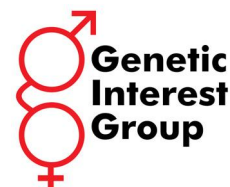


Consultation Response

Tomorrow's Doctors 2009: a draft for consultation

General Medical Council

Response by the Genetic Interest Group



The Genetic Interest Group (GIG) is a national alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders.

Whilst individual rare diseases are seldom met, rare diseases itself is not uncommon: one in seventeen people in the UK will develop a rare condition at some point in their life¹. Four out of five of these conditions are genetic in origin² whether inherited, due to a chromosomal abnormality, or caused by a spontaneous point mutation.

Rare disease requires special consideration:

- Symptoms of conditions such as autism and epilepsy can often mask rare disease.
- Rare inherited conditions can occur at significantly higher prevalence in groups of particular ethnic origin, or groups with higher rates of consanguinity.
- Rare conditions are often multifactorial in nature and require specialised coordinated care.
- Care for particular rare conditions may be at odds with best practice for treatment of comparable common conditions.

Outcomes 1 - The doctor as a scholar and a scientist

Paragraph 152 details the requirements for application of biomedical scientific principles (including genetics and molecular biology), method and knowledge to medical practice. A recurring requirement in this section of the guidance is knowledge surrounding common conditions. Whilst GIG would not expect all medical practitioners to have expert knowledge on individual rare diseases, we believe that all doctors should be aware of the strong likelihood that they will see patients affected by rare disease during their careers, and aware of the possible courses of action in such an eventuality. Many of our member charities' members report barriers to treatment of rare disease at the primary care level, such as difficulties in achieving referral to specialist care; we believe greater awareness of rare disease across the medical professions would begin to mediate this problem.

Outcomes 2 - The doctor as a practitioner

Paragraphs 158g and 158h stipulate the involvement of patients in the formulation of treatment plans and promote patient choice. Whilst applauding these statements, we would like to add that our member charities' members have frequently described experiences with healthcare professionals during which their expert knowledge of their own condition is marginalised and/or ignored. We feel that doctors should recognise that patients can in many instances contribute a great deal more than simple informed choice when planning their healthcare. Requests from patients for specialist care, when evidence-based, should be supported and enabled.

Outcomes 3 - The doctor as a professional

Paragraph 165e requires the recognition of equal treatment for patients and that opportunities may be limited by other's perception. In our own research, we have found disparity in access to familial cancer treatment between White British groups and ethnic minority groups³. GIG therefore applauds inclusion of this requirement in the draft guidance.

References

1. Communication on Rare Diseases: Europe's Challenges, European Commission, Nov 2008.
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2. Rare Diseases: Understanding this Public Health Priority, Eurordis, Dec 2005.
http://www.eurordis.org/article.php3?id_article=918

3. Ethnic Monitoring in Clinical Genetics, Dr P Mehta, Genetic Interest Group, Jul 2003.
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