Clinical genetics

Educating the gatekeepers

The 'Education of Genetic Disorders through Multimedia Sources: Knowledge, Education and Dissemination (KEDs)' project team in the National Centre for Medical Genetics outlines the initiative that counters a manpower crisis in clinical genetic services

The poor provision of clinical genetics services in Ireland was highlighted at the 2013 European Society of Human Genetics meeting held in Paris recently: Inequalities in Genetic Services (Stanislas Lyonnet and Jörg Schmidtke) https://www.eshg.org/sunday2013.0.html WS09.

A project team comprising **Dr Sally Ann Lynch,** Consultant Clinical Geneticist, **Dr Jillian Casey**, PhD Researcher, **Dr Alana Ward**, PhD Genetic Counsellor, **Rosie O'Shea**, Genetic counsellor, and **Jackie Turner**, Genetic Counsellor, is behind the 'Education of Genetic Disorders through Multimedia Sources: Knowledge, Exchange and Dissemination (KEDs)' project, which looks to address the manpower crisis that currently exits in clinical genetic services.

The National Centre for Medical Genetics (NCMG) struggles to provide a comprehensive service, with staff ratios at 10-20 per cent of equivalent international units. Given the straitened economic environment and reductions in health service head counts, staffing levels are unlikely to improve for the foreseeable future (see figure 1).

This situation means that it is difficult for the NCMG to even 'stand still' in terms of the service that it can provide. While the service comes under increasing pressure; science and technology marches on. "We have seen a doubling in the number of genes which we now know can cause disease *(see figure 2)*," the team stated. This in turn has led to huge increases in requests for genetic tests and counselling.

'Precious' appointment slots

In 2012, the NCMG recognised that waiting times for clinics were already exceeding 12 months, and so introduced a triage process in 2012 to optimise referrals. This process results in unnecessary appointments being avoided if the patient tests negative, thereby freeing-up precious appointment slots. The clinical genetics service cannot offer regular updates to our referrers using traditional methods because they are too labour intensive. Focusing on new media techniques, however, allows the service to reach its referrers without having to be physically present.

Details of the 'Education of Genetic Disorders through Multimedia Sources: Knowledge, Exchange and Dissemination (KEDs)' project are available on http://www.ucd.ie/medicine/rarediseases/.

In parallel, research by members of the team, joint-funded by a number of agencies including the Health Research Board (HRB) and National Children's Research Centre (through the Medical Research Charities Group) and Temple Street Children's Hospital has successfully identified three new disease genes prevalent in the Irish population.

The research means that patients could be offered a) genetic-based diagnostic tests avoiding invasive diagnostic procedures; b) carrier testing; and c) accurate risk assessment and genetic counselling. However, the research and its relevance to the service's population was in danger of being ignored, as the service did not have the ability to translate its findings into routine clinical practice.

Clinical translation

A top-up KEDs grant of approximately €9,000 from the HRB, relating to the disease gene research, has been used to address some of these barriers and enabled the clinical genetics service to:

a) Translate testing of our three genes from the research into the diagnostic laboratory at NCMG.
b) Develop a microsite, http://www.ucd.ie/medicine/rarediseases/ hosted by UCD, which gives practical information on the clinical genetics service, including how referrals are processed. In addition, the NCMG has included information on common genetic disorders such as haemochromatosis (in collaboration with **Dr Stephen Stewart**, Mater Hospital), as well as providing information on its disease gene identification and its relevance to Irish health.
c) Develop and publish a handbook answering common clinical queries to the service, which can be downloaded by healthcare professionals from the website.
d) Host a number of workshops for healthcare professionals throughout the country, highlighting the NCMG's research, the website and launching the handbook.
e) Publish posters instructing healthcare practitioners on how to go about testing for cystic fibrosis, and;
f) Develop short, animated videos explaining consanguinity and autosomal recessive inheritance for use by both healthcare professionals and their patients (which are a work in progress). These will be hosted on the website.

Educating healthcare professionals

The NCMG has included scenarios guiding healthcare professionals through common genetic queries such as: Patients with family history of cystic fibrosis, x-linked disorders, Down's syndrome, intellectual disability or concerns about consanguinity.

They sourced support from two GPs, **Dr Mel McEvoy** and **Dr Peter O'Keane**, and a specialist midwife, Jane Dalrymple, to ensure their handbook was clear and usable. Mark Byrne, Communications Manager, UCD School of Medicine and Medical Science, has been instrumental in the development of the handbook and microsite.

Education programmes elsewhere in Europe have used similar formats to raise awareness of clinical genetics services locally and how to best utilise what they can offer. Healthcare professionals in primary care are often the gatekeepers for families to access such services and with the current under-resourcing, this is a timely project to help facilitate knowledge of clinical genetics and how to effectively help families in the current climate.

The Centre's ultimate aim is to improve the genetic health of the Irish population through education of healthcare professionals, as it currently cannot provide this service in person. Time will tell whether the efforts are effective.


