EDITORIAL

A new impact factor for EJHG in 2022

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The impact factor for EJHG in 2022 is 5.2 (compared to 5.3 in 2021). EJHG now ranks 30th out of 172 Genetics & Heredity journals. This is the journal's highest ranking since 2011.

What papers could you cite in this month's issue of EJHG? It is relatively easy to select which genomic tests should be used based upon their diagnostic performance. However, designing a clinical service that meets families needs is somewhat different. Crellin et al. performed a systematic review to identify what matters to parents of children with a rare condition [1]. Parents value feeling "cared for", continuity of clinician involved, being kept informed and being offered psychosocial support and follow up. Many of these factors can be supported by individual clinician behaviours.

Phenotypic studies of rare genetic conditions are vital to allow us to inform families of likely symptoms. FINCA syndrome is characterised by Fibrosis, Neurodegeneration, and cerebral angiomatosis. In this issue, a new series of FINCA patients emphasises that the initial presentation can be with non-specific developmental delay [2]. Evidence of genotype phenotype correlation is presented. Hedberg-Oldfors et al. report biallelic RNH1 variants as a cause of a novel developmental disorder [3]. The presentation was with cataracts and infection induced developmental regression. Accurate variant classification is vital for genomic diagnosis. Peripheral blood DNA methylation signatures are emerging as a useful diagnostic tool: here for Renpenning syndrome [4]. Bouassida et al. report that 2p25.3 duplications have a non-specific phenotype of autism or intellectual disability, they are frequently inherited from apparently unaffected parents [5].

Some people affected by a genetic condition choose to use a reproductive medicine technique so that they can have a child without the condition. Neurofibromatosis type 1 (NF1) is an autosomal dominant, fully penetrant condition with marked variation in expressivity. In this issue, the Dutch experience of Preimplantation Genetic Testing (PGT) for NF1 is reported [6]. Complications of in vitro fertilisation (IVF) were not more frequent in women with NF1 than in women without NF1. A pregnancy rate of 24% per cycle was reported, comparable with other autosomal dominant conditions. Bettinaglio studied genomic factors that may explain why some people with NF1 have predominantly spinal involvement, this may aid clinical counselling in future [7].

Direct to consumer (DTC) genetic testing remains controversial. Many clinicians view DTC results with caution, and not equivalent to clinical laboratory testing. Ruehl et al. used an online survey to assess people's interest in, and expectations of, DTC before and after exposure to sample DTC disclaimer notices [8]. They found that people with greater self-reported genetics knowledge read the DTC disclaimer notice more closely. However, there was no association between how deeply the DTC disclaimers were processed and the expectancies of what DTC could tell people. The effectiveness and appropriateness of DTC disclaimers requires further study.

Genetic testing can play an important role in helping people understand their risk of cancer. Henkel et al. used a 123 gene cancer panel to sequence 6941 German people with breast or ovarian cancer [9]. They refined this to a core 14 gene panel for breast and ovarian cancer. This core panel would identify a monogenic cause in 10% of patients, the expanded 123 gene panel only diagnosed an additional 1% of patients and very significantly increased the workload associated with variants of uncertain significance. Of course, environmental risk factors play an important role in carcinogenesis. Maina et al. report a mendelian randomisation study examining the role of different types of obesity in pancreatic cancer [10].

Does Clinical Genetics have a role in palliative medicine? It is possible that people nearing the end of life with genetic causes of cancer or neurological conditions might undertake genetic testing to try and inform the healthcare of their relatives. Undertaking genetic testing in this setting was reported as challenging, as it may not be perceived to be relevant but that it was important to consider the benefits to the family [11].

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