatory Neuropathies www.gaincharity.org.uk
Corticobasal degeneration (CBD)	Corticobasal degeneration is a progressive neurological disorder characterised by nerve cell loss and atrophy of multiple areas of the brain including the cerebral cortex and the basal ganglia.	Progressive	Adulthood – age 40 and older	2,000	No	–	PSPA www.pspassociation.org.uk
Dravet syndrome (a rare form of epilepsy)	Dravet syndrome is a catastrophic form of treatment-resistant epilepsy that also encompasses intellectual disabilities and a spectrum of associated conditions which may include autism, behavioural problems and difficulties with speech, mobility, feeding and sleep (with 85% survival into adulthood).	Progressive	Infancy – the average age of onset is five months	Approx. 4,000 (1 in 15,000 of the population)[6]	Yes (85%+ have a SCN1A mutation)	Yes	Dravet Syndrome UK www.dravet.org.uk

6 Source: Dravet Syndrome UK website. Available here: www.dravet.org.uk. Accessed October 2020.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Duchenne muscular dystrophy	Duchenne muscular dystrophy is a neuromuscular wasting condition caused by the lack of a protein called dystrophin. It usually affects only boys; there are also eight other forms of muscular dystrophy.	Progressive	Early childhood	2,500	Yes	Yes	Muscular Dystrophy UK www.musculardystrophyuk.org
Early onset Parkinson's	Parkinson's disease (PD) belongs to a group of conditions called motor system disorders, which cause unintended or uncontrollable movements of the body. Early onset Parkinson's starts under the age of 50.	Progressive	Defined as Parkinson's which starts under the age of 50	17,000[7]	No	–	Spotlight YOPD spotlightyopd.org
Encephalitis	Encephalitis is a condition associated with inflammation of the brain. It is generally caused either by an infection or through an autoimmune reaction.	Sudden onset	Any age	Affects up to 6,000 people in the UK each year.[8]	No	–	Encephalitis Society www.encephalitis.info/Default.aspx
Guillain-Barré syndrome	Guillain-Barré syndrome (GBS) is a rare neurological disorder in which the body's immune system attacks part of the peripheral nervous system.	Sudden onset	Childhood and adulthood	Incidence is up to 1,000 cases each year (there is no prevalence figure available)	No	–	Guillain-Barré and Associated Inflammatory Neuropathies www.gaincharity.org.uk

7 Source: Spotlight YOPD website. Available here: spotlightyopd.org. Accessed October 2020.
8 Encephalitis Society website. Available here: www.encephalitis.info/Default.aspx. Accessed October 2020.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Familial dysautonomia or Riley-Day syndrome	Dysautonomia refers to a disorder of autonomic nervous system (ANS) function that generally involves failure of the sympathetic or para-sympathetic components of the ANS.	Progressive	Childhood	Not available	Yes	No	FD-UK.ORG familialdysautonomia.co.uk
Hereditary Spastic paraplegia (HSP)	Hereditary spastic paraplegia (HSP), also called familial spastic paraparesis (FSP), refers to a group of inherited disorders that are characterised by progressive weakness and spasticity (stiffness) of the legs.	Progressive	Childhood and adulthood	Estimates range from 1 in 11,000 people to 1 in 77,000 people[9]	Yes	Yes	The Hereditary Spastic Paraplegia Support Group hspgroup.org
Huntington's disease	Huntington's disease (HD) is an inherited disorder that causes brain cells, called neurons, to die in various areas of the brain, including those that help to control voluntary (intentional) movement.	Progressive	Adulthood	6,739	Yes	Yes	Huntington's Disease Association www.hda.org.uk
Idiopathic intracranial hypertension	Idiopathic intracranial hypertension (IIH), also known as benign intracranial hypertension, is a condition associated with raised fluid pressure around the brain.	Sudden onset	Late childhood and adulthood	6,060	No	–	IIH-UK www.iih.org.uk

