9 Voluntary organisations

Introduction

A focus group for representatives of voluntary organisations for people with IMDs was held on 23 June 2005. Those present included the following:

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<th>Name</th>
<th>Position and Organisation</th>
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<tr>
<td>Hilary Burton</td>
<td>Consultant in Public Health Medicine, PHGU</td>
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<td>Tanya Collin-Histed</td>
<td>Executive Committee Member, UK Gaucher Association</td>
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<td>Brendan Gogarty</td>
<td>Public Involvement Officer, PHGU</td>
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<td>Anne Hale</td>
<td>Executive Director, Global Organisation for Lysosomal Diseases</td>
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<td>Steve Hannigan</td>
<td>Executive Director, CLIMB</td>
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<td>Christine Lavery</td>
<td>Chief Executive, Society for Mucopolysaccharide Disease</td>
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<td>Ann Phillips</td>
<td>President/Co-founder, Association of Glycogen Storage Disease.</td>
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Following presentation of the background to the work and some of the main findings of the services review, the group was asked to discuss their experiences of the services and their views on unmet needs for service users, focusing on diagnosis, initial treatment and long-term care. They were then asked to make recommendations on ways in which the services could be developed and improved.

Main unmet needs and recommendations

2.1 Diagnosis and misdiagnosis

There are regularly problems of misdiagnosis and lengthy time-lags before a diagnosis is made. This is often due to difficulties that non-specialist GPs and other practitioners have in recognising rare disorders that they are likely never to have seen before. Once someone suspects an IMD, it often takes a long time to get the relevant testing done. Testing may be slow. This may be a problem of out-of-date equipment or having to wait for a batch of samples before running the rest; sometimes it is because the laboratory service has only one tandem mass spectrometer and there is no back-up if it breaks down.

The next problem that patients and families encounter is communication during the time of the diagnosis. There are often problems of parents being given erroneous or imprecise diagnosis. This can lead to much distress as they look conditions up on the internet and see depressing and shocking information. This might turn out to be misleading or untrue when the final detailed diagnosis is made. There are delays in getting supporting information about the condition to the patient and parents. The voluntary groups thought that parents should only be told in general terms about the tests that are being done or what is suspected. Only when a precise diagnosis is known, and the appropriate information about the condition has been sought by the health professional, should the exact diagnosis be given to the parents. This should be immediately supported by information about the condition and the contact details of the relevant voluntary organisation. It was thought that this could be expedited by laboratories including information with the diagnostic result when a positive result is returned.

In summary, as with many other rare and serious diseases, voluntary groups thought that there should be standards around the process of communication of a diagnosis.
2.2  Initial treatment and care

Quote from McArdle perspective

This is often very poor indeed or non-existent. Quite often more harm is done than good. Many consultants who get involved in diagnosis are excited about finding a rare disorder but have no experience of it. They may only ever see one or two cases in their careers. There is a tendency to tell the patient that nothing can be done.

Representatives thought that quite often there was a reluctance to refer to the relevant expert owing to:
- lack of knowledge about the condition
- rarity of conditions
- lack of knowledge about the best expert services
- concern over expense
- desire to hang on to interesting cases.

They thought that there should be nationally defined care pathways for each condition (some examples are provided by MPS) which start at the time of diagnosis and cover specialist referral and management. In general, the time from diagnosis to getting to see a specialist is too long. Information about how to access these care pathways should be sent out to the health professional with any positive diagnostic test result.

Specialist services need to be available and accessible from the patient’s home. But this is frequently not the case. The representative from CLIMB, the umbrella group for IMDs, thought that fewer than half of patients are looked after by specialist services and many have to travel long distances to access specialist services. Services should be commissioned on a geographical basis to cover the whole of the country.

The problem of lack of availability of services was made worse because services were often dependent on the availability of one consultant, making them very vulnerable to disruption when the individual is away, or, longer term, at retirement. Services need to be robust and comprehensive, including a whole multi-disciplinary team and sufficient clinical staff to provide cover on a 24-hour basis.

2.3  Standards of provision at out-patient services

Parents were concerned that there is no care or supervision for the child or for brothers and sisters when the child’s case is being discussed. This means that either diseases and prognosis may be discussed in front of the child inappropriately, or both parents are not able to be present. It also means that brothers and sisters have to be left at home — a situation that parents do not like, firstly because finding care for them may be difficult and, more importantly, because it cuts them off from the attention focused on the sick child and provides no support for them in understanding what is happening. It was recommended that care should be provided in out-patient clinics for children and their siblings, to enable consultation with parents.

2.4  Ongoing care and support

‘The patient is often left with little or no care and support. We need all patients to be referred to a specialist centre where there is in-depth knowledge of management or failing that good communication channels from specialist centres to local facilities to support patients near home’

Patients and families have long-term problems accessing adequate support. They experience:
- poor understanding of disease by local health services, social services and education
- financial problems and problems getting allowances (voluntary organisations spend a
lot of time helping people with forms and being advocates for them)
● a major gap in respite care and education
● support services that are not comprehensive or integrated, do not reach the whole population, and are inconsistent.

They recommend that there should be good networks of care, including specialist centres and networks which include more local services, with professionals educated and supported by the specialist centre. In general, there needs to be more support for people with long-term disabilities on a national basis.

In IMD services voluntary groups noted a deficiency in psychology services, few transitional services for adolescence and a large gap in services for adults. All of these need to be developed for all regions.

Pre-implantation diagnosis is rarely available for couples with IMDs. It was thought that the reasons behind this should be explored further.

2.5 Organisational gaps

Disease register

There is no NHS disease register. However, much reliance is placed on the MPS register which is maintained by the voluntary organisation to cover London and Manchester patients and is thought to be 96 per cent correct. NHS services should invest in this, building on registers currently available through voluntary organisations.

Education of health professionals

Other health professionals and commissioners need to have more education about IMDs. Voluntary organisations could be involved in the provision and should be considered an integral part. However, they are not often asked.

3 Main recommendations

Many of the problems are characteristic problems of management of rare disorders.

1. Management of rare disorders should become a central concern of government and NHS, with ring-fenced money and a body (possibly an ombudsman) to raise awareness and champion issues.
2. Disease registers should be established and developed with proper funding to ensure quality, sustainability and accessibility.
3. There needs to be a programme of education for health professionals and commissioners. Voluntary organisations should be considered as providers. This would be supported by the voluntary sector, but would need some funding.
4. Comprehensive and specialist services should be accessible to all patients. This will require:
   ● development of robust services with specialist centres and networks to cover the UK geographically
   ● development of a comprehensive network of transitional and adult services in association with main paediatric and laboratory centres
   ● raising awareness of the presence of these services and prompting health professionals to refer patients on diagnosis
   ● ensuring services are commissioned
   ● specialist education and CPD programmes for all members of the multi-disciplinary team.
5. Voluntary organisations could help to increase pressure for development of services by supporting their members in seeking specialist management.

6. Professionals should work together with voluntary organisations to develop care pathways for all conditions, which would be centrally available and could be accessed and sent out by the laboratory when the diagnosis is made.

7. The issue of accessibility to pre-implantation genetic diagnosis should be investigated further.

8. Health services should work with voluntary organisations, social services and education to ensure comprehensive support services for those with long-term disabilities and health problems.