



**Genetic Alliance UK**  
Supporting. Campaigning. Uniting.

## Consultation Response

Department of Health

Liberating the NHS: An Information Revolution

Response by Genetic Alliance UK

### Introduction

1. Genetic Alliance UK (formerly Genetic Interest Group) 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 130 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. We are grateful for the opportunity to comment on this consultation. This response has been endorsed by the trustees and members of Genetic Alliance UK.
4. 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 28<sup>th</sup> February 2011. Rare Disease UK has submitted a response to this consultation, which should be considered as complimentary to this submission; the Rare Disease UK submission describes elements of the recommendations pertinent to the information strategy. 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.

### Information for patients with genetic conditions

5. Information is vital to patients. As this consultation document has recognised, information plays a key role in every step a patient takes through their healthcare journey. This is particularly true for patients with genetic conditions, due to their familial nature and, for those genetic conditions which are rare and poorly understood, because information is scarce.

6. 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.
7. 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.

#### Information for patients with rare conditions

8. 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.
9. 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.
10. 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 conditions<sup>1</sup>

## 1. An information revolution

### Question 1: What currently works well in terms of information for health and adult social care and what needs to change?

11. 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 gravis<sup>1</sup>

12. 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<sup>2</sup>), 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.
13. 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 syndrome<sup>1</sup>

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

Patient with craniopharyngioma<sup>1</sup>

## Question 2: What do you think are the most important uses of information, and who are the most important users of it?

14. 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.
15. 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.
16. 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.

17. 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.

**Question 4: Given the current financial climate, how can the ambitions set out in this consultation – to make better use of information and technology to help drive better care and better outcomes – be delivered in the most effective and efficient way?**

18. The information revolution should be seen as investment in efficiency and improvement. Patients with complex genetic conditions frequently describe their experience of poor understanding of their condition from healthcare professionals, and poor signposting and poor diagnosis in the NHS; all of this leads to inappropriate interventions and ineffective episodes of care provision. Timely information, delivered in the correct context can cut wastage of this sort drastically.

19. There are savings and efficiencies that can be made with recognition of good quality work already being carried out. Patient organisations are a valuable source of good quality information both for patients and for healthcare professionals, particularly in the case of genetic conditions where added familial context is often necessary and in the area of rare diseases where patient organisations possess the most comprehensive and highest quality information in many cases.

20. Accreditation schemes such as the new Information Standard should work to facilitate the accreditation of small organisations which provide valuable information to their members. Current schemes are time-consuming to administer, a hurdle which can become a barrier to small, often voluntary, organisations achieving accreditation. The Information Standard, for example, currently requires onerous protocols which effectively rule out small voluntary organisations; their current facilitation methods are valuable, and should be continued, but do not solve the issue of the large time investment necessary to implement their requirements.

## 2. Information for patients, service users, carers and the public

**Question 6: As a patient or service user, would you be interested in having easy access to and control over your care records? What benefits do you think this would bring?**

21. Most genetic diseases are multifactorial, affecting a number of different systems in the patient's body. Care for most of these conditions is provided by healthcare professionals who are specialists in the treatment of each particular body system. Coordination of care for cases such as these is the best solution, but in the vast majority of cases, no care coordinator is available. In these situations patients are frequently frustrated either by the necessity of repeating their story to every healthcare professional they meet, or because their care record has not been forwarded properly from the previous specialist.

“My husband’s care is split between two hospitals... One never requests records from the other so tests are duplicated and delays blamed on each other. No one takes overall responsibility and there is no one person to refer to for even simple requests let alone really important ones. Repeated requests for information go unanswered.”  
Wife of a patient with multiple myeloma<sup>1</sup>

22. Greater access and control to patient records will contribute to a reduction of this frustration, but this will essentially cast the patient themselves in the role of the care coordinator. Patient access and control of their records should not be seen as a substitute for expert care coordination.

**Question 7: As a patient or service user, in what ways would it be useful for you to be able to communicate with your GP and other health and care professionals on-line, or would you prefer face-to-face contact?**

23. On-line communication with healthcare professionals has traditionally been discussed in the context either of care for very remote communities who may live a long way from their GP, or of care for rare conditions, where specialists may be located at a great distance from the patient.

24. Face-to-face contact, in our experience, is the preferred medium for communication between patients and their healthcare professionals. This is certainly the case for local healthcare providers such as GPs. The use of such technology should not replace face-to-face communication unless it is necessary due to circumstances such as remoteness or distance from specialised care, or it is the preferred option of the patient.

**Question 8: Please indicate any particular issues, including any risks and safeguards, which may need to be taken into account in sharing records in the ways identified in this consultation document.**

25. Genetic health records are currently kept separately from a patient's NHS record. They are maintained by the patient's genetic service in recognition of the special nature of genetic information and to keep secure information that has implications for a wider familial group than would usually be the case for medical information.

