A patient centred approach to care planning for patients with chronic genetic diseases

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Abstract

This essay proposes seven pre-requisites for the creation of effective programmes of care and support for patients living with the consequences of chronic genetic diseases. It then goes on to discuss the role of patient organisations and other stakeholders in bringing about the development and implementation of these.

Introduction

Throughout the world there are millions of patients affected by chronic, life limiting conditions. For these patients there is no *magic bullet*, no single, simple intervention that will make the condition go away. Rather they need to find ways of managing their condition in ways that will fit their situation and their aspirations, making best use of the interventions and support available to them so that, as far as possible, they can fit their disease to their life, rather than having to make their life fit round the disease(s) which affects them. For many, this might mean searching out the *least worst* rather than the *best possible* combination among the options available due to the paucity of expertise available, or the limitations on our current abilities to alter the course of a given condition and improve the quality and/or the quantity of life for those affected.

For patients and families living with chronic disease to be able to gain and retain the degree of control over their lives that they wish to given their situation and the limits imposed by their condition a partnership approach, with all stakeholders contributing their specific expertise and knowledge is essential. In this the patient and the family are central, with a seat at the table where decisions will be made

and a voice that is listened to by the other stakeholders as a right, rather than as a result of the condescension of others. This partnership will need a flexible mandate that allows the contributors to adopt a holistic approach to problem solving, thinking outside the box when the occasion demands in order to bring together the elements of care, medicine, technology, information, policy and advocacy in ways that are fit for purpose, and which make good use of available expertise and resources in the pursuit of patient and family led health gain.

This applies whether the patient's condition is rare or common, but the centrality of the patient in the case of rare diseases is paramount given that they are the source of many relevant insights into their condition, what works and what doesn't in its management and so in guiding the use of scarce resources to best advantage.

It is also important to recognise that, whilst many common chronic diseases that are currently the focus of much attention because of the demands that those affected make on the healthcare systems to which they look for care are potentially preventable, chronic ill health arising from rare and genetic causes is generally unavoidable in the absence of effective therapies to treat or cure. Unlike Type 2 diabetes, or COPD, no amount of healthy eating will help to prevent genetic disease, even if it might help ameliorate some of the symptoms. This being so, we need to plan for the provision of effective, user-friendly services that will meet the needs of patients and their carers in a timely, sustainable and user-friendly manner.

Pre-requisites for a model for the management of chronic rare conditions

Much of the attention currently focussed on chronic disease care is directed along public health lines towards interventions that will reduce the likelihood of chronic disease occurring in middle or old age through low cost lifestyle or environmental interventions that will eliminate, reduce or delay the onset of symptoms. This priority arises from anxieties about the ability of health care systems in the developed world to come with what many see as a tsunami of demand that is foreseen to be about to strike. In no way would I want to imply that this is not an essential focus for public health, but it is essential to recognise that, even if these programmes are successful beyond the wildest dreams of their initiators, there will still be a very substantial number of people who will have life limiting chronic diseases over which they have no choice as to whether or not they will experience the consequences of. The seeds of the disease are inherent and cross the generations.

So the first pre-requisite is that all concerned recognise that there is no one size fits all model that can be dropped onto the entire population and used to push square pegs (rare disease patients) into round holes (public health based disease risk reduction plans).

While it is sometimes the case that families affected with rare diseases will be found clustered in the vicinity of hospitals able to provide

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high levels of disease specific specialist care this is the exception rather than the rule. Far more usually affected families are scattered throughout the population, looking to local hospitals and community care for assistance in the management of their situation. This means that planning and provision of care needs to be strategic as well as pragmatic. A helicopter view enables everyone to see the big picture and look to concentrating those elements of a care programme in centres where the critical mass necessary to maintain expertise and secure sustainability can be generated. This avoids unnecessary duplication and reduces the risk of local enthusiast going off at a tangent and providing less than optimal support.

The second pre-requisite, therefore, is that an overview of needs is taken from a sufficiently elevated position to be able to see the situation in the round and allocate resources accordingly. This also increases the chance of the issue landing on the desks of people of sufficient seniority to allow decisions to be made. One off decisions are frequently taken by relatively junior people who, by virtue of their standpoint, are unable to see the bigger picture and as a consequence run the risk of establishing precedents that will make the subsequent development of higher level strategies more, rather than less difficult.

