



Working to benefit all people affected by genetic disorders

Introduction

This consultation response to the Patients Rights Bill is based on the views of 60 patients, carers, charity workers and healthcare professionals, all with experience of genetic conditions. It is the result of a consultation event ran by the Genetic Interest Group Scotland on the 9th of December 2009.

The right to accessing healthcare, the right to proper communication regardless of background and the right to information were voted as the most important facets of the bill.

The importance of improving access and communication for vulnerable groups of patients such as those with rare and complex conditions, learning difficulties, physical disability and for people whose first language is not English were themes that emerged throughout the day. There was also the consensus view that patient participation was lacking and that NHS Scotland was missing out on the invaluable resource of patient experience.

In this response, direct quotes are highlighted in blue and were offered by patients or their direct carers unless followed by "HCP," the abbreviation we use throughout to refer to healthcare professionals, including genetic counsellors, specialist nurses, clinical geneticists and laboratory scientists working in Scotland.

Q1. The Right to Access

Key points

- Patients welcomed a reduction in waiting times. They were concerned that the use of the criterion “referral to treatment” in the bill was too narrow. Patients affected by genetic conditions principally interpreted the term “access” to imply the right to an appointment with an informed (specialist) health professional.
- Collectively, patients had experienced huge inequality in terms of access to services. Patients commented that the referral process felt like a “postcode lottery.” Healthcare professionals (HCPs) argued that more trained staff would resolve the issue.
- The first barrier experienced by most genetics patients was one of transport or physical access to the hospitals.
- Patients argued that access to social care services should be offered through the health service and that NHS Scotland should work closely with Social Care services to deliver this.

Suggested entitlements

1) *The right to a waiting time guarantee for “referral to treatment”*

Many genetic conditions are chronic and require long-term management, often under the care of numerous services, rather than discrete episodes of treatment, and patients are concerned that these should be managed effectively to allow care coordination and continuity. Furthermore, some patients seen at Genetics Clinics don't have a confirmed diagnosis for much of their lives, so treatment can be hard to define. A more useful measure of waiting times for genetic services would be to measure the length of time before the patient accesses an expert. In illustration:

“Genetic disorders are different, the waiting time model is “too simplistic” for genetic services.” (HCP)

“In cancer genetic services the waiting times are completely unrealistic as the process of referral to treatment requires the collation of a large number of family member's medical histories with appropriate consent” from these relatives. The timescale for obtaining this consent “is outside of NHS staff control.” (HCP)

“Waiting times should be prioritised by need and severity *i.e.* rationalised.” (NHS Service Commissioner)

2) *The right to access specialist care*

Patients had a strong desire to be seen by a HPC who knew of their condition and had experience of managing it previously. The right to access specialist and sub-specialist services (expert professionals) irrespective of whether they are based nearby, in Scotland or in Europe was particularly important to people with rare genetic conditions and people without a diagnosis.

3) *The right to equity of access*

The principle of equity of access came up in several forms, but in general it required health services to be more flexible, such as provision of a choice of appointment times. A Tuberous Sclerosis patient commented:

“A choice of appointment times would be very helpful. Due to my work I often have to rearrange and this can [sometimes] be [...] an absolute nightmare depending on the department and speciality. I have to work like anyone else, what am I expected to do? [Individual healthcare professionals can have] a bearing on how you are treated. For example, I recently went to the back of a queue for trying to alter an appointment which wasn't even for the doctor I had been referred to.” (Patient)

Equity of access was also expressed in terms of the right to the same levels of support and service from the NHS irrespective of the rarity of the condition. It is the experience of many genetic patients that many of the HCPs they have been in contact with lack knowledge of their condition. For example:

“Good service in terms of screening, the provision of information and follow up tends to be determined by the commitment of the treating physician and varies from doctor to doctor. You can easily tell if a physician is interested in your condition or not.” (Patient)

4) *The right to the support necessary to enable patients to access care*

(i) *Transport*

Difficulty with transport to clinics was experienced in a variety of ways by most patients attending the consultation event.

The biggest concerns were cost and the unreliability:

- Patients felt that transport for treatment of their illness was a financial burden due to the remoteness of many Scottish communities.
- There is often no financial support for travel. These problems were exacerbated where patients had physical and / or mental (learning) disability. Many people affected by genetic conditions have mobility problems but do not fall under the criteria to achieve financial support through “disabled” status.

For example, a relative of a person with Marfan's Syndrome (who did not qualify for support) described how her brother had difficulty accessing his hospital and frequently could not attend medical appointments that were necessary for the treatment of his life-

threatening condition.

Similarly, a mother of a child with Fragile X said that she was paying for private transport for her son to attend school, as she could not get financial assistance because public transport was available although this transport could not be relied upon to be suitable. This decision failed to recognise that her child's access to public transport was barred by the sensory overload her child experienced when using public transport.

(ii) Disabled access

"If a clinic is not for orthopaedic patients it is assumed that access (stairs, doors) are not a problem. Access for disabled people is usually limited to the front door of the building, but once I get inside I find myself stuck between fire doors!"
(Patient)

One carer said that she experienced problems with disabled access to toilet facilities.

