



Genetic Alliance UK
Supporting. Campaigning. Uniting.

Consultation Response

NHS Future Forum

How to improve information for users of services and professionals

Response by Genetic Alliance UK

1. Genetic Alliance UK is the national charity supporting all those affected by genetic conditions. Genetic Alliance UK aims to improve the lives of people affected by genetic conditions by ensuring that high quality services and information are available to all who need them. Our membership represents more than 140 voluntary organisations working for a wide range of conditions, many of which pose complex health and social care needs.
2. A baby with a genetic condition is born every half an hour in the UK; of these only 4 in 10 will have their condition cured or ameliorated, the rest will die or live with a lifelong chronic condition. Most of the patients and families supported by our members are frequent users of the NHS and require good quality coordinated care from a wide variety of local, regional and national centres in many areas of specialisation.
3. Rare Disease UK is an initiative of Genetic Alliance UK. It is a multi-stakeholder organisation which brings together patients, healthcare professionals, academics, industry and commissioners to promote strategic planning for rare diseases in the UK. Rare Disease UK will publish its recommendations for a strategy for rare diseases on the 28th February 2011. Rare Disease UK has submitted a response to this consultation, which should be considered as complimentary to this submission. This response contains quotes from the “Experiences of Rare Diseases: An Insight from Patients and Families” (2010) publication by Rare Disease UK, which illustrate patients’ views on information value and provision.
4. We are grateful for the opportunity to comment on this consultation.

What information about health and care do you need – and in what form?

5. There are more than 6000 rare conditions, of which the vast majority are genetic conditions. Information for patients diagnosed with a rare condition, or for a couple who are aware that they are at risk of having a child affected by a genetic condition is scarce. No healthcare professional can possibly recognise or understand every rare genetic condition. At diagnosis patients may be told, often incorrectly, that they are the only case in the country, or one of twenty in the world. This information does not empower patients, it isolates them.
6. At diagnosis patients with a rare condition would like to know the same information as a patient with diagnosed with a common condition: how their ability to work, have children, care for their children will be affected; how their daily routine will be affected; how long they be able to continue to drive; etc. This information is much harder to obtain for those affected by rare genetic conditions.

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7. In some cases, the information simply does not exist; but in many cases the information is out there, but access requires effort and resourcefulness from a healthcare professional. The simple recognition of how frequently rare conditions occur, 3.5 million people in the UK will be affected by a rare condition in their life, is a first step for health professionals that can help patients with rare conditions.

“Any [information] would be great, I’ve not had one leaflet. My baby has no skin or skull, and I leave hospital, without even a health visitor’s home appointment, support group, nothing!!”

Mother of child with multiple complex rare conditions¹

What works well already? What doesn’t work?

8. There is currently a dearth of information on rare genetic disease in the NHS. In some cases, patients are left to their own devices after their diagnosis, because their GP does not know what to do next. For every GP who works hard on their patient’s behalf, and researches the most valuable next steps and appropriate specialists, there is a GP who does not fulfil their role as their patient’s advocate in the NHS, and leaves them adrift and without access to tertiary care.

“[We received a] phone call for diagnosis on New Year’s Eve with the comment from GP ‘I don’t know anything about it go on the computer and look it up’. We didn’t have a computer!!!”

Relative of a patient with myasthenia gravis¹

9. For GPs to fulfil their responsibilities to all their patients, they should recognise the frequency of rare disease in the UK (3.5 million people in the UK will be affected by a rare disorder at some point in their life), be prepared to admit that they do not have knowledge of a rare condition, and understand the best route by which to obtain valuable information for their patient.
10. GPs and other healthcare professionals should consider patient organisations as potential sources for information on rare genetic conditions. Patient organisations do not only provide information relevant to patients, they can be the sole source of information on a particular health condition, and are frequently capable of advising clinicians on palliative care, diagnosis and natural history of conditions.

“All of our information and research comes via the Lowe Syndrome Association in the USA. Diagnosis was made possible by development of a reliable biochemical assay by the Lowe Syndrome Association.”

Mother of a child with Lowe syndrome¹

“Support groups, helplines and newsletters from the Pituitary Foundation have been invaluable for information before my operation and after diagnosis and treatment.”

Patient with craniopharyngioma¹

How can we open up access to information and support people to use it?

