

A response from the Genetic Interest Group

The Genetic Interest Group (GIG) is a UK-wide alliance of organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders. Its primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them. GIG is the only UK organisation of its kind.

The Genetic Interest Group welcomes the opportunity to comment on this consultation, and would be happy to discuss any of this response further.

General comments.

The overall effects on health and well-being for the population as a whole will without doubt benefit from many of the changes outlined in this consultation paper, however, if Commissioners seek only 'local' views then patients with rare genetic conditions will remain *invisible*. It is unlikely that community groups representing the needs of such patients and their families exist at local level¹ and only by engaging nationally with Patient Support Groups or alliances representing their interests will a full picture of their health and care needs be collated for the Joint Strategic Needs Assessment (JSNA).

The ideal expressed in the suggestion "to shift care closer to home" is not always appropriate for patients affected by or at risk of genetic conditions;

*"Thankfully I have now found an oral surgeon and dermatologist who have a broad knowledge of the condition (Gorlin Syndrome). I would gladly travel 100+ miles to see them. I only live 1 road away from my local hospital so I think this speaks volumes about a) the poor service and understanding at x General. b) the reassurance and understanding at Leeds. In my opinion, it is only down to luck or determination that you are referred to the "right people". I could easily be lost in the system with no follow up."*²

What patients want is echoed by your drive to deliver services that are better integrated around the needs of individuals and the "establishment of multi-disciplinary teams across health and social care". Patients would prefer to have a 'Centre of Excellence responsible for their care with one lead Clinician acting in a co-ordinating role'³. Perhaps it might be better for PCTs to look at funding networks of expertise (similar to Cancer Networks but based on systems e.g. neurological networks, endocrine networks, orthopaedic networks etc. that specialise in rare disorders) by contributing to a central national budget. This would ease the difficulties for patients who are looking for specialist treatments and

¹ Lane, A., Report on Progress to DOH: Development Officer, Patient & Public Participation, Genetic Interest Group & West Midlands Regional Clinical Genetics Unit, May 2004

² Genetic Interest Group Family Route Map Survey, 2007; unpublished data

³ Allford, A., Winter M., Family Route Map Project: Report of a series of six Focus Groups, Genetic Interest Group, March 2007 http://www.gig.org.uk/docs/FocusGroupReport_final_colour.pdf

surveillance and help prevent the deleterious effect on their health and well-being from delays in diagnosis and treatment;

“x (name of son) was nearly five before we found out he’d been diagnosed with it (Barth Syndrome)...he’d had a (heart) transplant at Great Ormond Street Hospital but there was clearly something else going on.”⁸

The Genetic Interest Group welcomes the ‘person-centred care and support’ aimed at helping patients and families and endorses the need to provide GPs with increased availability to physiotherapy and psychological therapy services, a further finding informed by patients, families and carers who participated in GIG’s recently held Focus Groups³.

‘Disinvesting’ from procedures or services where evidence suggests they are of limited or negligible effectiveness poses considerable difficulties for patients with rare genetic conditions as most have no protocols, no care pathways and no evidence base yet! Decisions to ‘disinvest’ must be taken nationally and with full consultation with patients who use, or seek to use such services or procedures. Making decisions at local level will further hinder the work to establish such treatments and procedures in helping patients with these rare disorders. ‘Managing exceptions’ whilst appropriate can be divisive if local people are given choices to make between services/procedures e.g. 10 people receiving heart bypass surgery vs. 2-3 sessions of infertility treatment for one couple, however a toolkit is available that offers advice around how to engage patients and the public in a meaningful way, when commissioning for long-term conditions⁴.

Commissioners are recommended “...to ensure that exception processes for interventions not normally funded are agreed and locally owned. This includes securing agreement with providers, clinicians, operational staff and local people.”(Annex B, B15). How can this be achieved in practice? Decisions around treatments and surveillance for patients with rare genetic conditions may not be well understood and these patients already consider themselves as expensive which increases the burden of illness on them;

“My local GP doesn’t want to know because of financial reasons”³

Collaborative commissioning arrangements should continue to enable patients with rare genetic conditions to access specialised services and many specialised service definitions currently exist, for example, **‘Specialised Services National Definition Set: 27 Specialised endocrinology services (adult)’⁵**, further specialised service definitions when published will hopefully provide essential frameworks for care.

⁴ <http://www.commissioningforthelongterm.org.uk> accessed 18/05/07

⁵ <http://www.shca.info/framepage.htm> accessed 18/05/07

Question 1

Individuals and families at risk of rare genetic conditions want access to experts who have experience of managing and treating these conditions. Such experts may not be locally available or even within a NHS Trust that has an existing contract with that patients' PCT. Often it is difficult for the patient to obtain information either about the condition or importantly, clinicians who have experience of managing and treating the condition.

"...absolutely no-one takes responsibility for managing you, pointing you in the right direction."⁸

It is therefore difficult for patients to take greater control over decisions about their own health when in some cases they are being discouraged and/or prevented from consulting relevant experts or receiving treatment outside of their local area.

