Consultation on UK Plan for Rare Disease
Neurological Alliance response

About the Neurological Alliance
The Neurological Alliance is the only collective voice for more than 70 national and regional brain and spine organisations working together to make life better for 8 million children, young people and adults in England with a neurological condition.

Our vision is for a better quality of life for each individual diagnosed with a neurological condition. Our mission is to raise awareness and understanding of neurological conditions to ensure that every person diagnosed with a neurological condition has access to high quality, joined up services and information from their first symptoms, throughout their life.

Overview
The Neurological Alliance welcomes the opportunity to comment on this consultation on a UK Plan for Rare Diseases.

With the majority of the 324[1] known neurological conditions classified as rare, a UK Plan for Rare Diseases has the potential to enhance the quality of, and access to, appropriate care and support for the thousands of people living with a rare neurological condition in the UK.

Many elements of the proposed plan are strong and have the genuine potential to improve care and outcomes for people with rare diseases. We fully endorse the plan's proposals aimed at enhancing integration and coordination across primary, secondary and tertiary care, harnessing technology to assist GPs in diagnosis and referral, and the establishment of expert centres.

Other areas of the plan need to be fortified if it is to transform care and support for people with rare diseases and move beyond the status quo of service provision and research. We are particularly disappointed that the vital role performed by patient representative organisations and support groups is not fully recognised in the consultation and that, as such, they are not a core component of the plan's vision.

Our member organisations have a wealth of experience and expertise to offer patients, the public and health and social care services. Without regarding patient organisations as equal partners alongside healthcare professionals and relevant health and social care structures, their full potential will remain untapped.

The plan also needs to acknowledge the pressing need to develop the UK research base in respect of rare disease.

We would stress that, in order for the plan's vision to be realised, it must be adequately resourced, including the provision of dedicated leadership at a national level, and be underpinned by a robust implementation strategy. The failure of the of the National Service Framework for Long Term Conditions as compared to those

[1] *National Service Framework for Long Term Conditions* - Department of Health, 2005

National Service Frameworks allocated these elements highlights their centrality to success and we would urge the four health departments to take this into full consideration when devising the final plan. We would also emphasises the need for close co-ordination between the four departments if care and support for people with rare diseases is to be provided on an equitable basis across the UK.

1. Do you agree that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, particularly in neurology and genetics?

Achieving early and accurate diagnosis represents a key challenge across the neurological spectrum; the complexity, manifestation and rarity of the majority of neurological conditions frequently results in diagnostic delay, to the detriment of the health outcomes of those individuals affected.

Securing a diagnosis is absolutely essential in enabling individuals to access appropriate care, support and, where available, treatment. For those with a suspected neurological condition, timely referral to a neurologist is crucial to achieving this. The imperative of early and accurate diagnosis is particularly pronounced for certain neurological conditions; for those with encephalitis, for example, acquired brain injury can result from a delay in diagnosis. Non-diagnosis also leads to cost inefficiency and places additional stress upon the individual concerned and their family.

Given the importance of diagnosis to all people with rare conditions, we support the proposed exploration of the potential of expert clinical systems to reduce diagnostic delay, including clearly defined care pathways to expert centres. Computerised databases and algorithms are particularly important, delivering key diagnostic information to healthcare professionals in their working environment. As people with neurological conditions may be referred to other specialists and undergo inappropriate interventions before they are referred to neurological expertise, technology can provide GPs with an invaluable source of support to enable appropriate referral for neurological and other rare conditions.

We also strongly support the development of core diagnostic indicators such as red flags to aid GPs in the process of timely referral to a specialist having identified a particular symptom or set of symptoms. Such indicators would also have the potential to aid development of core training materials and raise the profile amongst healthcare professionals of symptoms to be alert to.

Diagnostic delay can also be mitigated by effective training for healthcare professionals and raising public awareness of rare diseases. In respect of the latter, the UK Plan for Rare Disease represents a real opportunity to raise the profile of rare diseases both publically and amongst health and social care professionals and we encourage the departments responsible to consider how best to capitalise on this.

2. Can you suggest ways of rare disease featuring more prominently in speciality training for doctors?

We appreciate that due to their very nature, not all rare conditions can be covered in the context of specialist training. Given the impossibility of equipping specialists with training in all rare conditions, we strongly recommend the explicit signposting of

patient support and patient representative groups during the training process; as the plan rightly specifies, these organisations 'powerfully articulate the needs and priorities of people with rare diseases'.

