Draft Northern Ireland Rare Diseases Implementation Plan

Response Form

Please provide your details before submitting your response (see the attached advice on the confidentiality of consultation responses).

I am responding:

As an individual	
As an organisation	X

(please tick the appropriate box)

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Responses must be received no later than 5pm on Monday 19 January 2015. Thank you for your comments.

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1. Do you think the actions to **empower those with rare diseases** will allow Northern Ireland to meet its commitments? If no, please provide details.

We welcome the involvement of medical professionals, universities, service users and carer groups in the development of the plan.

Some actions could be made more specific. The Northern Ireland Electronic Care Record (NIECR) does provide a basis for putting patients with rare diseases in charge of their own information, and development of a Patient Portal will be important.

It is very welcome that the Draft Plan calls for patients to have access to specialists elsewhere (mainly in the rest of the UK), but a critical component in the empowerment of rare disease patients is to ensure that broad-based generic services are available in Northern Ireland - for many disorders it may be unnecessary to travel to a distant

specialist service (or possibly only occasionally), but adequately resourced tertiary specialties are absolutely required on a regional basis. This calls for increased investment in Genetics Services among other specialised tertiary services dealing with rare diseases.

2. Do you think the actions to **identify and prevent rare diseases** will allow Northern Ireland to meet its commitments? If no, please provide details.

Access to carrier tests is unlikely to significantly impact this area unless patients within Northern Ireland have the same access to reproductive decision-making that is available in the rest of the UK.

The current law in relation to termination of pregnancy in Northern Ireland, particularly in the context of severe fetal abnormality (lethal and non-lethal) puts those in Northern Ireland in a different position to those in the rest of the UK, which could therefore negatively impact on this area of delivery of the plan. There is effectively no mechanism for prevention or prevention/mitigation of the impact of some of these very severe abnormalities on families. It is noted, however, that the Department of Justice in Northern Ireland is currently consulting on possible changes to the criminal law on abortion, in cases of lethal foetal abnormality.

The lack of clear commitment to developing the equipment, IT, bioinformatics and clinical infrastructure for Next Generation Genomics is concerning, as this is very clearly the direction of travel for the rest of the UK. The development of a Next Generation Sequencing core as the basis of a reconfigured overall service for Molecular Pathology provides the best way forward, and will serve patients much better than simply increasing existing services, pathways and practices.

3. Do you think the actions to **diagnose and allow early intervention** will allow Northern Ireland to meet its commitments? If no, please provide details.

The steps outlined are welcome, but could go further. Access to UK Genetic Testing Network (UKGTN) approved tests is important, but if rare genetic disease patients are to benefit from this plan, it is important to get a diagnosis as near to the start of the patient journey as possible, before embarking on a drawn-out, arduous and hugely expensive diagnostic process. We welcome the stress placed on education and training with a focus on early diagnosis and the exploitation of new genomic technologies.

We would urge some caution on diagnosis. By definition, rare diseases can be difficult to diagnose and the development of highly specialised centres for diagnosis and treatment needs to be balanced by measures to ensure that diagnosis is enabled in the less specialised settings where patients present. We understand that this difficulty has been recognised by the Rare Diseases Advisory Group and that a sub-committee is

meeting to produce some ideas.

It is important that Next Generation Sequencing capacity is at the forefront of genetic diagnostics moving forward in Northern Ireland. This will largely remove the need to send genetic tests elsewhere as the majority of genetic testing will be done in Northern Ireland, resulting in faster results and less money leaving the system.

Reference is made to the 100,000 Genomes Project (100KGP). Discussions are on-going as to how Northern Irish patients can access this, and following the Scottish model, it would seem that the only way would be for Northern Ireland to have its own Genomic Medicine Centre (GeMC) and for Government to identify the funding required to equip and operate this, and also to link in with the wider 100KGP.

Developing expertise in Northern Ireland, rather than just funding piecemeal UKGTN tests, may prevent unnecessary expense and put patients on the same footing as those in the rest of the UK. Developing expertise will also positively impact on research and future service development. The commitment to registries (Point 20.1; patient databases that proactively identify and input into patient management) is particularly welcome, but needs a very clear statement that resources will be provided to manage these, as they are active systems, not merely records.

