EDITORIAL

A new impact factor for *European Journal of Human Genetics*

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We are pleased to report that the *European Journal of Human Genetics* impact factor for 2020 has risen from 3.657 to 4.246. The *European Journal of Human Genetics* provides a great forum to ensure your genomics research is both read and cited. Our editorial performance continues to improve too; time to first decision and time to securing initial reviews have been very significantly reduced. Our social media following is growing, we now have over 6000 twitter followers, which we use to help promote the research we publish. Much of our improved editorial performance is due to the hard work of our global panel of Section Editors, whom we thank.

Variant interpretation remains a major challenge in Clinical Genetics. This month Savige et al. report proposed modifications to the American College of Medical Genetics criteria to improve interpretation of genetic variants in Alport syndrome [1]. For example, specific "mutation hot spots" are defined for the Alport syndrome genes. It seems likely that further disease specific modifications to American College of Medical Genetics criteria will be produced in future.

Exome and genome sequencing has led to the discovery of hundreds of genes and variants that cause neurodevelopmental disorders. Following this, novel drug targets and therapeutics are being developed which will lead to clinical trials. Clinical trials for treatments of children with neurodevelopmental conditions can be especially challenging. Turbitt describes the treatment priorities of parents of children with Fragile-X [2]. This will help design clinical trials in a manner that families are most likely to engage with.

Trio exome/genome sequencing has been used extensively to identify the cause of paediatric neurological disorders. In this issue, Wagener apply this technique to children with cancer [3]. Considerably fewer causal variants were identified in these children than would be found in those with neurodevelopmental disorders. A high proportion of those with features suggestive of a cancer predisposition syndrome had no causal variant. This suggests a need to look beyond the exome for causal variants: oligogenic, non-coding or novel genes for example.

Genomic research can also provide insights into common conditions. A novel Genome Wide Association Study analysis is reported, which identified new loci for primary biliary cholangitis in Japan [4]. Exome sequencing in critically ill COVID-19 patients identified variants in immune system genes as predisposing to cytokine storm [5]. Lastly, the potential for exome sequencing to improve diagnosis of inherited retinal disease is described [6].

Genome sequencing of (apparently) healthy individuals can also provide scientific insights. Here, Italian individuals with bi-allelic loss-of-function variants ("Human Knock Outs") are described [7]. Ten individuals with such variants in known recessive genes were identified. Most had no or only very mild phenotypes. This underscores the need for in depth phenotyping to understand the penetrance of such variants in "healthy" populations. Two studies of SETBP1 syndrome using in depth phenotyping, in this issue, provide valuable insights into this rare disease [8, 9]. Helping define the associated functional strengths and weaknesses, which will help inform families.

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AUTHOR CONTRIBUTIONS

Dr Alisdair McNeill conceived and wrote this editorial.

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

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