Dr Sally Ann Lynch


Dr Jillian Casey


Dr Alana Ward

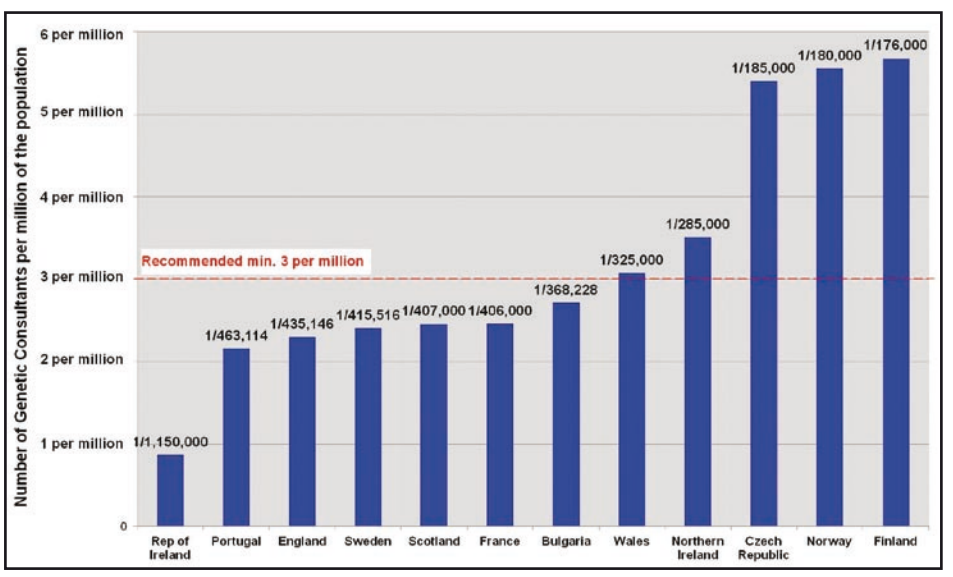


Figure 1: Consultant staffing levels across 12 European countries

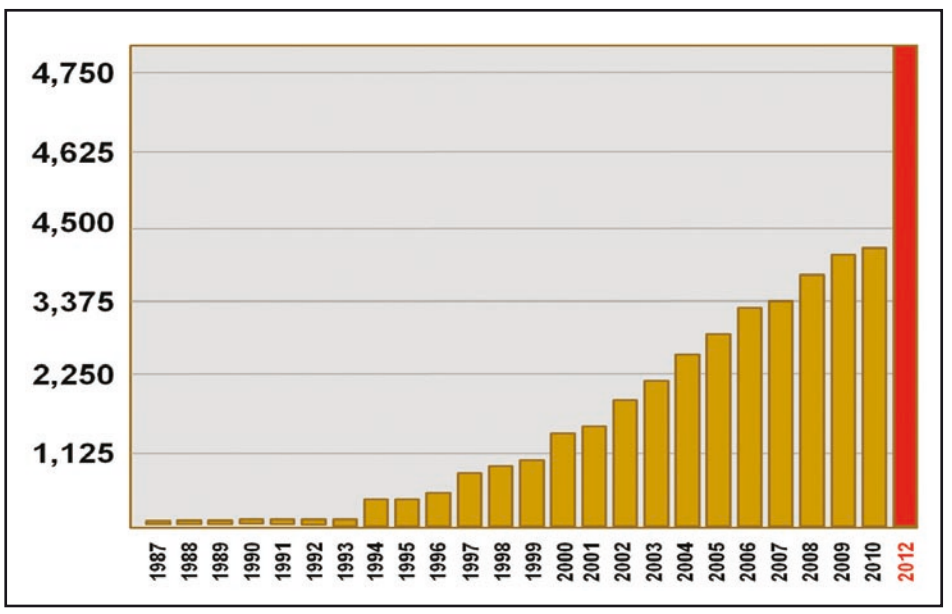
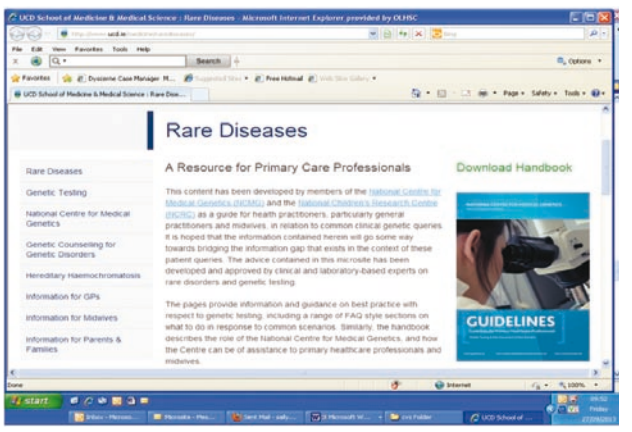
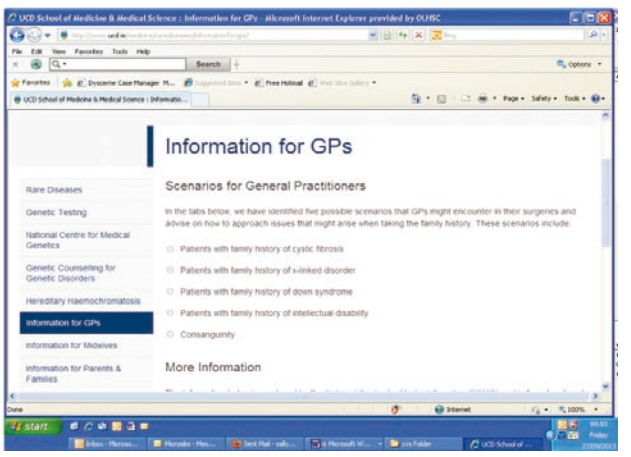


Figure 2: http://omim.org/statistics Number of new disease genes identified year on year.



Screen shot from the new microsite.



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