"My husband needs assistance when using the toilet, but in our local hospital the disabled toilets are within either the male or female toilets, so I have to wheel him in as far as I can and leave him to attempt to use the bathroom on his own."
(Patient)

(iii) Language

Non-native English speakers highlighted the importance of information in other languages at the point of access. This group of patients are often used to different health systems and are therefore not aware of their rights, the services available to them, or the knowledge of how to access services. Provision of language services also needs to be available throughout their care.

(iv) Learning Disability and Mental Incapacity

Access for people with mental incapacity or with learning difficulties was described as "doubly difficult". There was a feeling that these groups had to "jump through hoops" to access care and treatment. (The discussion on how better to support people with learning difficulties to interact with NHS Scotland is included under communication and participation.)

5) The right to access screening

The absence of organised screening for certain genetic conditions was felt to be highly inequitable (e.g. haemoglobinopathies). Closer links with community groups was raised as a rational cost-effective way to target ethnic minorities (who are at higher risk of certain genetic conditions) and to feed into screening programmes for small volume high-risk groups.

6) The right to treatment in a local setting

Whilst patients didn't mind travelling to see a specialist at key points in their

patient journey, they felt that often the more routine procedures and check ups could be done locally. Unfortunately, many patients reported difficulty in accessing GP services at busy times because their condition was “long-term” and did not fulfil the criteria for “emergencies”.

Omissions

The bill did not cover access to medicines.

What do patients and/or the NHS need to do to make this happen?

1) To provide equity of access, patients suggested that more funding and better use of the ambulance service was required. Many patients felt that a more flexible approach (access to a telephone conversation with a GP) would provide greater equity of access.

2) Equity of access could also be improved by creating a universal scale of ability and/or disability that covers all the barriers to access, as many people with severe genetic conditions do not qualify for the disabled badge.

3) Advertise existing services to all who need them.

'You can only access something that you are aware exists'. (Patient)

A specialist advisor from the Tuberous Sclerosis Association recalled how a specialist clinic was closed due to poor attendance, but when she met with patients she found that the vast majority had never been told about the clinic.

4) Information about services needs to be distributed in a number of languages and formats and by a number of methods to reach all patients. Representatives from community groups (such as MEHIP in Edinburgh and the Glasgow Black African Women's Association) were keen to support NHS Scotland to reach non-native English speakers. HCPs that attended the event also suggested that the NHS should use existing staff with language skills more effectively as they have both the medical and the language skills.

5) Both HCPs and patients were keen to use tele-health technology to overcome the issue of rurality in Scotland. There was the suggestion that tele-health could be delivered at the GP surgery so that technical support was available for the consultation.

6) The provision of a self-referral system as well as allocated appointments, would reduce fail-to-attends

2. The Right to Dignity & Respect

Key Points

- There needs to be greater consideration taken to the patient's individual experiences and circumstances.
- There needs to be more consideration for the dignity of people with learning difficulties, cognitive impairment and mental health difficulties.
- Children and young people deserve more consideration.
- Black and minority ethnic groups highlighted the need for equality and diversity training to support them. Ideally the training and subsequent policies would describe how NHS Scotland will achieve respect for diversity and involve consultation with relevant organisations

Suggested entitlements

1) *Respect for Diversity*

A person from West Africa highlighted the need for Equality and Diversity training:

“In my home country, it is normal to be able to ask a doctor to come to your home if you are ill. But I tried to ask for this in Britain and the doctor refused. I did not know that this was not normal here. And the doctor was cross with me. So there seems to be some acknowledgment in this Bill that people have different expectations and may not know how the health service works here; it is difficult for foreigners to understand it.” (Patient)

2) *Respect for people with learning difficulties and impaired cognitive abilities*

Failures in this area were highlighted for both adults and children with learning disabilities.

“Patients with multiple learning difficulties do not seem to be respected by health workers and their dignity is ignored. As a consequence parents whose children have multiple learning difficulties are afraid to leave their children alone in hospitals to be treated. This situation could be improved if staff were trained properly to understand the reality and complexity of some genetic conditions. Patients’ passports can and do help, and should be more widely used, but staff still need to be trained to use them properly and pay attention to them.” (Patient)

3) *Respect for patient knowledge, expertise and experience*

Many carers and patients said that they had had to become experts in their own condition because of widespread ignorance amongst health workers. Whilst no one expects medical staff to be experts in all conditions, there was a real sense of frustration and anger that the expertise garnered by day-to-day self-management, was not being taken on by health workers, for the benefit of other patients. The key barrier to sharing this knowledge amongst HCPs seemed to be that there is inadequate systems and resources dedicated to conveying it, and

that HCPs do not feel comfortable admitting their lack of experience or actually asking patients for information.