9 NHS website. Hereditary spastic paraplegia. Available here: www.nhs.uk/conditions/hereditary-spastic-paraplegia/#:~:text=It%27s%20difficult%20to%20know%20exactly,to%201%20in%2077%2C000%20people. Accessed October 2018.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Kennedy's disease	Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly in the arms and legs.	Progressive	Usually symptoms appear between the ages of 30 and 60	Around one in 40,000 people are affected	Yes	Yes	Motor Neurone Disease Association www.mndassociation.org
McArdle's disease	McArdle's disease is a genetic, neuromuscular disorder associated with muscle cramps and injury, and myoglobinuria induced by sudden, vigorous exercise.	Generally progressive	Generally, symptoms appear by the age of 15, but onset can be at any age	350	Yes	Some genetic glycogen storage tests available	Association of Glycose Storage Disease UK agsd.org.uk
Motor neurone disease	Motor neurone disease is a progressive neurological disorder that destroys cells that control essential muscle activity such as speaking, walking, breathing and swallowing.	Progressive	Adulthood	5,000 in UK	Generally not, but genetic in around 10% of cases	Yes, available for inherited cases	Motor Neurone Disease Association www.mndassociation.org

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Multiple system atrophy	Multiple system atrophy (MSA) is a progressive neurodegenerative disorder characterised by symptoms of autonomic nervous system failure such as fainting spells and bladder control problems, combined with motor control symptoms such as tremor, rigidity, and loss of muscle coordination.	Progressive	Adulthood	3,000 in UK*	No	–	Multiple System Atrophy Trust www.msatrust.org.uk
Myasthenias – a group of neuromuscular conditions, the most common of which is myasthenia gravis	All forms of myasthenia are due to problems in the communication between nerve cells and muscles. Myasthenia gravis is a chronic autoimmune neuromuscular disease characterised by varying degrees of weakness of the skeletal muscles of the body responsible for breathing and moving parts of the body, including the arms and legs.	Intermittent	Childhood and adulthood	10,109	Myasthenia gravis, ocular myasthenia and Lambert-Eaton myasthenic syndrome are autoimmune conditions, while congenital myasthenic syndrome (CMS) is genetic.	Only for Congenital Myasthenic Syndrome (CMS)	Myaware www.myaware.org

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Narcolepsy	Narcolepsy is a chronic neurological disorder caused by the brain's inability to regulate sleep-wake cycles.	Stable with changing needs	Childhood and adulthood	22,250	No	–	Narcolepsy UK www.narcolepsy.org.uk
Neurofibromatosis – including types 1 and 2 and schwannomatosis	Neurofibromatoses are genetic disorders that cause tumours to grow in the nervous system.	Progressive	Childhood and adulthood	24,093	Mostly genetic with some non-genetic cases	Yes	Nerve Tumours UK nervetumours.org.uk
Neuromyelitis optica	Neuromyelitis optica (NMO) is an autoimmune disease of the central nervous system that predominantly affects the optic nerves and spinal cord.	Progressive	Childhood and adulthood	842	No	–	Tranverse Myelitis Society www.myelitis.org.uk
Neurosarcoidosis	Neurosarcoidosis is a manifestation of sarcoidosis, a chronic inflammatory disorder, specifically in the nervous system.	Sudden onset	Childhood and adulthood	1,000	No	–	Sarcoidosis UK www.sarcoidosisuk.org

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Progressive supranuclear palsy	Progressive supranuclear palsy (PSP) is a brain disorder that causes serious and progressive problems with control of gait and balance, along with complex eye movement and cognitive problems.	Progressive	Adulthood	Up to 10,000 cases in the UK*	No	–	PSPA www.pspassociation.org.uk
Rett syndrome	Rett syndrome is a rare childhood neurological and developmental disorder that almost exclusively affects females. Infants appear to develop normally for the first several months before development stalls (typically between the ages of 6–18 months)	Progressive	Childhood	1,264	Yes	No	Rett UK www.rettuk.org
Ring chromosome 20 syndrome, also known as r(20) syndrome	Ring chromosome 20 epilepsy syndrome is a chromosomal abnormality characterised by medically intractable epilepsy, nocturnal subtle seizures, behavioural problems and mild mental impairment.	Intermittent	Childhood	Not available	Yes	No	Ring20 Research and Support UK ring20researchsupport.co.uk