While specialist centres have a key role to play in the provision of care and support for patients with chronic genetic conditions, most patients with the condition in question will not live in the shadow of such specialist facilities. Even for those fortunate enough to do so, many aspects of care and support which they may need will not be provided in a hospital setting. Instead they will be within the bailiwick of local and community services. They may not even be the responsibility of the health care system at all, but fall to other agencies in social care, education or elsewhere depending on jurisdictional boundaries in the country in question. Inadequate coordination of centrally and locally delivered element of care and support can create confusion, with patients and families getting mixed messages and partial services, falling through the net because of a breakdown in communication, or even receiving inappropriate interventions which hinder rather than help. For example, boys with Duchenne muscular dystrophy benefit from physiotherapy. Travelling to an expert centre to receive this regularly is not a viable option for most families. Community physiotherapists are frequently unfamiliar with the special needs of Duchenne boys although they are perfectly capable of providing a service if they know what to do. Inappropriate physiotherapy can worsen the situation for the boy. In the absence of a link to relevant expertise locally provided services may struggle to do the right thing and in the worst case may even do harm.

This means that the third pre-requisite for the provision of care and support for patients and families is that there must be proper integration between those elements which are provided centrally and those which are provided locally. Achieving this not only requires vertical integration between the centre and the community, it also requires horizontal integration across institutional boundaries with different agencies working together to a coherent and coordinated agenda. Over rigid insistence on only acting within the mandate of the Department or Institution leads to inefficiencies, inappropriate territorial behaviour by professionals and officials, too often creates a silo mentality which is inhibitory to progress because expenditure *here* will produce results *over there* which are outside the departmental orbit. The fallout from this territoriality impacts on patients and families caught in the middle of often arcane disputes which seem to defy logic and common sense.

Recent progress in biomedical research has seen the creation of unprecedented insights into the fundamental flaws in our basic biology that result in the chronic conditions that impair the quality and quantity of life for so many currently coping with the daily realities of life with a serious genetic disease. While it remains the case that the majority of patients with rare chronic diseases are still waiting for

innovative drugs that will alter the progression of their disease, for a growing number of conditions innovative interventions are being developed that hold out the promise of better times ahead. However, the path from bright idea to available intervention is a long one, and there are many obstacles in the way that threaten to derail the progress of a novel possibility before it gets to the point where patients can expect to be able to benefit. Biomedical research is moving from a model in which small molecules were tested on large populations to a situation where the reverse applies, and large molecules (and these are usually biological in origin and may even include genes and cells) are tested on smaller and smaller populations. History teaches us that we need a regulatory framework to reduce the risk of patients being exposed to undue risk, but a system based on a historical perspective runs the risk of failing to see forward facing opportunities derived from advances that have outstripped the regulators parameters. We need to change the rule book and allow the framework for bringing new possibilities to patients to reflect what we know and where we are heading, rather than judging today's and tomorrow's progress by yesterday's rules.

So the fourth pre-requisite is that the regulatory framework set up to check that new interventions (be they new drugs, devices or some other development) is based on a legislative framework that reflects what is actually going on in our bodies. It is much easier to create a legal framework that is based on our biology than it is to try and make our biology obey the law! Patients, ultimately, might be bound by the rulings of the European Court of Justice, but our metabolism and our genes are not.

When a new possibility is licensed this is not the end of the story. In an increasing number of health care systems there are further hazards to be negotiated before new products can routinely be used for patient benefit. Health care systems everywhere are caught between the *rock* of increasing possibility, and the *hard place* of constrained resources. More and more, health care systems are looking to Health Technology Assessment (HTA) to help decide between conflicting options when it comes to allocating resources that will not stretch far enough to permit everything that is a possibility to become a reality. The rationale for HTA is unarguable. Of course those charged with the responsibility of allocating the health care budget wisely need to have a robust framework to help them in this task. And HTA has the merit of seeming to be a scientific system that is independent of personal prejudice. Feed it the ingredients, turn the handle and the decision pops out at the other end fully formed and readily defensible. But most HTA models assume a very simple idea of cost effectiveness and clinical effectiveness. Things that matter to the health service are counted, but things that matter to patients or families may carry much less weight or even be discounted from the calculation entirely. Suppose, for example, that the provision of a novel therapy for a patient enabled their carer (who is usually a family member) to return to work because the patients care needs were reduced to the point where they no longer needed to have the constant presence of carers but could rely on alarms and on call response teams. This might well be seen as a benefit for the wellbeing of the carer, but it would be unlikely to weigh in the balance of the HTA assessors as the costs to the system would remain pretty much the same - or they might even increase due to the need to provide on call response services where previously the domiciliary care carried the load. Similarly, a small increase in a measurable outcome such as a patient's Forced Expiration Volume (FEV, a measure of lung function) may seem insignificant until you realise that it means that that individual is no longer dependent on mechanical respiration, which in turn means it is easier to hold a conversation and others can see their face, facilitating better social interactions and a much improved quality of life.

