EDITORIAL

Clinical genomics testing: mainstreaming and globalising

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In many countries, the practice of Clinical Genetics is changing, with genomic testing being incorporated into mainstream clinics and managed by non-genetics specialists. Of course this represents a paradigm shift, with extensive educational and service development needs. Slomp et al. report on the process of integrating a Genetic Counsellor into primary care (General Practice) [1]. The rationale of having a Genetic Counsellor in this setting is that it would reduce the need for referral to hospital departments and improve access to genetic testing. This paper emphasises that the process of integrating a Genetic Counsellor into a primary care setting is sequential - there is initial resistance and uncertainty about how to collaborate with a Genetic Counsellor among Primary Care Physicians. The authors identify a series of barriers and facilitators to integrating a Genetic Counsellor into this setting. Interestingly, van Vliet et al. report that haemoglobinopathies may be under-diagnosed in primary care, identifying a need for greater integration of genomic health professions into this sector [2].

A persistent challenge in utilising genetic testing in mainstream medicine is the process of informing relatives of their genetic risk. There is a conflict between the duty of confidentiality to the patient and the rights of relatives to understand their genetic risks. In some countries, there is a legal duty for the person with the genetic condition to inform their at risk relatives so they can seek Genetic Counselling. Phillips et al. describe the Belgian legal context for this important issue [3].

Routine use of genomic testing in mainstream medicine also has the potential to generate large amounts of genomic data. Ormondroyd et al. highlight the sources of genomic health data information in the United Kingdom (UK): clinical testing, large-scale research projects and direct-to-consumer testing [4]. A focus group study identified areas of concern for policy development in the important area of genomic data generation as part of routine healthcare.

The role of genetic testing in Oncology has been long established. In this issue, we publish an ERN GENTURIS guideline for diagnosis and surveillance of people affected by schwannomatosis [5]. Recommendations for genetic testing, imaging and annual surveillance are made. De Oliveira et al. report the findings of cancer multi-gene panel testing in a large cohort of Brazilian patients with cancer [6]. Results were broadly comparable to other populations, but there may be some relatively specific pathogenic variants in the Brazilian population. An independent study from Brazil explores the clinical and genetic correlations of microsatellite instability in colorectal cancer [7].

Clinical genomic testing in a specialist setting is likely to remain crucial for diagnosis of rare diseases. Harvey and colleagues identify a novel splice site variant in OPA1, characterising an underrecognised disease mechanism associated with this gene [8]. Pelizaeus-Merzbacher disease is recognised as genetically heterogeneous. In this issue we publish a report of a Pelizaeus-Merzbacher-like presentation associated with bi-allelic variants in MAL [9]. This should be considered in the differential diagnosis in appropriate cases. Genetic testing in rare disease also helps to define clinical syndromes, which may have useful functional and prognostic information. Genetic testing defines a broader range of individuals affected by Rubinstein-Taybi syndrome than by clinical criteria alone [10]. This cohort study provides valuable insights into the adult clinical picture in this rare disease, helping to guide clinical management. DYRK1A-associated syndrome can only be confidently diagnosed with genomic testing. In this issue, Morison et al. report on social interaction and communication profiles in this rare condition [11].

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AM conceived, wrote and edited this piece.

COMPETING INTERESTS

The author declares no competing interests.

ADDITIONAL INFORMATION

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