26. This special nature can be illustrated with the example of Huntington's disease. A child whose parent has the disease has a 50% chance that they too have the condition. Huntington's disease is a neurodegenerative life-limiting condition, the symptoms of which become apparent in middle age. It is a fully penetrant condition, which means that if you have the gene, you will get the disease. Many people who are at risk of the condition choose not to have the predictive test, and would rather live with a 50% risk than a 100% certainty. If a grandchild of patient with Huntington's disease decided to have a test, and the grandchild's parent has not, a positive result would have significance for both the grandchild and the grandchild's parent.

27. 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 the information revolution 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.

28. 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 histiocytosis<sup>1</sup>

29. Doctors will play a key role in the information revolution. They 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 uveitis<sup>1</sup>

"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 2a<sup>1</sup>

30. Paragraph 2.14 of the consultation document predicts the development of a market of products to package information and assist patients with the management of their own healthcare. Genetic Alliance UK expects these products to be free, as any cost attached to a patient's management of their own healthcare will create multi-tiered access to appropriate healthcare within the NHS, a situation which is in opposition to the NHS's founding principles.
31. As information is transferred to participants in this predicted marketplace of information management products, it leaves the NHS, and resides with a third party organisation. There should be a code of conduct for these organisations to ensure that they do not use patient information for any purpose the patient is unaware of, the lifespan of the information possession is clear, and all privacy implications are stated explicitly.

#### Question 9: What kinds of information and help would ensure that patients and service users are adequately supported when stressed and anxious?

32. Diagnosis is a key point in a patient's healthcare experience. This is the stage at which there health problems should be explained, and the implications and future decisions laid out. For many genetic diseases the implications are severe, and future treatments are scarce, ineffective, non-existent, or not available on the NHS. In these situations patient organisations can be extremely valuable sources of advice and support. Healthcare professionals should make themselves aware of appropriate patient organisations and other support organisations, and explicitly direct patients to organisations that are able to support and assist.

"We were informed in Yorkshire that the drug was a red drug and as such could only be prescribed by a hospital consultant. This resulted in my husband knowing there was a drug which would help him but having to liaise between a consultant and GP, a very upsetting experience."

Wife of a patient with myasthenia gravis<sup>1</sup>

33. Equally, failure to diagnose a condition is a distressing point in a patient's healthcare experience. A lack of a name to attach to their healthcare problem can leave a patient struggling to explain

themselves to employers, social support services, and benefit providers. Access to palliative care can be difficult without a name of a condition to open doors. Patients in this situation need information which will help them access useful palliative care services such as pain relief and social care, without becoming reliant on Accident & Emergency services as some undiagnosed patients must.

34. Diagnosis can often bring or come with devastating news, a short life expectancy or the loss of a child for example. In these cases, it is frequently impossible for a patient to absorb information, and it may be a number of days or weeks before they are in a position to receive information. There should be mechanisms within the NHS for a patient to receive information in stages, and at their own convenience.
35. The consultation document refers to NHS Choices as an important component of the future of information provision in the NHS. Our members have reported a number of problems with NHS Choices in the few years it has existed; these have included inappropriate images on pages describing health conditions, inaccurate information, and problems with changing, updating and extending information due to budgetary constraints.
36. As information becomes more central to the service the NHS provides to patients, it is important that NHS Choices is maintained and funded properly. As we have stated earlier, patient organisations, especially in the case of rare diseases, frequently have the best quality information available in the UK; these organisations should be clearly signposted from NHS Choices, and their complaints should be taken seriously.

**Question 10: As a patient or service user, what types of information do you consider important to help you make informed choices? Is it easy to find? Where do you look?**

37. As we have stated in our response to “Liberating the NHS: Greater choice and control” 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.
38. A second pertinent point from our response to “Liberating the NHS: Greater choice and control”, is that 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.
39. 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.

### 3. Information for improved outcomes

**Question 17: For which particular groups of service users or care organisation is the use of information across organisational boundaries particularly important?**

40. Services currently commissioned at a national and regional level are vital to our members. Of the 45 services commissioned nationally, 6 are services for patients with genetic conditions, and 27 of the remainder frequently treat those affected by genetic conditions. Of the 34 defined services in the Specialised Services National Definition Set (SSNDS), currently commissioned on a regional basis, 4 are services for patients with genetic conditions, and 28 of the remainder

frequently treat those affected by genetic conditions. Many patients with genetic conditions will need to use more than one of these services.

41. Since these services are in the main provided at one of a small number of specialist centres, it is very likely that patients will have to travel across organisational boundaries. Information exchange between these specialist centres and local healthcare provision will ensure all those involved in a patient's healthcare provision are aware of all components of their patient's care.



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