Introduction

1. Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all who need them. We actively support research and innovation across the field of genetic medicine.

2. Rare Disease UK (RDUK) is a multi-stakeholder campaign run by Genetic Alliance UK, working towards the delivery and implementation of the UK Strategy for Rare Diseases\(^1\), published by the Department of Health in November 2013.

3. RDUK’s Patient Empowerment Group works closely with Public Health England to ensure patient input and oversight in the development of public and patient information for the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS).

4. Data use, sharing and governance are critical issues in the rare and genetic disease arena. In this document we discuss approaches for governing use of patient data in NCARDRS and make our own recommendation for an approach supported by the patient community.

5. This document has been reviewed and approved by the Rare Disease UK Patient Empowerment Group.

The value of NCARDRS to the rare disease patient community

6. The scale of the challenge to the NHS that rare, genetic and undiagnosed conditions present is difficult to ascertain. Currently 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. (Where it is possible, it is usually due to independent data collection by a patient organisation or research project.) Far less is it possible to tell how old patients are and where they live.

7. Without this basic information it is impossible for the Government health departments in the UK and their NHSs to plan effectively and build a health service that is suitable for delivering healthcare for the patients that Genetic Alliance UK and its Rare Disease UK campaign represent.

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8. NCARDRS can deliver this information. The visibility and ability to plan alone will be step change for our community, but the full benefits that such a store of information could deliver are difficult to predict. In short, NCARDRS has the potential to be the biggest influence on improved healthcare for patients with rare diseases in England (and hopefully the rest of the UK) in a generation.

9. The value of NCARDRS to the broader rare disease community is shown by its significant utility for the implementation of the UK Strategy for Rare Diseases.

   In November 2013, the Department of Health published a UK Strategy for Rare Diseases, in this strategy all four Governments of the UK committed to:

   Commitment 7: “Support patients to register on databases, where these exist.”

   Commitment 18: "Standardise data collection, building upon existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.”

   Commitment 29 “Improve systems to record genetic and other relevant information accurately to record the incidence and prevalence of disease and support service planning and international planning.”

   Commitment 49: “Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.”

   UK Strategy for Rare Diseases, November 2013

10. These commitments recognise the important role that data sharing and research has to play in addressing the high level of unmet medical need within the rare disease community. With such small patient populations it is important to gather information from as many patients as possible in order to gain the best understanding of a condition.

The rare disease patient community’s attitude to data sharing

11. Many rare diseases are severe and life-limiting. For individuals or families affected by most rare diseases, the day-to-day challenges of managing a severe condition are made worse by the absence of an effective treatment or cure. These patients look to research as the source of new therapies to address their unmet health need. In order for progress to be made, patients recognise that the rarity of their conditions means that research relies on the effective sharing and use of their medical data, nationally and internationally.

12. Patients in the rare disease community therefore tend to be familiar with the need for their data to be shared and used widely. Given the small number of individuals affected by these conditions, patients recognise that collecting as much data as possible is vital to help research into the prevention, cause and treatment of rare conditions. Indeed we have anecdotal evidence of patients’ and patient organisations’ frustration at not having their information shared with researchers due to overly restrictive consent requirements.

13. Rare disease patients recognise that there is an inherent risk that they could be identified personally from some of their data. Despite this, rare disease patients are willing to share their medical data in order to drive research². For these patients, not sharing data would be detrimental to research efforts and as a result, to potential scientific advancements that could improve their quality of life.

Through an online engagement project, ‘My Condition, My DNA’, supported by the Medical Research Council, the Wellcome Trust and British In Vitro Diagnostics Association, Genetic Alliance UK sought the views of patients affected by rare and genetic conditions, both diagnosed and undiagnosed, on genome sequencing. When we asked patients what they thought about the use of genomic data for research, 93 per cent of participants said that they would want their genome sequences to be used.