The measures in this section appear to confound the efforts of such patients to take greater control as the focus is on needs assessment for the local community and not the individual. Already many PCTs have set their own guidelines where NICE guidelines have not been issued and this serves to cap the services provided to patients and exacerbate the postcode lottery which can only be eradicated by national services and contracts for people affected by rare genetic conditions.

"...we moved 10 miles down the road to a different Health Authority for health reasons... (we are) under 5 different hospitals...although it's a lot of travelling it works."⁸

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Question 2

Many people living with genetic disorders interact infrequently with their local health service. They often travel to attend clinics with specialist health professionals and bypass primary and secondary care services. There are however, areas where local services could help. Patients would like to be helped through the services they receive. They would like a co-ordinator of care, a person who can take responsibility for their continued surveillance and treatment. They would also like to be told of social benefits they may well be entitled to receive. Helping to facilitate patients care and journey to specialist services could be carried out by local services. Interacting with national voluntary organisations to

⁶ Allford, A., Winter M., Family Route Map Project: Report of a series of six Focus Groups, Genetic Interest Group, March 2007 http://www.gig.org.uk/docs/FocusGroupReport_final_colour.pdf

gain the views and experiences of patients could be a starting area for their views to be heard on local issues.

A co-ordinator of care would provide an ideal link between the local care providers and patients accessing care in other areas of the country.

Question 3

a) Commissioners undertaking an assessment of an individual's needs who has a complex multisystem rare genetic disorder require clinical knowledge far beyond that of many Primary Care and Secondary Care teams. This should be undertaken in liaison with a MDT Assessment (including an expert with knowledge of managing the particular condition) of an individual and not based on statistical analysis and algorithms of risk for more common disease such as exist for hypertension and coronary vascular disease. Furthermore, what does this mean in terms of funding allocation, if an individual has been 'assessed' will there be an amount allocated for that individual (that is either ring-fenced or capped)? And will this take place through the local services or by a national funding budget?

b) & c) The Strategic Health Authority have a role in assessing the needs of the local community together with expertise in this field, including information and planning tools, Sharing of information and resources are required to complete the JSNAs but are there sufficient numbers of Commissioners to do all of this work?

GIG welcomes the proposed guidance to be published later this year on person centred and integrated planning and acknowledges that to ensure that all patients with genetic (and therefore long term) conditions have a personal care plan by 2010 is an ambitious target. However, in work that GIG has carried out having a co-ordinated care plan is what many patients feel they are currently lacking, and subject to the detail contained within, this proposal has the potential to help provide some guidance on what care, treatment and surveillance patients should be receiving.

Question 4

Are there systems currently in place to pool information and share resources, or is it an impossible exercise in IT at the present time? Listening to patients might be an appropriate place to start, the majority of patients, their families and carers told GIG in Focus Groups and interviews for the Family Route Map Project that healthcare professionals do not communicate well with them and do not listen to what they say. Inter-professional communication was cited as almost non-existent. This does not confidently provide a firm platform for sharing information when professionals involved in care do not communicate effectively. Improving this inter-professional communication will be key to creating Joint Strategic Needs Assessments, it will also be important to highlight that not all conditions will have the necessary local presence for them to be considered at this level. Many rare conditions will fall outside the standard template for JSNA's and this needs to be taken into consideration to ensure that patients are not left behind.

Question 5

Further information is first required to establish if Children's Trusts are satisfactorily accommodating commissioning for children and young people through their services. Parents of children with complex conditions resulting from genetic disorders are frustrated with the system of care for their children.

*"As a family with a disabled child you're just bottom of the pile."*²

However, data could be used to model future needs generically but individual assessment would be extremely difficult for Commissioners to undertake without appropriate skills, training, and development.

Question 8

Identification of children at risk of a genetic condition may mean that other people know the status of a child before the child or in some cases their parents, know themselves. This could lead to choices being presented to them before necessary, for example, before a child reaches reproductive age, and also before information has been provided to allow them to make informed choice leading to possible psychological harm or damaging family relationships.

Question 9

Commissioners need to be working with the DOH, *patients and the public* on this.

Question 11

There should be one for each of the main service segments and arguably a further segment added for 'rare conditions'.

Question 12

Commissioners need to be aware of the possible impact on services such as physiotherapy and psychology services even with 'step approach' screening in place. Many patients and families involved in the GIG Family Route Map project already want access to these services and are finding it difficult to get them, demand is only set to increase and the current scarcity is noticeable.

Question 14

Individuals with rare conditions may need surveillance and interventions outside of the scope of local contractors and require flexible funding to be able to do so. Special arrangements should be put in place for this to happen at a national level for all rare conditions in order to eliminate the current postcode lottery³.

Question 17

Special National Groups including patient and professional bodies are required to guarantee inclusion of patients with rare genetic disorders. These groups should also have a 'voice' and be consulted by Commissioners to ensure accountability.

Question 18

Local authorities should be involved in planning for their populations' needs but may not have sufficient expertise in health to do so currently.

Question 20

A specific training module on genetics and the affects on individuals and families at risk of genetic conditions, including the social/psychological aspects would provide an opportunity to begin to understand the complexity of such conditions.

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22/05/07