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Spina bifida	Spina bifida is a neural tube defect (a disorder involving incomplete development of the brain, spinal cord, and/or their protective coverings) caused by the failure of the fetus's spine to close properly during the first month of pregnancy.	Stable with changing needs	At birth	Approx. 100 births a year[10]	Influenced by genetic and environmental factors	Preterm maternal testing	Shine Charity www.shinecharity.org.uk
Spinal muscular atrophy – the most common form is 5q SMA, which includes SMA types 1, 2, 3 and 4. There are some other very rare forms, information about which can be found at smauk.org.uk/rarer-forms-of-sma	Spinal muscular atrophy refers to a group of hereditary diseases that damage and kill specialised nerve cells in the brain and spinal cord (motor neurons). Motor neurons control movement in the arms, legs, face, chest, throat and tongue, as well as skeletal muscle activity including speaking, walking, swallowing and breathing.	Progressive	Types 1–3 in childhood, but type 4 can be in over 18s	660–1,320 in UK*	Yes	Yes	Spinal Muscular Atrophy UK smauk.org.uk
Syringomyelia	Syringomyelia is a disorder in which a fluid-filled cyst forms within the spinal cord. This cyst, called a syrinx, expands and elongates over time, damaging the spinal cord.	Progressive	All ages, but most commonly presents in 20s and 30s	Not available, but prevalence has been quoted as 8.4 cases per 100,000[11]	Only rarely	–	Ann Conroy Trust www.annconroytrust.org

10 NHS England, Service Specification: Open fetal surgery to treat fetuses with open spina bifida. Available here: www.england.nhs.uk/commissioning/wp-content/uploads/sites/12/2018/12/Open-fetal-surgery-to-treat-fetuses-with-open-spina-bifida.pdf. Accessed October 2020.

11 Dr Mary Lowth, Patient.info website. Syringomyelia and syringobulbia. Available here: patient.info/doctor/syringomyelia-and-syringobulbia. Accessed October 2020.

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Transverse myelitis	Transverse myelitis is an inflammation of the spinal cord. The inflammation interrupts communications between nerve fibres in the spinal cord and the rest of the body, affecting sensation and nerve signalling	Sudden onset	More commonly starts in adulthood although children can also be affected	3,820	No	–	Tranverse Myelitis Society www.myelitis.org.uk
Tuberous sclerosis complex	Tuberous sclerosis (TSC) is a rare genetic disease that causes benign tumours to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs and skin. It usually affects the central nervous system.	Progressive	Usually starts between the ages of 15 and 30	6,720	Yes	Yes	Tuberous Sclerosis Association www.tuberous-sclerosis.org

Appendix A: Prevalence of rare neurological conditions in England and the availability of a genetic test

Rare neurological condition	Brief description[1]	Type	Age of onset	Prevalence in England, unless otherwise stated (figures sourced from Neuro Numbers 2019,[2] unless otherwise indicated)	Genetic condition (yes/no)	Availability of genetic tests[3]	Patient group
Wilson's disease	Wilson's disease is a genetic disorder in which copper builds up in the body, mainly in the liver and brain; neurological symptoms relate to speech, swallowing or physical coordination and uncontrolled movements or muscle stiffness.	Progressive without treatment	Symptoms most commonly appear in adolescence, although they can appear earlier or later in life	c.1,850 (1 in 30,000 of the population)	Yes	Yes	Wilson's Disease Support Group – UK www.wilsonsdisease.org.uk/WDSG-P0.asp

Disclaimer
Please note: This list of conditions is not exhaustive; further information about each condition, including symptoms, is available on each of the patient group websites listed above. Also, while every reasonable effort has been made to ensure that the information in this Appendix is complete, correct and up-to-date, this cannot be guaranteed and the Neurological Alliance shall not be liable for any damages incurred as a result of its use.