“In my experience nobody speaks to patients themselves, or to their parents, to find out from them what they know about their own conditions. Their expertise goes unused.” (Patient)

“Having more specialist nurses would help, but I've noticed that nurses are sometimes not listened to either, by more senior staff. There is a real hierarchy in the health service, with patients at the bottom.” (HCP)

4) The right to be seen as a person and an individual, and treated in a holistic manner

Patients reported that in their experience “professional health workers see a ‘condition,’ not the whole person.” This is especially dangerous for patients with conditions that can be variable or affect many organ systems. Therefore, a multidisciplinary approach to healthcare that sees the patient as an individual was welcomed. One patient summed up the general discussion by saying:

“There is a big need for the individual assessment of needs, to overcome the current assumption that any given condition always has the same symptoms.” (Patient)

Omissions

The right to express your frustration over inadequate service

It was felt that the bill did not acknowledge that patients often became angry after several incidents of being ignored, misrepresented or unsupported. Patients felt that they had the right to express their frustration in these circumstances, as respect is a two-way process and is based on listening and understanding each individual's point of view.

“Patients do get angry, with good reason, if they think they're not being listened to and they're suffering. The Bill needs to acknowledge this.” (HCP)

Q3. The Right to Safe and Effective Care

Key Points

- Patients **did not agree** that they should have responsibility for raising concerns over the safety of their treatment, until education existed that explained, in lay terms, what constitutes “safe” and “unsafe” care.
- *Patients believed that safe treatment required healthcare professionals (HCP) to be adequately qualified and trained HCPs.* Several patients had wanted to access information on their clinician’s training history and performance to date, but had been unable to. The vast majority of patients had never heard of the Scottish Patient Safety Alliance.
- Patients **did not entirely agree** with the responsibility of complying with the instructions regarding their medication, as they interpreted that this conflicts with the right to refuse treatment. This blanket responsibility doesn’t take account of occasions when adverse reactions occur to a new treatment in the absence of short-term availability of a HCP.
- The right to a timely diagnosis was missing from this section, and patients felt it constituted a form of unsafe care.
- Lack of access to effective treatment on the grounds of cost (due to rulings made by the Scottish Medicines Consortium) was often felt to contribute to ineffective care.
- The bill focuses too much on the hospital setting and infectious diseases rather than community based treatment, self-management and social care issues. Genetic health care is a growing area, and its unique requirements should be catered for.
- Both HCP and patients thought that safe and effective care was the responsibility of NHS Scotland as a service provider. There was some consternation over whether patients should carry any responsibility for this.

Suggested entitlements

1) *The right to feel safe and reassured*

“The differences in my experience of being treated for Tuberous Sclerosis have largely depended on the attitudes of the staff treating me. For example, I remember a nurse who had no consideration for how frightened I was whilst I was having a kidney scan. Staff need to be mindful that when they provide reassurance the whole experience can be less painful on many levels.” (Patient)

2) *The right to education about “safe and effective care”*

Patients need to have information about what current safety standards are in place if they are to be able to determine if their care is unsafe. One patient commented, “How do you reach someone who knows?” Also, patients should have the right to information about the full range of treatment options. One patient described his experience of asking questions as: “sometimes you feel like an inconvenience.”

3) The right to be treated by adequately qualified and trained HCPs

Patients and HCPs said that training was the cornerstone of safe and effective care. Genetic healthcare is rapidly developing and expanding across the specialisms, and many HCPs have had little genetic healthcare education. HCPs reported that inadequate training is a common barrier:

“The taking of accurate family histories is fundamental to appropriate screening of other family members.” (HCP)

4) The right to a care pathway to ensure safe and effective care

Safe and effective care is dependent on HCPs from different teams and specialities communicating effectively and maintaining continuity of care. Most of the consulted patients using Genetics Services raised this point:

“Safe and effective care means that all the care a person receives should be somehow threaded or linked together.”

“Safe care means a package of care *i.e.* a pathway.”

“It should represent a holistic approach through multidisciplinary teams 'joining forces'.”

5) The right to have your care tailored to your individual circumstances

For patients with alpha-1 anti-trypsin deficiency access to lightweight mobile oxygen delivery systems is restricted. The cheaper heavier systems are available, but this does not take account of the fact that people, who need oxygen as a consequence of a genetic condition, are likely to be relatively young, with family and work responsibilities and need to remain mobile.

Areas for improvement with regards to safe and effective care included:

1) Better training around differential diagnosis

Patients commented that for effective care there is an importance on reducing the time to diagnosis.

“I only got a diagnosis after I trawled through medical journals looking up my symptoms, and I had to see a physician in France to confirm my diagnosis. I felt that it was missed in Scotland because of the low incidence, but I thought that being from (West Africa) originally might have been a clue.” (Patient with sickle cell anaemia misdiagnosed as having anxiety disorder.)

In degenerative conditions an earlier diagnosis may enable lifestyle changes and treatments that may prevent or slow progression.

“I was misdiagnosed as having asthma rather than a degenerative lung condition of genetic origin and thus was not treated correctly.” (Patient with alpha-1 anti-trypsin deficiency)

Many patients also thought that access to psychological support around the time of diagnosis constituted good practice in terms of effective care.

2) *The provision of community based healthcare*

Safe and effective care in the community was inadequate in the views of most patients. One parent (of a disabled child with epilepsy and learning difficulties) had been legally awarded the right to manage his son’s care package after repeated failure of Health and Social care to provide adequately trained support workers. He felt that the NHS was “falling at the first hurdle because HCPs needed Epilepsy training.” He added, “you can have the *right* to anything but until the HCPs have the skills to deliver them, there is no real right.”