When we further asked patients and families whether their views on sharing their genomic information for research were dependent on the type of organisation that would be undertaking that research, around two thirds of respondents were happy for the NHS (80 per cent), universities (77 per cent) and charities (63 per cent) to use their data for research purposes. The fact that the majority of respondents supported genomic research within the NHS is a clear endorsement from our patient community that they welcome current and future initiatives that put the NHS at the heart of this type of research.

Our study also revealed that patients feel that it is important to have control over their genomic data and how it is used for research purposes. Patients supported the use of dynamic consent in a research setting in the same way as they value autonomy and choice about how their genomic data will be used and what they find out from it in a clinical context.

Consent is a vital part of the collection of genomic data for research and clinical use, it relies on patients ‘opting in’ to have relevant tests and to donate samples. Thereafter, the use of genomic data is an ongoing process between the patient and clinical and research stakeholders.

We believe that a consent based approach to collection of data for NCARDRS is not appropriate, and support an approach enabled by Section 251 of the NHS Act 2006:

Informed consent is the fundamental principle governing the use of patient identifiable information within health or social/community care research. It is recognised that there are situations where informed consent cannot be obtained. In England and Wales, Section 251 of the NHS Act 2006 (originally Section 60 of the Health and Social Care Act 2001) provides the statutory power to ensure that NHS patient identifiable information needed to support essential NHS activity can be used without the consent of patients. The power can be used only to support medical purposes that are in the interests of patients or the wider public, where consent is not a practicable alternative and where anonymised information will not suffice. Caldicott Guardians in Scotland and Medical Directors in Northern Ireland make decisions on the same basis.

In light of the value that data collection and the sharing of this information brings to the rare disease patient community, and given the willingness of the community to share medical data for research, Genetic Alliance UK supports a model of consent that would enable data collection from as many patients affected by rare conditions as possible. We would therefore support the use of the statutory power defined in Section 251 of the NHS Act 2006 for data collection for NCARDRS.

The key issue that informs this view is that of how complete a data set NCARDRS can create. It is crucial to the value of the system that it contains as complete a picture as possible. Small gaps in a data set with a remit to track rare diseases could lead to major inaccuracies in our understanding.

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of rare disease in the UK. We understand Public Health England are consulting with statisticians to understand the tolerance for opt-outs or non-consents, and will be interested to see this information, though we expect the tolerance to be low.

21. This position is also informed by the knowledge that this implementation of the Section 251 power is expected to allow for an informed opt-out. Those members of the rare disease community that do wish to remove their data from this record, or proactively decide not to have it recorded will be able to register such a decision with Public Health England. We agree that those choosing to do this should be made aware of the possible impact removal of such data could have on their own health care planning and that of others with the same or similar diseases.

22. In implementing the informed opt-out, Public Health England should take particular consideration of the fact that many individuals with rare diseases will have been added to the register in the early years of their lives. Patients should therefore be informed on a regular basis that their information is being held in this register, as part of regular awareness raising of the existence and the value of this register. Consideration should likewise be given to those patients who might not gain competency in adulthood, and measures should be implemented to take account of such individuals’ wishes and / or those of their guardians.

23. The other models of consent that we have examined, such as informed consent (opt-in) or dynamic consent, are valuable in other healthcare and research scenarios, but not appropriate for this use of data.

Safeguards and gaining patient trust
24. There has been much public debate about the sharing of medical data for research in recent years and concerns over data security, privacy and access. This has included the electronic sharing of patient information collected in general practice as part of care.data, and more recently has considered the potential use and abuse of the genomic data that will collected as part of the 100,000 Genomes Project.

25. It is essential that there are clear, functional systems in place to facilitate the collection, analysis and sharing of data whilst reassuring those that have their data collected through NCARDRS that their data will be stored and shared safely and accountably. Public Health England should work to strengthen patient and public confidence that the data they share is used ethically in order to improve patient care.

Conclusion
26. Patient data is of critical importance to quality improvement, service design, public health and medical research in the NHS. Genetic Alliance UK supports a model of consent that maximises the collection of data from as many patients affected by rare conditions as possible.

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