EDITORIAL

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The *European Journal of Human Genetics* is the official journal of the European Society of Human Genetics (ESHG). The ESHG-Young Committee (ESHG-Y) is an important part of ESHG. ESHG-Y is highly active and helps to promote and develop the careers of young Clinical Geneticists, for example by organising focussed training sessions and events [1].

Review articles are a great place for young geneticists to get a rapid overview of a topic. In this issue, Al Sayed et al. present an overview of genetic testing for congenital hypogonadotropic hypogonadism (CHH), a rare disease characterised by low circulating sex steroid concentrations and reproductive immaturity [2]. Genetic testing may be able to provide a clear diagnosis, especially when there is difficulty distinguishing delayed puberty from CHH. Pasquier et al. report a systematic review of clinician attitudes towards the implementation of reproductive carrier screening [3]. Eighteen papers were identified and findings were summarised under ten themes: such as ensuring all couples can access screening and the importance of information resources. Best et al. report using behaviour change theory to support practitioners offering reproductive carrier screening [4]. Expanding the availability of reproductive genetic carrier screening will require non-genetic specialists to play a role. Behaviour change theory can be used to identify the influences on the behaviour of clinicians and to identify interventions that support the new desired behaviour. Best et al. report barriers and enablers to non-specialist clinicians offering expanded carrier screening. Van Vliet et al. report the needs of patients and primary care doctors when offering haemoglobinopathy screening [5]. Some academics believe that polygenic risk scores may be used in reproductive decision making in future. Lowes et al. report a qualitative study of people who have received polygenic risk scores via direct to consumer testing [6]. Some found the information empowering while others were distressed. Diagnosis of a single gene disorder is key to offering reproductive options. Smith et al. report a qualitative analysis of what is perceived to be the family-level factors defining the utility of genome sequencing and a genomic diagnosis [7].

In this issue, we publish a series of clinical reports illuminating the genomic causes of several unusual presentations. In 1977, a condition of progressive gingival fibromatosis and deafness was reported by Jones. A specific variant in the REST gene is reported as causing Jones syndrome [8]. Nakano and Bánfi provide an insightful commentary on the historical aspects and genetic mechanisms [9]. Bottillo et al. provide further evidence that bi-allelic CHEK2 variants might be associated with cancer syndrome and chromosome instability [10]. Aerden et al. report a cohort of people with TRIP12-syndrome recruited via an ERN ITHACA call [11]. They report 35 variants affecting TRIP12, distributed throughout the entire gene with no clear variant hotspots. The clinical characteristics of 38 individuals with intragenic TRIP12 variants include variable developmental delay, intellectual disability, important speech delay, autism, obesity and distinct facial features. In 10%, there was developmental regression and 13% had seizures. Neyroud et al. report that LARS2 variants present with isolated premature ovarian failure and a linked comment by Vona [12, 13]. The clinical spectrum of DYNC2H1 variants is further described with Ellis van-Creveld like presentations noted [14].

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ADDITIONAL INFORMATION
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