3) *Rare conditions*

Most genetic conditions are relatively or very rare. Patients affected by rare conditions reported more instances of unsafe or ineffective care (as well as an absence of care altogether) because of the lack of awareness of their condition with the exception of one or two experts. HCPs thought that this situation could be improved by training:

“There is a lack of training for specific genetic conditions and the service is therefore lacking.” (HCP)

What do patients and/or the NHS need to do to make this happen?

1) Patients require education to understand the safety and efficacy of their care. They felt that looking at safety would provide the basis with which to identify training needs within the NHS. Both of these goals would require significant funding.

2) Auditing is necessary to see where the areas of weakness are in a service (in terms of identifying training and information needs). The service should have a greater commitment to training through enabling HCPs to have adequate time and resources that are proportionate to the needs identified.

3) Paper records need to be replaced by electronic records to allow important information to be accessed wherever the patient is treated. Some patients said that they would be happy to carry a memory stick or disk containing their entire medical history with them at all times as a back up.

4) The two most universally held suggestions for improvement were that NHS Scotland:

- a) Make better use of parent and/or carer’s experiences.

- b) Use existing voluntary and charity organisation's expertise in delivering care.

Many families with relatives affected by rare and complex genetic diseases are willing to let HCPs and social work trainees shadow them to learn how to manage these rare and challenging conditions. It was felt that by ignoring the families expertise, the NHS was missing out on useful training and failing to provide adequately trained staff at the same time.

Q4. The Right to Communication

Key points

- Communication between patients and HCPs must be a “two-way street.” It was felt that communication skills could be improved in all sectors of the health service.
“We want communication that [treats patients as equals].” (Patient)
- Communication was identified as being particularly important in the following circumstances: at the time of diagnosis; for patients managed by multiple specialities; for patients requiring both health and social care services; and for all patients during major transitions (e.g. from paediatric to adult care; from inpatient to outpatient care and for end of life care).
- Extra resources are required to ensure that disadvantaged patients are supported in their communication. Patients and healthcare professionals (HCPs) understood “disadvantaged” to mean non-native English speakers, those with visual, hearing and speech impairments, children and young people, people with learning difficulties, refugees and asylum seekers and people from socio-economically deprived areas.
“Problems with English can lead to difficulties. Some health professionals are not aware that they shouldn’t use family and friends as interpreters or that they need to make sure that the interpreter has the right dialect or gender.” (Patient)
- Patients agreed with all of the provisions under the right to communication, especially the right to have your clinician introduce themselves and explain what they do.
- Patients disagreed that it is their responsibility to provide an accurate history of their condition. For many (medical) reasons patients might be unable to give a full and accurate account of their care. It also takes no account of the fact that genetics patients with complex disability would require an encyclopaedic memory to fulfil this responsibility. The onus should be on the HCP to establish the history and / or refer to an electronic patient record.

Suggested entitlements

1) Improved communication skills:

i) At the time of diagnosis

Patients were frustrated and angry when a diagnosis was delivered in an inappropriate way, and in an inappropriate place.

When genetic diagnoses are delivered by HCPs with inadequate knowledge of the condition, this communicates a sense that the standard of care they will receive could be vulnerable.

HCPs felt that more time was needed to allow for the news “to sink in” and for questions to be dealt with. Patients would like to be offered counselling (where appropriate) and given contact details for a HCP to ask for answers to questions that arise in the hours and days following diagnosis. Patients were extremely vocal about the need for information regarding local and national support groups to be given at the point of diagnosis, as this support and information resource is often invaluable.

ii) When a diagnosis is elusive

Patients often need more support when symptoms persist but diagnosis is not possible or easily achieved. Paradoxically, this is often the situation where the least support is offered.

“I know if I hadn’t been so persistent we still wouldn’t know the cause of my son’s condition”. (Patient)

(iii) For patients affected by learning disability, dementia (or lack of competence) or mental health problems.

“Patients sometimes have difficulties taking in information, particularly if they have learning difficulties. It is, therefore, important not just to communicate but to make sure that the information has been taken in and understood.” (HCP)

(iv) For children and young people.

“It is necessary to make sure that children receive the communication they need. Decisions are often made without talking to children. Incorrect assumptions can be made that they have a learning difficulty. Play should be valued as a form of communication. Children’s hospices are a good example of how this can be done e.g. Rachel House Children’s Hospice in Kinross and Robin House Children’s Hospice in Balloch.” (HCP)

2) The right to have your clinicians introduce themselves

“You are seldom told who will be treating you, and it helps. It makes things less clinical.” (Patient)

“It would be helpful if HCPs introduced themselves and explained how they fit into the service.” (Patient)

Omissions

What is currently missing from this right is the entitlement to care co-ordination

for families affected by complex multi-system genetic disease.

What do patients and/or the NHS need to do to make this happen?

1) Patients need more time to be allotted for communication within an appointment. Complex long-term genetic conditions require expert knowledge to be managed in a safe and effective manner. An example of good practice was provided by a specialist nurse working for the charity dEBra:

[“We hold regular specialist clinics to provide families with 'protected time' during which they can talk to a specialist about the specific problems that their child experiences.”](#)

2) The provision of services that broaden the number of ways medical information is communicated. E.g: The Scottish Consortium for Learning Disability offers training on how to make information accessible and how to communicate with people with learning disabilities. Likewise, the use of independent translators for people who do not speak English is far better practice than using a relative.

