



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

## Consultation Response

Department of Health

Liberating the NHS: Greater choice and control

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.

### A patient's relationship with their doctor

4. GPs will play an enormously important role in facilitating their patients' access to choice and control of their healthcare. Our members' experiences of their GPs' attitudes to them as patients with rare and/or intractable conditions vary tremendously. For every GP who fully delivers on their role as a patient's advocate within the NHS, there is a GP who leaves their patient adrift in the system; or even worse acts as a barrier preventing access to specialised care.
5. Our members frequently report that their GP will not listen to their requests for access to diagnostics and/or specialised care. Patients in this situation are less concerned with choice, than with accessing healthcare at all. In many of our members' situations there will not be a choice of care provider or treatment plan, there will be one service for their condition, and many are struggling to access this.

“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>

“Many meetings with medical professionals become us informing them about the condition!”

A relative/carer of a person with non bullous erythrodermic ichthyosis<sup>1</sup>

6. Though this consultation states:

2.1 “Everyone should have choice and control over their care and treatment, and choice of any willing provider wherever relevant.”

7. It is later explicitly stated that GPs will retain the ability to block patients’ access to the care they consider to be most appropriate to them:

2.11 “It will be for the healthcare professional making the referral to decide what is clinically appropriate.”

8. While Genetic Alliance UK does not advocate a complete revocation of controls ensuring that a patient is accessing healthcare appropriate to them, we do not see these proposals contributing significantly to a solution solving the problems that our members can frequently face. These issues can be resolved by GPs engaging in a more balanced dialogue with patients, accepting evidence that patients bring to their appointments, admitting that they have not had experience of rare genetic conditions, and taking the time to understand their condition.

## Care planning

9. We are pleased to see recognition in this consultation document of the large population of patients with long term conditions, the large proportion of these that suffer from comorbidity, and the importance of care planning for these patients. Coordinated care is essential for the majority of genetic conditions which require multifactorial care from multiple care providers; and is vital for those affected by rare genetic conditions, as care providers are unlikely to have encountered these conditions previously in their career.

10. We believe the estimate that 70% of patients currently have care plans does not apply to the population represented by Genetic Alliance UK. The proportion of patients represented by our members that have a care plan is much lower, and we would hope to see this increased as a priority. We refer the Department to the publication of the recommendations towards a strategy for rare diseases which will be published in February 2011 by Rare Disease UK, an initiative of Genetic Alliance UK.

“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>

## Shared healthcare decisions

11. It should be recognised that many patients will not wish to exercise greater control and choice in their healthcare. Patients who wish to rely on their healthcare provider to design a care plan for them, and choose the best possible care for them, should be allowed to do so, and should not have to make decisions they do not wish to or do not feel able to.
12. Healthcare providers should ensure that all patients are fully informed of their options regarding choice. This requires communication rather than just information provision. Health professionals should be sure that their patient has understood where they have an option to choose, and how they may exercise this right; but should not assume that the patient will choose to choose.

“In my role as a Researcher in healthcare lots of people tell me they do not know what to do for the best when presented with choices about treatment options. For example, a patient diagnosed with breast cancer was asked what she wanted to do i.e. chemotherapy before surgery, or after surgery. Shock and the necessity to begin treatment as soon as possible made it too difficult to comprehend how to decide and instead she asked the Consultant what they would do in her situation. This sensible question led to her receiving the best chance of successful treatment.”

Anna Allford, Project Officer, Genetic Alliance UK

13. For all patients to be able to access choice within the NHS there should be recognition of the necessity for more communication between patient and healthcare professional, and that improvement and greater investment will be necessary in support services for communication with those patients who do not have English as a first language.
14. Language and literacy are not the only barriers to accessing choice within the NHS. A patient’s level of health literacy will affect their confidence to engage or disagree with healthcare professionals. For these initiatives to be successful and equitable, those patients with poor health literacy should be supported by independent advocates, and efforts should be made to increase the population’s health literacy.

## Value of patient organisations

15. We are pleased to see recognition of the assistance and support that patient groups can provide in this area. We believe this recognition should be broader; patient groups can provide information, care planning assistance and further guidance alongside assistance in understanding information and making choice. We believe the support patient groups can provide to the health service and social services should be recognised, and where applicable projects developed where patient organisations can be funded to deliver this extra value for patients with specified healthcare needs.

“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>

“All support given comes via TSA [Tuberous Sclerosis Association] - clinic, online sites, research, written info, family liaison workers etc.”  
Relative of patient with tuberous sclerosis<sup>1</sup>

### Choice for those with rare genetic conditions

16. For those with rare conditions, choice is not always possible, and is not always appropriate. There may be one single specialist in England who is capable of providing care for people affected with a particular condition. In this situation, there should be no choice, patients in this situation should be treated by the expert in their condition.
17. In these cases, when choice is offered, patients with rare conditions will reply: “where can I access the best care?” Healthcare professionals should be able to judge when their patient’s care is best given by a specialist, and they should be able to identify the appropriate specialist and assist their patient to access them.

“I tried having highly specialised blood tests taken and analysed at a local hospital and then getting my results at the specialist oncology centre I attend over 100 miles away. This was not useful for that centre or myself (despite what had seemed more convenient) as these extremely sensitive tests can only show changes in my disease status if consistently accurately measured by the same lab over a period of time.”

Rare cancer patient

18. Choices should never be offered that could damage the quality of care provided, expert advice should be sought to avoid this from occurring.
19. Travel is a key issue for those with rare diseases. Patients may live many hundreds of miles from the specialist they most want to access. Choice is valuable here. Many patients will consider a face-to-face meeting as essential and will not balk at the many hours of travel necessary, others will prefer to make use of new technology and use a local consultant who is in liaison with the appropriate specialist. Patients should be supported as much as possible in this choice, and regional and administrative boundaries should not become factors in their decision.

Director  
Genetic Alliance UK

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)