EDITORIAL

# Hitting the heights with CiteScore

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This month brings the release of the 2024 citation metrics from scopus and clarivate. The European Journal of Human Genetics now has its highest ever ranking by CiteScore. The *European Journal of Human Genetics* now has a CiteScore of 9.9 and is ranked 8/99 in clinical genetic journals. The European Journal of Human Genetics is a highly regarded and well-read vehicle for some of the best clinical and basic science genomics research relevant to human genetics.

Documenting the clinical spectrum of rare genetic conditions is vital to help clinicians manage them. Watts and colleagues describe a novel series of people with Carpenter syndrome - adding laterality defects as a novel feature [1]. NOTCH1 variants are associated with Adams Oliver syndrome. A novel series of people with NOTCH1 variants identities a broader phenotypic spectrum - only a minority meet diagnostic criteria for Adams Oliver syndrome [2]. Tetralogy of Fallow was the most common congenital heart defect. Schoch et al. report novel variants in CDC45, causing Meier-Gorlin syndrome [3].

Genome sequencing technologies have enabled the identification of novel genomic variants and mechanisms for disease. Fifty percent of primary familial brain calcification patients remain undiagnosed. In this month's journal, 5 prime UTR variants in PDGFB are validated as a rare cause of primary familial brain calcification, with implications for genomic diagnostics [4]. DNA methylation profiling continues to develop as a clinically useful tool. Niceta et al. show that DNA methylation signatures can distinguish between VUS and pathogenic variants in KMT2D Kabuki syndrome - supporting the use of DNA methylation for variant classification in clinical practice [5]. De Sabando et al. report somatic instability of intermediate repeat lengths in Htt, but no association with neurological disease [6]. Prenatal use of whole genome or whole exome sequencing to diagnose the cause of fetal brain abnormalities is reviewed by Marchionni et al. [7].

Genome sequencing also facilitates the identification of novel rare conditions caused by single gene variants. Lu and colleagues characterise loss of function variants in MSL2 in a novel neurodevelopmental condition with dysmorphism and combinations of intellectual disability and autism [8]. This is another component of the epigenetic machinery causing early-onset neurological disease. Whole genome sequencing is reported to detect a cryptic chromosomal rearrangement in CCM2 causing multiple cerebral cavernomas in a family [9]. Detecting genomic variants is one thing, understanding their significance is another. Houge and colleagues report a comparison of the novel ABC variant reporting system against the standard ACMG system [10]. Functional studies of a genetic variants effect can help with classification. Calderan et al. report the use of a yeast 2-hybrid system to classify the pathogenicity of ACTA2 variants associated with aortic aneurysm [11].

Genomic analysis of rare conditions also helps illuminate the causation of more common diseases. Laver and colleagues report deletion of regulatory elements near FOXA2 as a cause of persistent congenital hyperinsulinism [12]. Adding to our understanding of the regulation of insulin secretion.

Genomic technologies also make a major contribution to understanding the cause of, and treating, cancers. Terradas et al. report NPAT as a novel gene predisposing to nonpolyposis colorectal cancer [13]. NPAT-deficient colorectal cancers have significantly higher aneuploidy levels. Genome sequencing for paediatric cancer patients can also be used as an "opportunistic genome screening" platform. Hammer-Hansen et al. report that around 3% of probands who had genome sequencing for paediatric cancer had an additional finding [14]. An average of 2.25 relatives per proband underwent cascade testing for these. It is interesting to note that the germline and somatic mutation rates of POLD1 variant carriers differ, the somatic mutation rate being higher [15]. This is explained by the fact that neoplasms lose the second (ie wild type) POLD1 allele.

Pearce et al. undertake a systematic review of the attitudes of the general public towards public health genomics [16]. The public has generally positive attitudes towards genomics, and high expectations of its benefits. In contrast to this paper - Muller et al. report a study in which members of the Australian public report concerns about genetic discrimination as deterring them from using genomics [17].

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## AUTHOR CONTRIBUTIONS

AM conceived, wrote and edited the paper.

## COMPETING INTERESTS

The author declares no competing interests.