Appendix B: Highly specialised services for rare neurological conditions, including paediatric neuroscience services[12]

Title	Service known as	NHS providers	NHS website – link to specification	UK-wide commissioning arrangements
Ataxia telangiectasia services for adults	AT adults	Royal Papworth Hospital NHS Foundation Trust	www.england.nhs.uk/wp-content/uploads/2018/08/Ataxia-telangiectasia-service-adult.pdf	Fully commissioned on behalf of England & Scotland
Ataxia telangiectasia services for children	AT Children	Nottingham University Hospitals NHS Trust	www.england.nhs.uk/wp-content/uploads/2018/08/Ataxia-telangiectasia-service-children.pdf	Fully commissioned on behalf of England & Scotland
Complex neurofibromatosis type 1 service (adults and children)	NF1	Manchester University NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust	www.england.nhs.uk/wp-content/uploads/2013/06/b13-comp-neurofib-1.pdf	Fully commissioned on behalf of England only
Diagnostic service for rare neuromuscular disorders (adults and children)	Rare Neuromuscular disorders	Great Ormond Street Hospital for Children NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle Upon Tyne Hospitals NHS Foundation Trust University College London Hospital NHS Foundation Trust	www.england.nhs.uk/wp-content/uploads/2013/06/d04-diagn-serv-rare-neuromusc.pdf	Fully commissioned on behalf of England & Scotland
McArdle's disease service (children)		University College London Hospital NHS Foundation Trust	www.england.nhs.uk/wp-content/uploads/2013/06/e06-mcardle-dis-adult.pdf	Fully commissioned on behalf of England & Scotland
Multiple sclerosis management service for children	Paediatric MS	Alder Hey Children's NHS Foundation Trust Cambridge University Hospitals NHS Foundation Trust Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust The Newcastle Upon Tyne Hospitals NHS Foundation Trust	www.england.nhs.uk/publication/multiple-sclerosis-management-service-for-children	Fully commissioned on behalf of England only

12 NHS England, NHS providers of highly specialised services and the UK-wide commissioning arrangements, updated July 2020. Available here: www.england.nhs.uk/publication/nhs-providers-of-highly-specialised-services. Accessed October 2020.

Appendix B: Highly specialised services for rare neurological conditions, including paediatric neuroscience services

Title	Service known as	NHS providers	NHS website – link to specification	UK-wide commissioning arrangements
Neurofibromatosis type 2 service (all ages)	NF2	Cambridge University Hospitals NHS Foundation Trust Manchester University NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Oxford University Hospitals NHS Trust	www.england.nhs.uk/wp-content/uploads/2013/06/b13-neurofib-2.pdf	Fully commissioned on behalf of England & Scotland
Neuromyelitis optica service (adults and children)	NMO	Oxford University Hospitals NHS Trust The Walton Centre NHS Foundation Trust	www.england.nhs.uk/wp-content/uploads/2013/06/d04-neuromyelitis-optica-serv.pdf	Fully commissioned on behalf of England & Scotland
Open fetal surgery to treat fetuses with spina bifida	Spina bifida	University College London Hospital NHS Foundation Trust – Fetal Surgery Centres (FSCs) in a shared care pathway together with local maternity units/regional Fetal Medicine Units (RFMUs) and regional Neurosurgery Centres.	www.england.nhs.uk/commissioning/publication/open-fetal-surgery-to-treat-fetuses-with-open-spina-bifida	Fully commissioned on behalf of UK (Pre-1991)

Explantory notes on the above neurological conditions[13,14]

- **Ataxia telangiectasia (AT)** is a rare, neurodegenerative and progressive condition that starts in early childhood causing severe disability and premature death. It affects many parts of the body and a wheelchair is often needed by the age of 10. The average life expectancy is 25 years. During the adult stage of the condition, there is increased susceptibility to leukaemias, lymphoma, pneumonia, chronic lung disease and neurological decline. Fewer than 100 adults in England have AT.
- **McArdle's disease** is a rare, genetic, neuromuscular disorder associated with muscle cramps and injury and myoglobinuria induced by sudden, vigorous exercise.
- **Neurofibromatosis type 1 (NF1)** is an inherited genetic disorder, characterised by formation of neurofibromas (tumours involving nerve tissue) in the skin, subcutaneous tissue, cranial nerves and spinal root nerve tissue. About one in 25,000 of the population has NF1. With complex NF1 there is a high risk of developing rare complications, which may affect most of the body's systems.
- **Neurofibromatosis type 2 (NF2)** is a genetic disorder characterised by the growth of non-cancerous tumours in the central nervous system.
- **Neuromyelitis optica (NMO)** (also known as Devic's disease) is a rare inflammatory demyelinating disorder of the central nervous system that typically presents as severe optic neuritis and longitudinally extensive myelitis, often followed by further severe attacks, which usually result in permanent disability (visual loss, limb weakness, respiratory muscle weakness). There are high mortality and morbidity rates associated with the condition.
- **Paediatric onset multiple sclerosis (POMS)** is a rare condition in which children under 18 develop multiple sclerosis (MS). MS is a condition of the central nervous system (CNS) characterised by chronic brain inflammation and occurs when the coating around nerve fibres (myelin) becomes damaged (known as demyelination).

