



Genetic Alliance UK
Supporting. Campaigning. Uniting.

Consultation Response

National Institute for Health and Clinical Excellence

National Quality Board Engagement Exercise

Response by Genetic Alliance UK

1. Genetic Alliance UK (formerly Genetic Interest Group) is the national charity supporting all those affected by genetic conditions. Genetic Alliance UK aims to improve the lives of people affected by genetic conditions by ensuring that high quality services and information are available to all who need them. Our membership represents more than 150 voluntary organisations working for a wide range of conditions.
2. Our response to this consultation was submitted by pro-forma. The following is a transcript of our response.

Question 1: Are there any significant gaps or omissions in the proposed library? If so, what are they?

3. In our response to the White Paper, Equity and Excellence: Liberating the NHS a year ago, we raised our concern that the production of just 150 Quality Standards will leave the majority of health conditions unsupported. Inevitably producing a disease specific Quality Standard for every condition is a never-ending task. This should not discourage us from attempting to create a set of Quality Standards which has something to offer every patient in the new NHS.
4. There are many thousands of rare genetic diseases for which it would be infeasible to develop Quality Standards. To ensure these patients have Quality Standards to support the delivery of their expected outcomes of care, we believe there should be a set of broad, pathology based Quality Standards such as that proposed for neurological conditions entitled “Neurological problems (relatively uncommon neurological problems e.g. muscular dystrophy)”. This would allow sets of rare genetic diseases which are dealt with by a particular medical specialty to be covered by a single Quality Standard.
5. The existence of Quality Standards become more valuable as conditions become more complex and less common, as it becomes more difficult for patients to intuit what high-quality care for their particular condition would look like.
6. We believe this approach would allow the Quality Standards to define high-quality care for as many patients as possible.

Question 2: Of the topics in the proposed library, are there any aspects of the care pathway or clinical area that it would be particularly important for the Quality Standard to address?

7. Genetic Alliance UK has recently relaunched Syndromes Without A Name (SWAN) the support group for children with undiagnosed genetic conditions. Based on our work with this community we believe it is essential that the following issues are addressed by the “Managing symptoms with an uncertain cause” Quality Standard:

Recognition of long-term uncertainty

8. A common issue for families with a child with an undiagnosed condition is a failure of the system to ensure continuity of contact with the family once diagnostic options have run out. Families have been told to “come back in ten years”; a direction which, if they had followed, would have meant they missed out on a valuable opportunity to participate in a clinical trial, and which fails to take account of the rapid change in healthcare knowledge currently occurring.
9. Families which have children with undiagnosed conditions should not be left to drift in this way. There should be a continuous relationship with a healthcare professional who should work to keep them informed of their options as they arise, and as their child ages.

Continuity of care into adulthood

10. For many children with undiagnosed conditions, their healthcare coordinator is their paediatrician. They are equipped with the knowledge to take an overview of their patient’s healthcare needs and provide ongoing contact with the family concerned. We have countless examples of children who benefit from good management from a paediatrician whose health deteriorates seriously when they become adults and their healthcare needs fall outside the remit of paediatrics. It is essential that patients are passed on effectively through the healthcare system as they age.

Patient held medical record

11. For patients with poorly understood health conditions, it may take many years for a physician to understand their healthcare needs and provide appropriate treatment. If patients such as these travel and become ill, it is imperative that their temporary physician understands their specific healthcare needs, and does not assume they will be able to treat the patient without guidance. A patient held medical record or a requirement to contact the lead physician should be a component of quality care.
12. Rare Disease UK (RDUK) is a multi-stakeholder initiative of Genetic Alliance UK. In February this year, RDUK published their strategy report outlining a vision for a rare disease plan in the UK. Large parts of this document are relevant to the Quality Standard “Long-term conditions, people with comorbidities, complex needs”.
13. In particular, Genetic Alliance UK would like to see designated Care Coordinators for patients with complex needs. Care Coordinators should liaise between patients and the services they use, and ensure that the right services are brought together at the right time. The Care Coordinator should be supported by the relevant centre of excellence to provide them with expert medical advice and the necessary knowledge of the condition. The Care Coordinator should assist in liaising between the different professionals involved in the care of a patient, again making sure that services are used more effectively.

14. RDUK’s strategy report is available here:
www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf

Question 3: Are there any other cross-cutting topics that address health improvement and / or patient experience issues for which a Quality Standard would be of benefit?

15. Genetic diagnosis is increasingly being delivered outside of the context of a regional genetic service as knowledge of the genetic component of conditions increase, and as technology becomes cheaper, better and more widely available.

16. This may or may not be appropriate, according to the specific case. For example a diagnostic test for some arrhythmias may be appropriately delivered by a cardiologist, but if the patient is making reproductive decisions, then genetic counselling regarding this diagnosis needs to be provided by a genetic counsellor or a clinical geneticist.
17. A Quality Standard on genetic testing and the provision of genetic counselling would ensure that as genetic healthcare becomes more frequently used across the NHS, the quality of genetic healthcare delivery remains high, and the appropriate healthcare professional is delivering it.

Question 4: Which are the topics where the interface with social care is particularly important and should be considered in developing the quality standard?

18. Genetic Alliance UK did not answer this question.

Question 5: What are your views on the Quality Standards published to date?

19. All of the Quality Standards published so far have an extensive list of “publication partners”. Genetic Alliance UK believes this approach is extremely valuable and hopes that this approach will continue for future Quality Standards.



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