Under point 14.1, a specific platform is mentioned as being trialled. We would welcome more information as to why this particular platform has been singled out. It may have been better to have indicated: '*As part of the Northern Ireland eStrategy, consideration will be given by the HSC to incorporate rare diseases using this or a similar type of platform*'.

4. Do you think the actions to **co-ordinate care** will allow Northern Ireland to meet its commitments? If no, please provide details.

There needs to be a clear commitment to managing patients efficiently by avoiding unnecessary multiple appointments. However Service Budget Agreements for specialised services such as Genetics are usually assessed on outpatient attendances. This creates a disincentive to collaborative multidisciplinary working, as other clinical practice (phone clinics, video conferencing, interacting with patients via email, etc) can be ignored in the final figures.

The intention to collaborate widely within the UK, the EU and internationally in delivering the plan is welcomed. Modern communication channels potentially allow specialists in Northern Ireland to share their expertise with centres in the rest of the UK, and vice versa for the benefit of patients.

The plan recognises that the delivery of low volume services may require collaboration with providers outside Northern Ireland and we agree that a four country dimension of strategic planning for these services should be maintained.

"Mainstreaming" of rare disease expertise out to primary and secondary care will become increasingly important, and the locus for the knowledge base required to do

this will necessarily focus around the patient and agreed protocols. The NIECR provides a logical hub for this, and in that respect Northern Ireland is ahead of the rest of the UK. We welcome the commitment in the Draft Plan (point 19.1) to build on this resource.

The development of a comprehensive rare disease registry in Northern Ireland (point 20.1) is a positive step, which our experts consider a prerequisite for any modern service and will facilitate not only clinical care and audit but also population based planning and clinical research. NI already has a cancer registry which has had considerable investment over the years, and that should be the model for rare diseases. (http://www.qub.ac.uk/research-centres/nicr/CancerData/) Consideration should also be given to the future establishment of a database that could cover all of the island of Ireland particularly as complex extended families may live in the other jurisdiction.

The mention of transitional care from paediatric to adult services is particularly welcome although it will be challenging. Liaison with the Northern Ireland Rare Disease Partnership and patient groups is also welcome, and will be important in driving this forward.

5. Do you think the actions on the **role of research** will allow Northern Ireland to meet its commitments? If no, please provide details.

Yes. There is a serious need to study and break down the barriers that currently prevent rare disease patients participating in research. These include: lack of timely diagnosis, lack of clinician time to do research or to recruit patients, lack of awareness of specific projects, difficulty in accessing assistance in the form of research nurses, the slow process of research proposals through Research Governance & Ethics, and differences in consent legislation between the rest of the UK and Northern Ireland. There needs to be clear delivery on the commitment to foster research, to provide patients with access, and to make sure the benefits flow back to the patients themselves.

There also needs to be a clear commitment to the development of Next Generation Sequencing (gene, panel, exome, genome) within Northern Ireland with a transition to clinical service provision. The response to point 42 could be much stronger. Northern Ireland has an opportunity to participate at the same level as the rest of the UK, and this would clearly be to the benefit of patients.

6. Do you agree with the Departments findings as set out on in Section 10 (Equality Screening) of this consultation document?

Yes.

a) Are the actions/proposals set out in this consultation document likely to have an adverse impact on any of the nine equality groups identified under Section 75 of the Northern Ireland Act 1998? If yes, please state the group or groups and provide comment on how these adverse impacts could be reduced or alleviated in the proposals.

b) Are you aware of any indication or evidence – qualitative or quantitative – that the actions/proposals set out in this consultation document may have an adverse impact on equality of opportunity or on good relations? If yes, please give details and comment on what you think should be added or removed to alleviate the adverse impact.

c) Is there an opportunity to better promote equality of opportunity or good relations? If yes, please give details as to how.

d) Are there any aspects of this action plan where potential human rights violations may occur?

7. Are there any **other actions** that should be considered?

With this Plan, the 100KGP, the NIECR, a highly motivated rare disease patient group, clinicians and their services, and other stakeholders; there is a chance to make a big difference to the lives of patients with rare diseases in Northern Ireland. The plan

recognises that successful delivery will require partnerships between Government, clinicians, researchers, industry and patient groups.

Investment in core genomic diagnosis is necessary to ensure that health professionals and others can access information to help those with rare diseases.

When the final plan is being prepared for implementation we would encourage the Department to consult again with both patients and professionals.

We welcome the intention to publish annually a report on progress against the plan commitments.