3) Clinicians need to acknowledge that some patients have a greater knowledge of their condition and that sometimes a carer or advocate will have a greater knowledge of the patient’s wishes.

[“Good communication involves a clinician explaining what they mean, listening to us and our concerns and offering choices.”](#) (Patient)

[“Clinicians can sometimes respond badly to patients who are clearly knowledgeable.”](#) (Patient)

Patients with learning difficulties and / or mental health difficulties, had often informed their carers (or advocate) that they wished to step out of dialogue with the clinician and be represented by them instead. Carers had experienced a resistance from clinicians to work in this way and were keen for this to change.

4) Several patients wanted to use care cards for communication with HCPs to reduce the amount of time, energy and sheer repetition it took to bring each new HCP up to speed.

Q5. The Right to Information

Key points

- A lack of accessible information in an appropriate format was a major barrier to patients feeling able to deal with, explain and manage their condition(s).
“When you are diagnosed with a rare condition, merely being told to “research it on the internet is completely inadequate.” (Patient)
- Patients rated the right to information as one of the most important in the Bill, but wanted clear guidelines on who could be expected to provide it and how.
“I’ve read it (the Bill) but it didn’t mean anything. It seemed more like guidelines than rules.” (Patient)
- Extra resources are required to provide information that is accessible to people who are vulnerable (to being excluded) such as those with learning disability or cognitive impairment.
- Informed consent can only be achieved by the provision of accessible, accurate information and both patients and healthcare professionals (HCPs) felt very strongly that there was a lack of both types of information within NHS services.

Suggested Entitlements

1) *Information at the point of diagnosis should include:*

i) *More time at the consultation for questions*

ii) *A named contact for patients to speak to if they have urgent questions*

iii) *Details of local / national voluntary organisations, advocacy services and other existing services that could help.*

“Professionals do a better job when they have the luxury of time. Then they can provide more individualised information. Unfortunately, due to time constraints getting the right information is more likely when people have the confidence to keep asking.”(HCP)

“The problem is, when you walk out of a consultation you have nowhere to go.” (Patient)

“Our son was tested at 3, but we didn’t find out what his diagnosis was until he was 23, where we came across it by accident when we asked to read his file. He had an extremely rare disorder and we had to go to the charity “Unique” to learn more about it. In our experience, after diagnosis, it was like the door was shut.” (Patient)

“When my son was diagnosed, we were given a list of associated conditions and

told “oh, your son will most likely lose his sight, but we don’t know much about this condition. You can look it up on the internet.” (Patient)

This highlights a key issue that should be addressed; how clinicians should present the situation of a lack of knowledge to their patient?

2) The provision of information needs to be flexible

The timing of information provision was cited as a frequent problem. Greater flexibility is required to provide the right amount of detail at the right time; crucially this will vary from individual to individual.

“People are not best placed to receive a mass of information when they had just been given bad news.” (HCP)

“In terms of providing information to patients about their diagnoses my experience is that one size does not fit all.” (HCP)

3) The provision of information needs to be ongoing

“I was given a folder when my child was one week old and that was it.” (Mother of a child with Haemophilia)

“After diagnosis you need to be able to go back to the information you were given. You also need to receive information when you want, ask questions when you want, rather than receive it all at once.” (Patient)

“I feel it is important to keep parents informed of treatment options whilst the child is still young.” (Patient)

Patients experienced problems around the dissemination of information as breakthroughs in knowledge are made. Conflict tended to arise as patients and/or carers kept up-to-date with the latest clinical research, while their clinicians did not wish to discuss it with them at appointments. Most patients thought that this illustrated a lack of respect for patient knowledge and/or experience and that better communication was needed.

4) For complex long-term conditions information must be backed up with education (preferably through access to self-management programmes).

“The lack of a simple checklist for managing a complex condition lead to unacceptable delays in the administration of pain relief for one of my patients.” (HCP)

“In genetics you are seen once, but there is a real danger of you slipping through the net after your first consultation. There is no call back mechanism or monitoring. This is inappropriate for long-term conditions. This differs from the services provided by the voluntary sector versus the NHS.” (HCP)

In general, patients wanted practical information first (at diagnosis) and more detailed medical knowledge later, after coming to terms with the diagnosis.

“Knowing the faulty gene isn’t the information I need. What helps is knowing about treatment and management for the condition.” (Patient)

“Information that has a point is useful. Not every patient wants to have all of the information. Medical professionals are right to filter information – give us what is useful.” (Patient)

*5) Considerable improvements with regards to the information provided for:
i) Rare conditions*

Most patients appreciated a clinician’s honesty if they admitted to not knowing a lot about their condition. Although some patients found this unnerving, most accepted that it was an unavoidable consequence of being medically “rare.”

However, these patients were very frustrated and disappointed that their clinicians did not take them seriously (as a valid source of information) or take interest in their expertise and knowledge to share with others.