All of which means that the fifth pre-requisite is that HTA systems must be fit for purpose. They must bring into their calculations more than the things that can be counted and the things that impact on the health care system. They need to take a broader societal framework if

they are to carry the confidence of those on whose behalf they are mandated to make judgements. Otherwise there is a danger that they run the risk of knowing the price of everything, but the value of nothing, and politicians are enabled to hide behind an apparently robust but actually partial bulwark when justifying their unwillingness to provide desired interventions for patients and families with chronic diseases.

In parallel with the questions addressed by the HTA process, novel interventions have to go through national systems for establishing pricing and reimbursement. This can be complex and the system varies from country to country across the EU, with some countries taking much longer than others to agree the price that they are prepared to pay. It would not be unfair to say that delays at this point in the process from bright idea to available intervention often seem, to patients and families at least, to be exercises in rationing by payers, or politicking by manufacturers juggling access for patients with the need to set the best possible price for their product across a basket of jurisdictions. Space does not permit the different systems in use across the EU to be detailed here, and systems are often in a state of flux as they try to secure a workable deal. In the UK there is a significant change under way with a shift to a framework for new drugs known as *Value Based Pricing*. The idea underpinning this is simple - those drugs which deliver greater value should potentially be able to command a higher price - but there is currently (September 2012) a distinct lack of clarity as to how this will actually work in practice. Nor is it clear who will have a say in establishing value, and what the price will be for patients who might have benefitted in the case where a novel therapy is unavailable in the event of a failure to agree.

This means that the sixth pre-requisite is that the system by which prices and access are determined must be transparent, robust, rational comprehensible, and work according to agreed timetables in order to avoid tactical game playing by vested interests at the expense of patients waiting to benefit from useful interventions that get stuck in the limbo of national negotiations about their price and availability.

Suppose a novel intervention that is intended to offer the prospect of an improvement in their condition to patients with a serious chronic disease has made the journey from the research laboratory through clinical development, licensing, HTA and pricing and reimbursement. Logically one might expect that eligible patients with an unmet health need would then be able to access it with minimal further delay. In some cases this is the case, but it is by no means universally so. In order to take advantage of novel possibilities professionals must be updated on an ongoing basis so they can incorporate novel possibilities in their clinical practice, and the healthcare system must be sufficiently flexible in its organisation to accommodate new ways of working. Health care planners and policy makers can sometimes seem to have a lot in common with generals - brilliant at fighting the last war, but not so good at preparing for the next. Our health care systems have a historic legacy that frames their thinking which needs to be discarded if the possibilities on the horizon created by the latest research and development are to be realised. All too often changing the way in which health and social care is organised is like trying to turn a supertanker. Moving the steering wheel now produces a slight alteration in course many miles down the road as institutional inertia resists the pressure to move from the previous line until the pressure for change becomes absolutely irresistible.

Hence the seventh pre-requisite for improving services and support for patients with chronic diseases is that there must be ongoing programmes of continuing professional development (CPD) for all medical professionals designed to keep them up to date with new ways of working and able to incorporate novel possibilities into everyday professional practice. This is important, it is necessary, but it is not sufficient. It must be accompanied by institutional investment in novel structures and new ways of working. It is of little use, for example, to develop remote monitoring arrangements that allow patients to remain in their

own homes and reduce the necessity for regular clinic attendance if the relevant community care professional are not equipped to receive and act upon the information thus transmitted.

