CONSULTATION RESPONSE FORM

UK Plan on Rare Disease

Closing date for responses: Friday 25th May 2012. Please send your replies to: rarediseasesconsultationresponses@dh.gsi.gov.uk or alternatively send by post to: Sarah Bramley-Harker
Department of Health
Screening & Specialised Services Team
Room 5W35, Quarry House
Quarry Hill
Leeds, LS2 7UE

Please fill in and/or tick the appropriate response.

Name	Dr A D Dwarakanath FRCP Edin
Contact address	9 Queen Street, Edinburgh
Organisation representing (if appropriate)	Royal College of Physicians of Edinburgh
Postcode	EH2 1JQ
Contact telephone	0131-247 3608
Email	l.lockhart@rcpe.ac.uk

Before submitting your response please make sure that it has been saved in a name *[e.g. A N Other]* that will make it easier for us to track. Many thanks.

Please indicate the country the consultation and your comments relate:

UK-wide	√	*and/or:*	
England		*Northern Ireland*	
Scotland		*Wales*	

Are you responding:

- *as a member of the public*
- *as a health or social care professional*
- *on behalf of an organisation* √

If you are responding on behalf of an organisation, please indicate which type of organisation you represent:

NHS	
Social Care	
Private Health/Independent Sector	
Third Sector	
Regulatory Body	
Professional Body	√
Education	
Trade Union	
Local Authority	
Trade Body	
Other (Please give details)	

Consultation Questions

1. 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

Yes. As the document highlights, rare conditions are uncommon and unlikely to present frequently to the General Practitioner or general physician and so the majority of patients can anticipate prompt diagnosis, appropriate investigation and treatment for their common diseases.

Achieving the right balance between delayed diagnosis of a rare condition versus over-investigation of variants of common disease is challenging and the demand on limited resources for health care must be recognised. All trained doctors should be competent to recognise significant symptoms and signs, to investigate appropriately and treat when indicated. Unusual signs and symptoms, significantly abnormal results of investigation or failure of treatment should alert the clinician to possible atypical or rare disease. At this point, ready access to decision programmes may offer a valuable aid to further investigation and a prioritised consumption of resources in search of definitive diagnosis and treatment.

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

   Comments

It is important to ensure a firm general grounding before specialist training and then maintaining links among specialties with joint education events when rare diseases that may present to several different specialists or involve care across specialties are highlighted. Again, there is a need for the well trained generalist as well as the organ specific specialist to provide effective holistic care. Patient narratives and feedback as lessons learnt can be effective learning tools but trainees often lose sight of complex patients due to rotation of posts.

Tagging patients with mystery signs and symptoms so their journey can be followed by a trainee could help raise awareness and empathy for undiagnosed and rare diseases.

3. 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

Possibly. However, screening programmes have tightly defined parameters for clinical and cost effectiveness and must have acceptable specificity and sensitivity to justify either universal or targeted screening of at risk populations. The latter is likely to be more relevant for many rare diseases though 'group disorder' screening may become realistic whereby a common marker for a group of rare disorders might indicate the need for more extensive specific investigation eg screening for raised blood carnitine, steroid or other products of metabolic disorders in the newborn.

Detection of an untreatable and serious rare disease through screening raises ethical questions both for the parents in relation to genetic counselling and family planning and for professionals who may be giving information without thereby helping their infant patient.

4. 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

Centres of expertise for rare diseases should be welcomed by families and health professionals for the reasons outlined in the consultation document. Clinical expertise through wider experience of the condition, technical and laboratory backup to enhance investigation, monitoring and treatment and a focus for research will enhance clinical services.

Direct contact with dedicated experts and with other families experiencing similar challenges reduces isolation for those affected and their families and provides peer support, practical learning and teaching through shared experience and participation in research at the most meaningful level. The development and maintenance of centres of expertise in individual or groups of related conditions facilitates access to new therapies and their improvement and production.

5. 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

No, not by diagnosis alone as this excludes those orphan conditions that have not yet been defined and whose sufferers may receive less care if a disease related coordinator consumes resources for solely the named conditions.

There is a role for named care co-ordinators whose remit includes all individuals with conditions requiring multiple or widely scattered services. The role of the co-ordinators should be determined by users as well as by service planners and commissioners.

6. 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

Yes, all of these are important – there has to be recognition that clinicians running these services need adequate time for CPD and to liaise with similarly expert clinicians.

6. 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

Yes, but also the centre must address the need to know and publicise its wider regional or national links and have knowledge of other relevant networks including clinical, laboratory, technical research and teaching.

8. CONSULTATION QUESTION: In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

This could be done by involving local media in describing the activity of local services to the population, including real life stories from those who have benefitted from services.

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

Comments

Yes, and awareness should be raised of it among the public and professionals.	

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

Comments

Families and friends not only need information and support from the health services, but can provide information and support to the patient as well.

11. 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

Yes, but they must be flexible enough to reassure those individuals who are anxious regarding any identifiable data that total anonymity is feasible if universal registration of all patients is required. The development of registers would take time and infrastructure and it is important that this is not underestimated.

There would need to be a mechanism to address the right to removal of data on the request of an individual.

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

Yes, for example:

- children's rights in relation to screening, data collection, research involvement and consent to ongoing registration that was initiated by parents or guardians.
- genetic counselling for wider family by experts in rare disease identification and heredity.
- centres of expertise offer an ideal situation for the use of telemedicine to support and educate families and their local health care teams as well as trainees and students in all health care fields.

Before submitting your response, please make sure that it has been saved in a name *[e.g. A N Other]* which will make it easier for us to track.

Please email or send this form back by 25 May 2012 to:

By email: **rarediseasesconsultationresponses@dh.gsi.gov.uk**

By post: **Sarah Bramley-Harker**

Department of Health

Screening & Specialised Services Team

Room 5W35, Quarry House

Quarry Hill

Leeds, LS2 7UE

Many thanks for your response.