“As parents we keep up with the latest information but are not taken seriously (by the medical profession) as experts in this (very rare) condition.” (Patient)

ii) Learning Disabilities

Patient information that is “easy to read” is required for people with learning difficulties. Several carers and voluntary sector workers pointed to the importance of checking for understanding in a consultation. Some patients affected by learning disability do not always understand or accept that they have a learning disability, and so would not think to ask for help.

What do patients and/or the NHS need to do to make this happen?

1) Employ more staff within the NHS to manage patient information.

“It doesn’t just appear and get replenished; there is a lack of joined up thinking in the NHS” (HCP)

2) Creating a support service database as a resource for clinicians.

“We need more logical and ordered web portals to link up and list all of the voluntary sector groups that do work for genetics patients. Here you could list all genetic conditions and under each one, all support services. I don’t think that we are utilising the existing technology and information that we already have to support patients. A database is needed that doctors can hook up with and deliver information to patients as soon as they need it.” (HCP)

3) Patients suggested the Genetic Interest Group or the Long-term Conditions Alliance Scotland could host a list of support services or be better utilised by

clinicians.

4) Patient information could be developed using current models of good practice. The Clinical Cancer Services were mentioned for excellence in the provision of information through partnership working with organisations such as Macmillan Cancer Care. Patients also praised the Muscular Dystrophy Campaign's introduction of care cards. Ideally, patients would like cards to be chipped so that they could carry their up-to-date medical records with them in case of future emergencies.

5) Patients particularly valued the peer support they received from meeting other families in the same or similar situations as themselves. Family days organised by Managed Clinical Networks were highlighted as a very effective way for patients to share information and support each other.

“Similar experience is often of the greatest value because it is based on lived experience”. (Patient)

Q6. The Right to Participation

Key points

- The right to participate was hugely valued by all patients, but for several different reasons. Some patients felt that access to specialist services was dependent on whether they had a good relationship with their clinician and whether, through working together, the clinician listened to, and valued their observations. Other patients valued participation because of the long-term chronic nature of their illness.
- Patients thought that this right should include the right to have a named person (such as a partner, carer or advocate) act on your behalf. This right needs to be supported by changing data protection laws to facilitate access to pertinent medical information by the named person
- Ethnic minority groups felt that the onus should be on the NHS and Scottish Government to provide better outreach services. They suggested that this could be achieved in part by better community engagement with pre-existing groups (e.g. Glasgow Women's Black African Group with regard to Sickle Cell Anaemia).
- Patients thought that sending an "ownership report" to every house in Scotland was a waste of resources. They thought that funding patient education and staff training would engender a greater cultural shift towards participation.

Suggested entitlements

1) Participation should be a right, not an obligation

While many patients welcomed the right to participate in their health care, some found it an unwanted burden. Flexibility is required of HCPs to support people who do not wish to be involved at every stage of their care and also to include and facilitate participation for those who want to be involved.

2) The right to ongoing participation

This was qualified as the right to be informed of new advances relevant to your condition.

3) The right to Informed Consent

Patients felt informed consent should be an ongoing process with sufficient provision of time and good quality information. It was noted that HCPs needed to ask more probing questions to ensure patients had actually understood what was being explained and the implications.

“First meetings need more time for patients to consider their options and for the HCP to check back that the facts, implications and consequences of any given course of action were grasped.” (HCP)

In the case of patients affected by learning disability, more effort was required to communicate in a manner that is understandable by the individual involved. It was felt that the amount of capacity a person has to express their opinion or preference was underestimated and under-explored due to time pressure.

Patients said they can feel under pressure to try to understand the decisions they are being asked to participate in, particularly if there are numerous decisions, or if the patient has learning difficulties or low self-esteem as a result of their condition. These latter patients in particular may not have the confidence to inform the HCP of their true views. This may translate into them appearing agreeable during the consent process, but not attending subsequent appointments. This already contributes to patients falling out of the healthcare system and there was concern that the bill might put more pressure on patients to appear to be participating in their healthcare decisions.

4) Consent is an ongoing process.

Genetic HCPs said that there is a need to make patients aware that their consent to information, tests and treatment could mean that they are contacted again, years in the future. Genetic patients said that if they were contacted years later, with further information that is likely to be life changing, they would prefer for the HCPs to get in touch personally to ensure that they still want the information.

5) The right to involve a named person such as a partner, carer or advocate

Patients would like the fact that they are entitled to bring a carer, family member or advocate to their appointments, to be made explicit and publicised at the start of their patient journey.

Where there is capacity, patients said they should be able to nominate another person to participate on their behalf (e.g. to support them to access medical information, apply for benefits, receive treatments or complain etc).

This is an area that is managed well for people with mental health difficulties under the Mental Health (Care and Treatment) (Scotland) Act due to the provision of Named Persons and Advance Statements. The same kind of legislation could be very usefully applied widely.

6) The commitment to participation should be evidenced by the provision of more time in clinic

Parents and carers of children with learning difficulties felt that their observations were often ignored during a professional assessment. Due to the brevity of the assessment they were concerned that an inaccurate picture was being ascertained and that not enough effort went into drawing out their child's abilities.

What do patients and/or the NHS need to do to make this happen?

