CONSULTATION RESPONSE FORM

UK Plan on Rare Disease

Consultation Questions

Overall statement: We at Unique have worked very closely with Rare Disease UK over the last 3.5 years, including participating in the working group on the Patient Care, Information and Support section in RDUK’s report ‘Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy’ and contributing to their report ‘Experiences of Rare Diseases: An Insight from Patients and Families’. Furthermore, we participated in RDUK’s stakeholder event in London on 3rd May 2012 about the consultation to the UK plan for rare diseases. We endorse fully RDUK’s response to the Department of Health (England) consultation on the UK plan for rare diseases. In our response to the consultation questions in this document, we build on RDUK’s response with points especially relevant to our thousands of member families affected by rare chromosome disorders in the UK.

CONSULTATION QUESTION: Do you agree that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, particularly in neurology and genetics?

Comments

Unique agrees that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, not only in neurology and genetics but across the spectrum of conditions. In our experience, there is sometimes a tendency in primary and secondary care to look at individual symptoms in isolation rather than as a complex matrix of symptoms emanating from an underlying chromosome disorder. Any tool to encourage clinicians to consider testing for a rare chromosome disorder in a timely fashion and then to refer on to appropriate colleagues is to be welcomed. We would suggest that awareness of the possibility of a diagnosis of a rare disorder should be highlighted in all aspects of health professionals’ training, both pre- and post-graduation. Development of e-learning packages to support this initiative would be very helpful, being both cost- and time-efficient for the health professional and for the NHS.

CONSULTATION QUESTION: Can you suggest ways of rare disease featuring more prominently in speciality training for doctors?

Comments
It is very important that the collective commonality of individually rare diseases is recognised by medical and other NHS staff. Although individually rare, rare chromosome disorders are collectively numerous, affecting at least 1 in every 200 live born babies. With the gradual (albeit patchy) roll out across the NHS of array CGH analysis for the detection of chromosome abnormalities, including sub-microscopic deletions and duplications of DNA, potentially many more patients could receive a diagnosis should they be afforded the opportunity of such testing. Although they might not expect it at the moment, it is inevitable that health professionals will come across patients with a rare chromosome disorder but they might not consider such disorders in assessing the patient. There must be improved undergraduate and postgraduate training and continuing professional development, not only for doctors but for ALL health professionals to ensure timely diagnosis. Detailed knowledge of all individual rare chromosome disorders would obviously not be possible nor expected for non-specialist health professionals but knowledge of the most up to date technology available to diagnose such disorders is imperative if the diagnostic process is to be made more efficient, both in terms of time and cost, to the NHS, to the individual health professional and to the patient. Rare chromosome disorders tend to lead to complex, multisystem effects, sometimes passing down through generations. The ability to take a decent family pedigree and recognise the signs that would trigger suspicion of a rare chromosome disorder and thus the necessary testing and referrals should be a basic part of every doctor’s (and nurse’s) toolkit, drummed into them as undergraduates and on through their professional careers.

CONSULTATION QUESTION Do you agree that the UK National Screening Committee should take into account the benefit of screening in reducing the ‘diagnostic odyssey’ and in allowing informed choice for subsequent family planning?

Comments

We endorse the comments on this question made in RDUK’s response to the Department of Health (England) on the consultation on the UK plan for rare diseases.

CONSULTATION QUESTION How can the NHS best ensure research in rare diseases carried out by the NIHR biomedical research centres and units is rapidly transferred into practice for the benefit of UK patients and their families and carers?

Comments
We endorse the comments made in answer to this question in RDUK’s response to the Department of Health (England) on the consultation on the UK plan for rare diseases. Specifically:

- Rare disease research should be identified as a priority area by major public funding bodies.
- Those with expertise in rare disorders should be included on committees of funding bodies.
- Support should be given for the development of clinical research networks.
- The NIHR Clinical Genetics Networks should be expanded to include all rare diseases.
- The R&D approval system should be streamlined for rare diseases.
- R&D approval should be granted to the Regional Genetics Services.
- Support should be given to the development of a network of genetic services host Trusts.
- R&D approval for rare disease research should cover all four home nations.
- Requirements for R&D approval for research into rare diseases should be more proportionate to the research being proposed.
- R&D fees should be standardized between sites.

CONSULTATION QUESTION: Do you agree that commissioners of care for people with rare diseases should assess options for improved care coordination, including named care coordinators?

Comments

We at Unique are of the firm belief that commissioners of care for people with rare diseases must assess options for improved care coordination. As mentioned before, rare chromosome disorders lead to a complexity of symptoms affecting not only different body systems and organs but also a varying mixture of cognitive and physical impairment, mental health needs, social and/or educational difficulties, as well as lifestyle changes for the whole family. Too often families and individuals face a shocking fragmentation of services which has a major impact on their daily lives and well-being. Appropriate and timely care coordination is essential to avoid these distressing barriers to achieving as normal a life as possible. Each patient with such a complex, multisystem chromosome disorder should be assigned a named, capable and empowered care coordinator. Too often, care coordination falls on the shoulders of exhausted family carers themselves who might not have the energy, capacity or inclination to carry out this role on top of the demands of everyday hands on caring. Too often, professionals do not provide care centred on the needs of the individual but rather centred on those individual aspects of the person’s disorder related to their specific professional interests. This is particularly true beyond transition into adulthood, when the paediatrician is no longer involved to coordinate at least some of the care. Too often, no health professionals will take responsibility for coordination of care of the most complex and sick patients once they...
transition into adulthood – a situation desperately worrying for the patient and their family. Each individual with a rare chromosome disorder, whatever their age, should be provided with a personalised care plan which should be reviewed regularly, updated according to need and from which the named care coordinator can work to provide the affected individual and their family with the best, most efficient and appropriate care possible.