13 NHS England, Highly Specialised Services 2018. Available here: www.england.nhs.uk/commissioning/wp-content/uploads/sites/12/2018/12/Highly-Specialised-Services-2018-v2.pdf. Accessed October 2020.
14 NHS England, Specialised Services, Women and Children, E04. Paediatric Neurosciences. Available here: www.england.nhs.uk/commissioning/spec-services/npc-crg/group-e/e04. Accessed October 2020.

Appendix C: Neuroscience Centres in England[15]

London

Charing Cross Hospital
Fulham Palace Road
London W6 8RF

Cromwell Hospital (private)
Cromwell Road
London SW5 0TU

Kings College Hospital
Denmark Hill
London SE5 9RS

The National Hospital for
Neurology and Neurosurgery
Queen Square
London WC1N 3BG

The Royal Free
Pond Street
London NW3 2QG

Barts and the London Centre for Neurosciences
Royal London Hospital
Whitechapel Road
London E1 1BB

St George's Hospital
Blackshaw Road
London SW17 0QT

The Wellington Hospital (private)
Wellington Place
London NW8 9LE

East England

Addenbrooke's Hospital
Hills Road
Cambridge CB2 0QQ

Queen's Hospital
Rom Valley Way
Romford
Essex RM7 0AG

East Midlands

Queen's Medical Centre
Derby Road
Nottingham NG7 2UH

North East England

James Cook University Hospital
Marton Road
Middlesbrough TS4 3BW

Regional Neurosciences Centre
Royal Victoria Infirmary
Queen Victoria Road
Newcastle upon Tyne NE1 4LP

North West England

Greater Manchester Neurosciences Centre
Salford Royal NHS Foundation Trust
Stott Lane
Salford M6 8HD

Royal Preston Hospital
Sharoe Green Lane
Fulwood
Preston PR2 9HT

Chorley and South Ribble Hospital
Preston Road
Chorley PR7 1PP

The Walton Centre for Neurology
and Neurosurgery
Lower Lane
Fazakerley
Liverpool L9 7LJ

15 Brain & Spine Foundation, adapted from List of Neurocentres in the UK. Available here: www.brainandspine.org.uk/wp-content/uploads/2018/03/BSF_List-of-Neurocentres-in-the-UK.pdf. Accessed October 2020.

Appendix C: Neuroscience Centres in England

South East England

Hurstwood Park Neurosciences Centre
Lewes Road
Haywards Heath RH16 4EX

Wessex Neurological Centre
Southampton General Hospital
Tremona Road
Southampton SO16 6YD

John Radcliffe Hospital
Headley Way
Headington
Oxford OX3 9DU

South West England

Derriford Hospital
Derriford Road
Plymouth PL6 8DH

Southmead Hospital Bristol
(formerly Frenchay Hospital)
Brunel Building
Southmead Road
Westbury-on-Trym
Bristol BS10 5NB

West Midlands

University Hospital of North Staffordshire
City General
Newcastle Road
Stoke on Trent ST4 6QG

Queen Elizabeth Hospital
Edgbaston
Birmingham B15 2TH

University Hospitals Coventry
and Warwickshire
Clifford Bridge Road
Walsgrave
Coventry CV2 2DX

Yorkshire and The Humber

Hull Royal Infirmary
Anlaby Road
Hull HU3 2JZ

Leeds Centre for Neurosciences
Leeds General Infirmary
Great George Street
Leeds LS1 3EX

