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EDITORIAL

What's new in EJHG in April

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Welcome to the April 2021 issue of *EJHG*. What can you learn from us this month? Well, Hochstenbach and colleagues provide evidence that, with the rise in use of genomic techniques, laboratories are losing competence in traditional cytogenetics [1]. This might negatively impact patient care. Phelan-McDermid syndrome is a classic chromosome deletion disorder, which can be diagnosed by cytogenetics. Morgan and co-workers define the speech and language phenotype, which will help guide clinical care [2]. The genomic technologies which have replaced cytogenetics are vital for identifying the aetiology of neurodevelopmental disorders. Balasubramanian et al. use exome sequencing to identify a cohort of individuals with *SIN3A* related disorder and define the phenotype [3]. Exome sequencing is also a powerful technique to identify novel causes of rare disease. In this issue, Bell and colleagues describe *GIMAP6* as a novel immunodeficiency gene [4]. Liisa et al. use the same technique to study the cause of a common neurological disorder—vascular dementia—illustrating the applicability of exome sequencing across the disease spectrum [5]. Long read sequencing may help resolve variants not identified by traditional exome techniques (Gilisen), but may not be ready for clinical use just yet [6]. Despite our increasing understanding of how to use genomic techniques for diagnosis, functional validation of variants in experimental models is often required. This is illustrated by a report of *ABL1* pathogenic variant clustering in the functional domain of the protein [7]. All of our advances in genomic testing are for naught if we cannot engage patients and families in using them. Genomic techniques can reveal a myriad of unanticipated findings, complicating the consent process. Harriet describes the new concept of "dynamic consent" in response to this [8]. Practical application of dynamic consent is considered by "CTRL" an online platform, designed to aid with the consent proves for people considering a genomic test [9]. We hope you enjoy this month's issue, our social media channels are open for feedback and discussion (twitter @ejhg_journal).

Compliance with ethical standards

Conflict of interest Salary support was received from UK NHS and University of Sheffield.

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