CONSULTATION QUESTION: Do you agree that this list of criteria for expert centres should be the basis for future shaping of services?

- Co-ordinated care
- Adequate caseload for expertise
- Not dependent on a single clinician
- Arrangements for the transition from children’s to adults’ services
- Engaged with people with rare conditions
- Research active.

Comments

We at Unique believe that all the listed criteria for expert centres are essential. Specialist expert centres should be commissioned in each region for rare chromosome disorders. It would be most logical to expand the remit of the existing regional genetics centres to take on this role over a patient’s lifetime since they already see most rare chromosome disorder patients at diagnosis and for a few patients on an ongoing basis. Clinical geneticists and genetic counsellors are already used to working with patients in an holistic fashion, taking into account the complex needs of their patients and their families, and then communicating those needs to the patient’s local health professionals, at least at the time of diagnosis. They have the skills to make ideal care coordinators, or to train such coordinators, if given sufficient financial and staffing resources. We would support a “hub and spoke” model of communication so that expert centres can inform and educate local services about the patient’s specific disorder and their consequent needs. We would like to see patients being able to access all the relevant specialist professionals they need in the expert centre at the same time, to reduce the number of medical and other appointments they have to attend at present and to circumvent the constant need to repeat their stories to numerous professionals. Streamlining the service in this manner would surely lead in the long term to financial costs savings for the NHS as well as more efficient use of the health and other professionals’ time. We would also suggest that the use of VOIP might facilitate this multi-professional approach – patients and their families and the relevant expert professionals would not necessarily always have to meet in a physical venue but rather with the right communications equipment, they could have some “virtual” consultations, with the patient and family in their home surroundings or in a local professional’s
office or surgery. Alternatively, the experts involved from the specialist centres might be able to hold satellite clinics in venues local to the patient, if sufficient patients lived in that area. Such streamlined, multi-disciplinary service would facilitate dissemination of information and training about specific rare chromosome disorders, lessening dependence on a single expert clinician.

With regard to transition from children’s to adult’s services, this is a huge area of concern among Unique families, as alluded to in the previous section. So often families report to us being “cut adrift” from the supportive services they have enjoyed under the care of paediatric services. Families are left with no professionals to coordinate their “child’s” care, even when that child is profoundly disabled and medically extremely complex. Too often, coordination of the young adult’s ongoing care is a matter of chance, dependent on the goodwill and compassion of individual paediatric clinicians willing to manipulate the paediatric system to allow them to stay under their care for as long as possible. This situation cannot be allowed to continue. There are now far too many children coming through the system who have survived thanks only to developments in modern medicine and who only a relatively short time ago would have succumbed in childhood and yet do not have a named professional to coordinate their care. Equitable, timely and appropriate care coordination for these complex cases, particularly in adulthood, is desperately needed.

It is critical that expert centres engage with patients with rare disorders and their families, as well as with relevant patient organisations, where they exist. Patients and patient organisations must be helped and empowered to be active partners with expert centres and should be adequately funded to do so. Expert centres should be engaged in research, since they should have sufficient patients and expertise to warrant this.
CONSULTATION QUESTION: Do you agree that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance?

Comments

It is crucial that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance. The hub and spoke model mentioned previously would facilitate this. Moreover, expert centres should also network with other expert centres both nationally and internationally, especially for the rarest disorders where global reach is necessary to “find” sufficient patients with that disorder. Such streamlined, multi-centre networking would aid dissemination of knowledge about specific rare chromosome disorders and greatly enhance high quality training to the benefit of patient and professional alike.

Genetic testing

We at Unique would like to raise some points about genetic testing not mentioned in the consultation document but which are very important. Very pertinent points are raised in the RDUK’s response document. In particular, we agree that when a laboratory report is issued, the report should include a) advice on who to contact for further information and b) a recommendation where the patient should be referred to. This should happen systematically and as a routine and we would suggest that the reports should include signposting to relevant support organisations like Unique, from which valuable, verified information about the impact on the patient and their families of specific rare (chromosome) disorders can be obtained. We would also highlight the present inequity of access to the most up to date testing for rare chromosome disorders. There is no doubt from Unique’s surveys that patients in certain regions of the UK are far more likely to have access to e.g. array CGH analysis to diagnose rare chromosome disorders than do patients in other areas such as Wales, where we understand array CGH analysis is still not routinely available through the NHS. This is despite the well-documented greatly increased rate of diagnosis in developmentally delayed and cognitively impaired children afforded by array CGH analysis (up from ca. 7% to ca. 20%). The benefits to the patient and their family of receiving an early diagnosis are manifold and cannot be underestimated, not least the curtailment of the endless round of health appointments and unnecessary and often distressing tests and screening. The additional costs to the NHS of this inefficient “diagnostic odyssey” must be enormous.
CONSULTATION QUESTION: In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