- 1) Provide more time for consultations and provide a contact number and clear pathway for getting back in touch after initial consultation.
- 2) Patients wanted organisations and/or specific locations where patients and professionals can discuss issues together.
- 3) Patients wanted HCPs to strive to gain informed consent and recognise that it is an ongoing process.
- 4) Families living with a rare disorder wanted to be used as a training resource for HPCs, so that other families can benefit from their experiences.
- 5) Patients wanted the NHS to raise awareness of the right to participate from the very beginning of the patient journey, and develop processes that support patients to participate.

Q7. The Right to Privacy and Confidentiality

Key points

- The Bill should make reference to other acts that deal with confidentiality, and should not create duplication of legislation.
- The right to privacy should make specific reference to genetic healthcare, because “genetic information is family information”.
- Patients and healthcare professionals (HCPs) would like the Bill to offer illustration of how to interpret the balance between the rights to confidentiality for individuals, family members and other parties. It should provide circumstances where the rights of other parties or relatives would override an individual’s rights to privacy.
- Patients and genetic HCPs recognise the benefits individuals and families gain from sharing genetic information, when it is conducted with sensitivity and safety. This requires HCPs to understand the information and consenting mechanisms that enable this.
- There is a need for HCPs and patients to be aware of privacy issues when making decisions on behalf of those that do not have capacity, and sensitivity to identify when an individual may indeed have capacity.
- Both patients and HCPs thought that this right needed to be delivered in a way that is forward thinking and congruent with new technologies

Suggested entitlements

1) Minimum complexity of Legislation

There is a need to limit the complexity of legislation. There are several acts that already impact in privacy, including the Human Rights Act, and the Data Protection act. There was concern from HCPs that further legislation has the potential to cause further confusion, which could restrict the flow of information that currently constitutes good clinical practice.

2) Legal right to confidentiality about genetic information

Individuals' genetic information should not be made available to any third parties, medical or not, without the specific written consent of the patient, except in exceptional circumstances which could be outlined as case examples in the Bill. It should be made an offence for other parties to demand this information as a condition for supplying any service to an individual.

3) Appreciation that the shared nature of genetic information has an impact on families, not just individuals.

The right of an individual to privacy can conflict with safe and effective genetic healthcare. Genetic healthcare often requires information about a patient's relatives. For example, in the case of familial arrhythmias, communicating genetic health information gives relatives the opportunity to access risk reduction treatments and reduce premature deaths.

4) Appreciation that the rights of other relatives or parties may override an individual's rights to privacy.

Whilst the consultation participants supported the right to an individual's privacy, they believed that in genetic healthcare, there can be an overriding need to communicate family health information with at-risk relatives.

5) Advice and support that enable and supports relatives to communicate genetic information where appropriate.

HCPs explained that although individuals might appreciate that relatives could benefit from sharing genetic healthcare information between one another, in practice, it is often not shared. They explained that this happens:

- When relatives withhold information
- If relatives identify that they do not want to receive the information
- Unintentionally, when families have different perceptions of the family history, and who is at risk.

HCPs need to help patients to know which of their relatives could benefit from genetic information. They also need to be supportive because of the strain people can feel because of the implied responsibility to communicate this family genetic information.

6) Wider appreciation of the appropriate ethical handling of genetic healthcare information.

The 'complex balancing between the right to privacy and the right to information in genetic healthcare' is challenging to both patients and HCPs.

HCPs identified that genetic healthcare is becoming applicable to increasing numbers of patients and HCPs. For example, the newborn screening for Cystic Fibrosis means that midwives and new parents are dealing with the option of genetic testing, which can lead to ethical challenges caused by the implied responsibilities to inform relatives, and children of their test results.

7) Patients and professionals believed that in most circumstances, individuals should be given the opportunity to opt into sharing information.

This enables good clinical genetic healthcare through the benefits that information sharing:

- NHS regional genetic services already work in collaboration on genetic tests. HCPs said that the introduction of shared IT systems would help these services to coordinate the genetic tests, in order to use resources most effectively and to enable accurate interpretation of genetic

information.

- The education of HCPs can be improved by patients giving consent to be included in on-line or traditional education. Educational materials should acknowledge that consent was given.
- Patients spoke of the benefits of information sharing: The charity Unique holds a register of individuals with rare chromosomal conditions, with the consent of the individuals or their guardians. This has created a rich information resource about extremely rare conditions, enabling HCPs and families with these chromosomal conditions to obtain information, and assisting in the development of information leaflets. It is the individual's right to decide whether to contribute to the register, and many families do not see sharing the information as an invasion of their privacy.
- Some patients want to consent to permit their information to be used by genetic researchers or
- Some patients want to consent for the health service and charities to put them in contact with other individuals with their genetic condition.

They believe that they would need to consent as individuals, believing that an individual cannot consent for the whole family.

8) Effective mechanisms must support that appropriate information can flow effectively.

There is potential for data loss or abuse from those that have access to the information systems. However, the need for timely healthcare means that there must be efficient mechanisms that allow patients, relatives and HCPs to swiftly and securely identify if there is consent for information.

9) There should be an appreciation of the individuals right to privacy (and information when making decisions on behalf of those that do not have capacity.