The development and ongoing maintenance of clinical guidelines and patient pathways also offer crucial support to healthcare professionals in the provision of the best quality care for patients with rare conditions.

In addition to specialist training for doctors, the inclusion of rare disease in the training for other professionals should also be considered; dentists, for example, require training to allow them to identify potential cases of trigeminal neuralgia.

In terms of Continuing Professional Development (CPD), e-learning should be exploited to enhance understanding of rare diseases. A specific rare disease module could be developed, for example.

3. Do you agree that the UK National Screening Committee should take into account the benefit of screening in reducing the 'diagnostic odyssey' and in allowing informed choice for subsequent family planning?

Not all neurological conditions can be screened for at birth. Where diagnosis of a rare disease in early infancy is possible and treatment can reduce the overall impact of the condition on the individual's quality of life, however, we would support the inclusion of the condition in UK screening programmes.

Where screening is not an option, comprehensive education and training can help address the 'diagnostic odyssey'.

4. How can the NHS best ensure research in rare diseases carried out by the NIHR biomedical research centres and units is rapidly transferred into practice for the benefit of UK patients and their families and carers?

Whilst we agree that the overall UK research base is strong, we would emphasise that this strength is not reflected in research into neurological conditions and other rare diseases. With research that focuses on more prevalent conditions typically being favoured over research into rare diseases, the level of funding for such research is currently not proportionate to the 3.5 million people comprising the UK's rare disease patient population.

In the absence of both accurate records on the epidemiology of rare diseases to enable appropriate budgeting and ring fenced funding for rare disease research, we are deeply concerned that rare diseases will remain a chronically underrepresented part of the UK research base.

Rapid adoption of research into practice is highly desirable and the multiple channels of telecommunication available in the modern health service offer a real opportunity to enable improved flows of information between all parts of the health system. Determining which means of accessing and receiving information is preferred by healthcare professionals would harness the greatest potential of technology to assist in the dissemination and uptake of research findings. There may also be potential for Royal Colleges to play a role in the dissemination of new research.

5. Do you agree that commissioners of care for people with rare diseases should assess options for improved care coordination, including named care coordinators?

As part of the Neurological Alliance's current campaign, *'It's time for a better deal for neurology'*, we have been actively campaigning for the option for all people with a neurological condition to have both a care plan and named care coordinator. The evidence base in support of care planning and named coordination is extensive and we would fully endorse the inclusion of the provision of both for all people with a rare condition in the final UK Plan for Rare Diseases.

As a large proportion of people with neurological conditions have a high level of interaction with health and social care services, integration across and within health and social care is a core priority. If integration is to be successfully realised, commissioners must appreciate its practical value and importance to patients' experience of care.

We are concerned that the existing tariff structure presents a barrier to effective care coordination; currently, if a patient visits a consultant and a specialist nurse on the same hospital visit, the visit to the specialist nurse is paid at a reduced rate compared to if the patient had made two separate trips. These perverse incentives need to be resolved.

6. Do you agree that this list of criteria for expert centres should be the basis for future shaping of services?

- Co-ordinated care
- Adequate caseload for expertise
- Not dependent on a single clinician
- Arrangements for the transition from children's to adults' services
- Engaged with people with rare conditions
- Research active

We agree with criteria described for expert centres, but do not regard the list as exhaustive. In particular, we note that the criteria do not specify the need for multidisciplinary team working. Expert centres must be multidisciplinary environments in which service users have access to the relevant expertise on a single site; this will improve early and accurate diagnosis, create an environment conducive to research and allow the accumulation of extensive knowledge about rare conditions and their effective treatments.

Additionally, we would welcome an emphasis in the criteria on the importance of coordinating the activity in expert centres with community services. Thorough and effective liaison with local health services, care and education providers is essential if a gulf between local and specialist services is to be avoided.
Expert centres must be able to provide necessary follow up care and support; whilst some patients may only need to be seen once or twice before being referred back to local services, others will require ongoing contact with the centre. It is also important that expert centres can operate flexibly where possible to prevent patients presenting to out of hours and the emergency services.

We also caution that in order for expert centres to fulfil their function in respect of research, a greater amount of research into rare diseases needs to be both initiated and sustained.

These criteria should be reviewed and assessed on an ongoing basis to ensure that specialist centres are meeting all the requirements and demands once they become operational.