Royal Hallamshire Hospital
Glossop Road
Sheffield S10 2JF

Appendix D: Neuroscience Clinical Reference Group (CRG) treatment recommendations (including for rare conditions)[16]

- **Treatment algorithms** – which provide a framework to aid decision-making on access to a particular treatment, to help reduce variations in access and ensure safe and effective prescribing. There is only one such algorithm advocated by the CRG, applying to disease modifying therapies (DMTS) for multiple sclerosis:
  - Treatment algorithm for multiple sclerosis disease modifying therapies
- **Service specifications** – developed by specialised clinicians, commissioners, expert patients and public health representatives, which define the standards of care expected from NHS providers funded by NHS England to deliver specialised care. The following service specifications fall within the scope of the Neuroscience CRG; some are for rare conditions:
  - A diagnostic service for rare neuromuscular disorders (all ages)
  - Neurointerventional services for acute ischaemic & haemorrhagic stroke
  - Neurosurgery (adult)
  - Neurosciences specialised neurology (adult)
  - Neuromyelitis optica service (adults and adolescents)
  - Neuropathology
- Service specifications falling within the scope of the Paediatric Neurosciences Clinical Reference Group (CRG):[17]
  - Paediatric neurosciences: neurosurgery
  - Paediatric neurosciences: neurology
  - Paediatric neurosciences: neurodisability
  - Paediatric neurosciences: neurorehabilitation
  - Refractory epilepsy specialist clinical advisory service
- **Policies** – a commissioning policy defines access to a particular treatment for a particular group of people. A NICE Technology Appraisal Guideline on the same topic will replace, or be incorporated into, a commissioning policy. These policies are developed to ensure consistency in access to treatments nationwide. The following policies fall within the scope of this CRG:

Commissioned:

- Deep brain stimulation (DBS) in movement disorders
- Disease modifying therapies for patients with multiple sclerosis (MS)
- Intrathecal baclofen (ITB) (for spasticity/dystonia)
- Levodopa-carbidopa intestinal gel (LCIG) (for Parkinson's)
- Mechanical thrombectomy for acute ischaemic stroke
- Vagal nerve stimulation for epilepsy

16 NHS England, Specialised Services. D04 Neurosciences. Available here: www.england.nhs.uk/commissioning/spec-services/npc-crg/group-d/d04. Accessed October 2020.

17 NHS England, Specialised Services. Women and Children. E04. Paediatric Neurosciences. Available here: www.england.nhs.uk/commissioning/spec-services/npc-crg/group-e/e04. Accessed October 2020.

Appendix D: Neuroscience Clinical Reference Group (CRG) treatment recommendations (including for rare conditions)[16]

Not routinely commissioned:

- Amifampridine phosphate for the treatment of Lambert-Easton myasthenic syndrome
- Deep brain stimulation for refractory epilepsy (all ages)
- Deep brain stimulation for refractory Tourette syndrome (adults)
- Fampridine for multiple sclerosis
- Rituximab for chronic inflammatory demyelinating polyradiculoneuropathy (CIDP), multifocal motor neuropathy (MMN), vasculitis of the peripheral nervous system & IgM paraprotein-associated demyelinating neuropathy (adults)

- **Policy statements** – in circumstances when NHS England needs to implement a commissioning policy quickly, a Policy Statement or Urgent Policy Statement is issued. These apply to neurology:
  - Alemtuzumab for treating relapsing-remitting multiple sclerosis – third cycle (all ages)
  - Amifampridine (firdapse) for Lambert Easton myasthenic syndrome (LEMS)
  - Cerebellar stimulator implants
  - Deep brain stimulation (DBS) for indications except movement disorders
  - Flow diverting devices for intracranial aneurysms
  - Infliximab for refractory or progressive neurosarcoidosis (adults and post-pubescent children)
  - Natalizumab-induced progressive multifocal leukoencephalopathy in relation to immune reconstitution inflammatory syndrome in multiple sclerosis
  - Rituximab biosimilar for the treatment of myasthenia gravis (adults)

THE
NEUROLOGICAL
ALLIANCE

Contact us

The Neurological Alliance
The Junction
Station Road
Watford
Herfordshire WD17 1EU

Email: info@neural.org.uk
Phone: 01923 882 590

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