11. All those who contribute to healthcare needs of a patient: clinicians, nursing staff, administrative staff, researchers, lab technicians, carers, parents, and patients themselves, benefit from access to good quality information. All these information needs are important; to prioritise one form of information over another or one information user over another would be wrong. It is important to

recognise the differences between the different participants in the healthcare pathway, and to ensure they can access the information relevant to their role.

12. Extraneous information can be upsetting in some contexts, and it can replace appropriate information in others. This is an area in which some healthcare professionals need to work to understand the most relevant and appropriate information that a patient and/or their carer require in their short appointment.
13. The majority of the thousands of genetic conditions that affect patients in the UK are rare. Little is known regarding their impact on patients and their incidence is not recorded by the NHS. This lack of information collection hampers research and effectively hides a health burden from commissioners. Where registries have been set up to monitor the incidence of rare genetic conditions, (for example the Registry for Mucopolysaccharide and Related Diseases founded by Genetic Alliance UK member The Society for Mucopolysaccharide Diseases (MPS) in 1980) they have facilitated the collection of data on the incidence of the rare genetic conditions and created a network of patients that both facilitates research and provides evidence to stimulate research.
14. The benefits which could be delivered by coordinated information collection by the NHS on the incidence of all rare genetic conditions are enormous. Commissioners would better understand the populations they serve, and be able to better plan care provision. Valuable research into unmet health needs would accelerate and be facilitated.

How can we ensure that information supports improved care and better integration of services (for example, commissioning, research, clinical audit, public health) whilst protecting patient confidentiality?

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17. Genetic healthcare records are unusual in the NHS in that they refer to more than one patient, if not explicitly then implicitly. A record stating that a patient has a condition which is inherited in an autosomal dominant pattern states that one of their parents also has the condition. A record stating that a patient has a condition which is X-linked states that his mother is a carrier, and that his sister has a 50% chance of being a carrier of the condition. These examples are myriad, but the principle is clear, this information is familial and must be recognised as such.
18. To account for the special nature of genetic health records, they are treated differently in the NHS, and are usually held separately from the standard NHS record at the patient's regional genetic service.
19. Genetic Alliance UK supports the current actions of regional genetic services, and agree that genetic information is different from other medical information due to its familial nature. We would argue though, that genetic records are not incredibly different from other records and do not require complete isolation. If that were the case, patients with genetic disorders would be isolated from the vast majority of benefits that information technology could bring. We call for

special attention to be given, and for sensible, proportionate measures to be introduced to address the added sensitivity and power of genetic healthcare records.

What help do you need to make best use of the information out there?

20. Information attached to and contained within patient records can be complicated and contain a lot of technical terms. Information in this form does very little to empower the majority of patients. Most patients will require support to interpret the information contained within their record.

“I understood some of the contents, but most of the information would have been understood by someone with a medical background.”
A patient with Langerhan’s cell histiocytosis¹

21. Doctors must interpret and communicate the complex information in a patient’s health record and they must find time to answer patient’s questions. Our members’ experiences of appointments with their GPs and specialists are highly varied. Some doctors do not afford patients the time and respect necessary to empower them with information and allow them to be equal partners in decision making. A culture change will be necessary for many doctors in the NHS, and training will be required to improve many doctors’ communication skills.

“Was told the bare minimum in a five minute chat most of which turned out to be incorrect.”
Relative of a patient with bilateral and chronic uveitis¹

“My GP was insistent, despite my protests, that the attacks were due to postnatal depression and had prescribed a cocktail of drugs to no avail. I was desperate.”
A patient with multiple endocrine neoplasia 2a¹

How can we ensure information is available that enables people to take more control of their own care and enable shared decision-making?

22. Patients look, in the first instance, towards their doctor for guidance on the impact their choices will have on their health. Without professional assistance, choice is not always valuable to patients, it can become a stressful addition to their healthcare experience.
23. For most patients with a rare genetic condition, choice is either not possible or not appropriate. For most patients in these situations, their sole preference is for the best possible treatment. Healthcare professionals should be equipped and prepared to assist patients in finding and understanding their treatment options and then to assist them in accessing this care.
24. Patient organisations, as we have explained elsewhere in this document, are a valuable information resource. They are able to provide condition specific information to patients on the issues that are most important to them.



Director
Genetic Alliance UK

1. Experiences of Rare Diseases: An Insight from Patients and Families, Limb et al (2010)
www.rare-disease.org.uk/documents/RDUK-Family-Report.pdf