Newborn cystic fibrosis (CF) screening detects infants with CF and atypical CF, but few HCPs or parents are aware of this. Atypical CF is often expected to be relatively mild, but there is little information about whether the current CF treatments will be appropriate. Several parents of children with atypical CF were concerned that their child has been labeled as having CF, and concerned that this could have an ongoing impact for their child: in their medical treatment, and in their ability to access employment and insurance, and for their psychological state. They were concerned that if the treatments are found to be ineffective for Atypical CF, their decision to participate in newborn CF screening and CF treatments on behalf of their child may not have been in the interests of their child, and their decision may be regretted by their child when it grows up.

What do patients and/or the NHS need to do to make this happen?

1) Both patients and HCPs would like case examples and educational materials about genetic privacy. These should clarify when the rights of relatives would override the rights to privacy of the individual.

For example, the Turners Syndrome Society has created an educational video to demonstrate to HCPs how best to cope with this balance in practice.

2) HCPs should provide appropriate information and support to individuals, in order to encourage that genetic healthcare information is communicated appropriately within families, whilst recognising that the communication can be emotionally sensitive for families.

3) The patients and genetic HCPs noted that patients carrying their own notes had apparently been successful in pregnancy care. They considered that patients carrying their own health record data on chips would enable them to share their information with relatives and their chosen healthcare providers.

4) Individuals should be given the opportunity to opt into sharing information in order to facilitate timely genetic healthcare, research, access to peer support and patient participation in HCP education.

5) There may be particular sensitivity about privacy at times of transition (e.g. from paediatrics to adult healthcare) and when there are capacity issues. HCPs need to have excellent understanding of privacy issues and communication skills when dealing with these patients and carers.

Q8. Independent support and redress

Key points

- Patients would welcome a new complaint procedure, providing that it is truly independent and that it has powers to discipline and change practice where appropriate.
- Patients thought that the description of this right was rather “woolly,” and that very clear and specific directives needed to be laid out to support the right in practice.
- Patients would like a system to be established where they could report their grievances but equally commend excellent service and good practice.
- Those that had experience of making a complaint pointed out that advocacy and support workers would be required to deliver this right in practice. Patients and their carers said that they were deterred from complaining due to the complexity, formality and time-consuming nature of the current process, as their primary concern was managing their condition.

Suggested entitlements

1) *Patients need to be confident that any complaint will not affect their future treatment.*

The main reason patients reported for not making a complaint is the fear of future discrimination. People affected by genetic conditions have a very limited choice (if any) of clinicians who can knowledgeably treat them. As a consequence, more often than not, complaints were not voiced for fear of the damage that this would do to the therapeutic relationship and the quality of future care.

“People feel anxious about complaining because they know that they will carry on seeing these doctors and nurses.” (Patient)

2) *Independent support and redress was defined by patients as satisfying the following criteria:*

- The process is truly independent
- The body responsible has powers to act where a complaint is upheld
- The individual (s) would be informed of the outcome and protected from discrimination in the future.

What do patients and/or the NHS need to do to make this happen?

1) Provide clear information from the point of first contact with a service about what to do if the service is unsatisfactory. One person referred to the Patient Information Centre at Edinburgh Royal Infirmary as an example of good practice in this area. Another person highlighted a service provided by the Scottish Health Council to assist in the grievance process.

2) Information about independent support and redress needs to be linked into the provision of information about safety, so that patients understand what constitutes “safe and effective care.”

3) Reduce the amount of time and effort required to feed your views back, and provide support services such as welfare rights workers and advocacy workers.
“It requires a huge amount of time and energy, something which you have very little of when you have genetic condition or are caring for someone who has. It can be very strenuous.” (Patient)

Q9. Other Rights?

1) The right to ongoing health education.

“One leaflet at diagnosis does not suffice.” (Patient)

All delegates at the event agreed that continuing education depends upon adequate resources for frontline health workers, patients and the general public.

2) The right to care co-ordination.

“I feel that for genetic conditions co-ordination of care is very important. I am (fingers crossed) well enough and well informed enough (by the Tuberos Sclerosis Association) to know what scans, tests and other treatments I need and when I need them, so that I can make sure that I have them. Luckily my GP and consultant physicians agree to my requests, but this might not happen if I wasn't a pharmacist.” (Patient & HCP)

3) The right to respite care.

4) The right to consistent practice across Scotland.

5) The right to have carers, advocacy workers, or a named representative (from a charity organisation) involved with your care.

6) The right to refuse or agree future contact over samples used in research (non-clinical).

7) The right to organised transitions, including from paediatric services to adult services.

8) The right to financial support.

9) The right to be treated for your symptoms, irrespective of whether your condition is fully recognised or understood.

10) The right to self-management courses for long-term conditions.

11) The right to social, emotional and psychological support (preferably integrated with the Clinical Genetics services).

Suggested forms that this could take included:

- A social worker
- A counsellor or psychologist
- Access to peer support

As one clinician concluded “the Bill is focussed on medical rights only and does not take account of social care, employment opportunities, the right to care for someone and be supported to do so, housing issues, the right to a relationship,

to education and to participation in society.”