The role of patients and families in the development of high quality care and support

Reading the above list of pre-requisites one might assume that the patient and his or her family are passive recipients of the wisdom and efforts of others, working dedicatedly to deliver high quality care and support to those with unavoidable chronic ill health. This is far from the case. Bringing about a framework within which patients and families can exercise the degree of control over their lives and opportunities that fits their circumstances and which reflects the very real limitations that many chronic diseases impose on those affected requires an active partnership between all stakeholders. Patients and families have a unique expertise into the way in which their disease limits their opportunities and experiences that goes far beyond that of medical, social care, educational and other professionals. These others have their unique expertise too. It is in coming together as equals in pursuit of a shared goal that permits the potential synergy there to be created to be realised.

This is unlikely to happen of its own accord. It needs a commitment from all concerned to bring it about. If we are to see the mantra *Nothing about us without us*, currently parroted by politicians and policy makers translated into a demonstrable commitment to engage with patients and families in a real and meaningful way then both sides will need to change. Patient organisations will need to invest time and energy in understanding how their health and social care system works - in theory and in practice. They will have to develop schemes for training and supporting their advocates to work strategically at a system level and pragmatically in support of individuals and families making their case in ways service providers and clinicians can understand. The recognition of a shared need and the provision of peer to peer support has been the inspiration behind the creation of many patient organisations. This will remain central to their purpose, but to become powerful as agents for change on behalf of their members patient organisations will need to raise their game and generate the evidence from their members to support the changes they wish to see brought about. As patients and as patient advocates we know only too well the suffering and hardship that chronic genetic diseases bring about for those affected. Sadly this is not enough to bring about the changes we seek. We must come with proposals that are worked through and evidence based if we are to earn the partnership status we deserve. This may seem like a daunting task, but there are resources available, often under the auspices of European umbrella bodies such as the European Patients Forum (EPF), The European Rare Diseases Organisation (EURORDIS) and the Thalassaemia International Federation (TIF) to help build the organisational capacity of patient organisations to engage at all levels. National umbrella bodies such as Genetic Alliance UK and the Dutch Genetic Alliance (VSOP) and multi stakeholder bodies such as the European Platform for Patient Organisations, Science and Industry (Epossi) also have much to offer.

However it is not just a matter for patient organisations to take the necessary action to bring about the development of high quality services and support for those with chronic diseases. The system must also move. If the rhetoric about patient engagement is to become reality then there must be a demonstrable willingness to invest resources to support the development of the strategic capacity of patient organisations to play the strategic role outlined above. The difficulties and dilemmas for patient representatives asked to participate in planning and developing services need to be accounted for. Simply asking for a patient representative to join a committee is inadequate if there is no

effort made to facilitate their being able to play an active role. This means, for example, training for the patient advocate. It also means training for the other members so the patient advocate is not left outside the gilded circle of professionals sitting round the table. It also means recognising that the professionals are at the table because it is their job to be there. The patient advocate may be there because he or she has taken time from their job, or have had to make arrangements for respite care for their affected family member so they are effectively paying for the opportunity to participate rather than being paid.

Services provided should aim to be delivered in a timely, appropriate and user friendly manner. They then need to be monitored to establish the extent to which they are meeting the needs of those receiving them. This needs to be carried out against criteria agreed to be appropriate by those receiving them, not just those thought to be appropriate by those responsible for planning and delivering them. Patient organisations are essential contributors to getting this framework right, but they need to be in at the start, not asked to rubber stamp decisions when they are done and dusted.

All stakeholders involved with the development and provision (and the receipt) of services and support designed to alleviate the impact of unavoidable chronic disease want to get it right. Nobody wants to see wasted resources, especially when there is severe downward pressure on many health care budget and a growing opportunity to use new knowledge to do more and exert a positive impact on the lives of those affected. I have identified seven pre-requisites for the planning and delivery of effective support for patients and families living with the daily consequences of unmet medical needs resulting from chronic genetic disease. Effective engagement with patient and with patient organisations will enable these to be met in a timely and a realistic manner. This will not only maximise the health gain to be had for the service provider, it will also reduce the waste of scarce resources through the provision of inappropriate or inadequate services. Most of all, it will enhance the quality, and in many cases the quantity, of life for those who, through no fault of their own are experiencing the daily reality of life with an incurable disease.

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