As evidenced by the recent National Audit Office and Public Accounts Committee reports into services for people with neurological conditions, significant variations in neurology services exist across the country[2]. In order to avoid the perpetuation of service variation and mitigate the potential for a postcode lottery, expert centres will need to provide comprehensive coverage across each of the four nations of the United Kingdom.

To oversee the establishment of this network of expert centres and promote the plan's ambitions to enhance service provision, equity of access and outcomes for people with rare diseases, we support the appointment of a national leader for rare diseases. We regard this as complementary to Neurological Alliance's campaign call for the appointment of a national leader for neurology; with many non-neurological diseases sharing a need for neurological expertise, and with non-rare neurological conditions affecting the lives of millions of people, there is a clear need for, and distinction between, these two roles.

7. Do you agree that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance?

It is essential that each expert centre knows its network of local hospitals and that, in turn, each local hospital knows the pathway to the expert centre. This will be vital in the provision of joined-up treatment and expert centres and their local hospitals will need to invest time to develop understanding, trust and effective working relationships to achieve continuity of care for patients.

Patient support and patient representative groups should also be seen as key partners in the network between expert centres and their local hospitals to encourage specialists to seek to advice and support from the hugely valuable resource these groups represent.

8. In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

We have considerable doubts that the voice of people with rare diseases will be heard within the context of Local HealthWatch. Whilst we believe that Local HealthWatch has the potential to act as a powerful consumer champion, in order to perform this role for people with rare conditions, who may be referred to services outside of their local area, provisions must be put in place to allow Local HealthWatch to have oversight of specialised service with which they are co-located.

[2] *Services for people with Neurological Conditions* - National Audit Office, 2011; *Services for people with neurological conditions* - Public Accounts Committee, 2012

For patient support and patient representative groups, involvement in umbrella groups such as Regional Neurological Alliances (RNAs), which currently number 17, and the Neurological Alliance offers the most effective means by which to get their voice heard at a local and national level. The RNAs and Alliance will be seeking to establish strong relationships with Local HealthWatch and HealthWatch England and encourage recognition of the value of these and other such umbrella organisations as HealthWatch is established.

We also support the introduction of an explicit duty on HealthWatch England to engage with the views of specialised service users.

9. Do you agree that the United Kingdom should continue to participate in the Orphanet project?

We agree that the UK should continue to participate in the Orphanet project; in providing a source of reliable international information, it has an important role to play.

10. What sources of patient information and support are available which are not listed in this plan?

Patient support and patient representative groups are a significant omission; we reiterate the need for the health departments in all four nations to recognise these organisations as key partners and vital sources of information on which to draw to deliver the optimum outcomes for people with rare diseases. NHS Choices presents an ideal portal through which to signpost healthcare professionals and the public to the relevant organisations.

We would also encourage recognition of the value of engaging patients in their direct care.

11. Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?

We entirely agree that registers are an important tool in rare disease; if they are well designed, accurate and kept up to date, they are beneficial to both research and care and can provide a fundamental means of monitoring outcomes, evaluating services and treatments. Registers also perform a vital function for commissioners in the planning of services.

As the consultation acknowledges, however, registers do require funding and the establishment of national registers does not represent a viable financial option for the vast majority of patient representative bodies and support groups. Their existence for different conditions is, as such, patchy at present.

Where registers are created, we strongly support their administration on a national basis to enable the collation of data from across the UK which can constitute a common core dataset.

12. Are there any areas of work that the UK Plan on Rare Disease needs to pay particular attention to in order to advance equality?

We are concerned that the consultation does not address the issues around access to medicines for people with rare conditions. We remain unclear on how value-based pricing will benefit people with rare conditions given the relatively high cost of many drugs for rare diseases, particularly where they address unmet need. In respect of the use of off-license or off-label medicines, we are not confident of the ability of NICE to provide comprehensive and timely assessment.

To prevent inequity in early access to medicines for people with rare disease, we suggest the central provision of or direction on the purchase of these drugs, without which there is a real risk that variations in uptake with appear across and within the four nations. Early access to medicines schemes for rare conditions should also be considered further.

Advancing the equality of people with rare conditions to ensure that they will also be contingent upon:

- commissioners, the NHS and key structures allocating proportionate resource to research into rare disease
- providing timely, equitable access to appropriate services
- delivering integrated care and support across health and social care