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EDITORIAL

Out now in May's EJHG

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What should you read first in the May edition of the journal? Perhaps start at the beginning, with one of the first papers? Implementing genome sequencing at national level in healthcare systems is recognised to raise multiple ethical issues. A paper by the UK-France Genomics and Ethics Network suggests these can be negotiated by redefining the rights of the individual and the collective responsibility to share genomic information for the common good [1]. The COVID-19 pandemic is the biggest challenge facing healthcare systems. How can the Clinical Genomics community help? One role has been to employ our experience of biobanking to set up multi-center biorepositories of samples from COVID-19 patients [2]. Screening family members for evidence of Marfan syndrome is resource intensive, but vital. However, Chesnau et al. [3] suggest that de novo variants are more common than previously recognised, with implications for planning how to screen families. Lack of stored DNA from individuals who died from cancer often precludes genomic testing, with implications for management of at risk relatives. Bennett et al. describe successful extraction of DNA from fixed, paraffin embedded tissue for genomic testing [4]. This will facilitate identification of causal variants in families and help with screening of at risk relatives. Evidence for the clinical utility of genome sequencing is growing. This paper describes improvements in molecular diagnosis using genome sequencing in polycystic kidney disease [5]. Genome sequencing also continues to expand our understanding of phenotypes. Here, TUBA1A is shown to cause congenital fibrosis of the extraocular muscles without the cortical malformations more typical of this gene [6]. We hope you enjoy the edition, our social media channels are open for feedback and discussion (@ejhg_journal).

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