While we at Unique are pleased that representation of patients with rare diseases on Local HealthWatch and HealthWatch(England) has been raised, we are concerned that Local HealthWatch will not be able to respond and react to the views of patients affected by individual rare chromosome disorders, since there will be few patients in each locality compared with patients with more common disorders and diseases. We endorse the recommendations made by RDUK in this respect.

Patient Information and support

We endorse the points raised in the RDUK’s response to the consultation document regarding patient information and support. An information prescription should indeed be made routinely available to every patient with a rare chromosome disorder when they are first diagnosed and should be the basis of a care management pathway coordinated by a suitable named professional. Information prescriptions should include:

- medical information including, where appropriate, possible treatment and/or therapy options
- when the patient will receive their personalised care plan
- information needed by carers and family
- information on financial support and welfare
- social information
- educational information
- how to access information on research into the condition
- patient support organisations and the information and services they provide
- which professionals are available to support the family, e.g. social services, therapists, behavioural specialists, psychologists and psychiatrists, respite/short-term care and support
- how to access psychological support and relationship counselling
- sibling support
- support for transition into adulthood

Families and patients with rare chromosome disorders so often feel totally isolated on receiving their diagnosis and would benefit greatly not only from being given information about the long term effects of their disorder but also having access to multiple sources of support, not least psychological support and counselling to help them cope.

We at Unique have worked hard over the last 28 years to gather information about the natural histories of specific rare chromosome disorders in our
comprehensive registry/database and have published over 150 information guides on specific rare chromosome disorders, reviewed by professionals expert in each particular disorder and by the families affected by that disorder, and researched and written by post-doctoral level members of staff. Many of the guides have been translated into a variety of languages. The guides are used by thousands of professionals and families worldwide. Affected families are often referred to us by their geneticists or local health professionals, although too often families are not given this information and have to resort to “Googling” or searching Facebook to find us. An information portal specifically for rare diseases in the UK, relevant to needs and cultures, should be developed so that patients and families are directed to sources of high quality information and support. In our experience though, internet-based portals would not suit all patients and families e.g. because they do not have access to the internet or they have intellectual impairment. We would suggest that PALS services in hospitals are not forgotten as a means of access to appropriate information. While accreditation of information is to be welcomed, like many other small third sector organisations, Unique has found the burden of complying with the demands of the Information Standard to be unacceptably, unnecessarily and disproportionately high. This has the perverse effect of the Information Standard discriminating against the high quality information produced by organisations like Unique, even though many NHS health professionals routinely use the information in clinic and routinely refer their patients and their families to these organisations. Furthermore, we are aware that some professionals in certain NHS institutions are prevented from disseminating to their patients and their families such high quality information because their local Trusts insist on having their own logo on any information given out by professionals working for that Trust.

CONSULTATION QUESTION: Do you agree that the United Kingdom should continue to participate in the Orphanet project?

Comments

| We at Unique fully support Orphanet but it is important to realise that it is only one resource and not the easiest of sites to use and navigate. |

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CONSULTATION QUESTION: What sources of patient information and support are available which are not listed in this plan?

Comments

We would reiterate RDUK’s response: that the DH should be concerned with and should be seeking to address in the plan for rare diseases what information and support is NOT available to patients and carers.

CONSULTATION QUESTION: Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?

Comments

Having devised, maintained and developed a comprehensive registry/database of rare chromosome disorders over the last 28 years, Unique has already demonstrated its strong belief that registers are an essential tool in rare diseases. Not only are they important to capture details and information about the natural histories of specific rare disorders, they are also hugely beneficial to facilitating research, gathering a critical mass of patients to make research feasible. While we agree that registers could be a core component of the service specification of expert centres, we would ask that existing registers/databases owned by charitable organisations like Unique are not overlooked, that they should be supported by funding bodies and that the extra demands placed on these organisations as more people are diagnosed and registered are provided for. Expert patients in organisations like Unique have a huge amount of information about rare disorders to offer the NHS and should not be ignored. Robust systems should be introduced for the NHS and health professionals to engage systematically with such organisations.

12. CONSULTATION QUESTION: Are there any areas of work that the UK Plan on Rare Disease needs to pay particular attention to in order to advance equality?

Comments

We are surprised that no mention has been made in the consultation document of monitoring the outcomes and effectiveness of a plan for rare diseases. This is surely essential if equality is to be achieved UK-wide for all rare disorders. Health departments will need to develop clear ways of assessing and measuring the impact of the plan. Furthermore it is not clear
from the consultation plan who will oversee and deliver the plan. There should be a designated team in the DH to take ownership, as well as a National Clinical Director for rare diseases to take overall responsibility.