European Journal of Human Genetics (2021) 29:889–890
https://doi.org/10.1038/s41431-021-00906-w

EDITORIAL

Clinical genomics—but faster

Alisdair McNeill[1,2]

Published online: 19 May 2021

Until recently clinical genomic testing was a slow and laborious process. Advances in sequencing and bioinformatics have made genomic testing more accessible and rapid in clinical practice. In this issue of *EJHG* John McDermott presents a new evolution of this concept—bedside testing for COVID-19 infection [1]. Elin Tonne describes the Norwegian experience of exome sequencing for craniosynostosis; clearly documenting the advantages of exomes for diagnosis in this group [2]. Exome and genome sequencing continues to identify novel genomic disease and assist in their characterisation. In this issue of *EJHG*, novel disorders associated with HEATR5B [3] and MADD [4] are described. This provides important insights into the diagnostic significance of variants in these genes. Our understanding of the molecular architecture of disease is also being revolutionised by exome sequencing. Previously heterozygous Junctophillin-3 variants were associated with an adult neurological phenotype, here bi-allelic variants are described in a neurodevelopmental disorder [5]. Widespread access to rapid, low-cost sequencing has its downsides. Francesca Forzano presents an ESHG position statement on how genetic testing and biobanks can be misused—with potential for discrimination—and how to avoid this [6]. In clinical practice, there is controversy over how to handle unexpected or incidental findings on exome sequencing done for developmental delay. With parents and clinicians sometimes having differing views on disclosure [7]. Increased sequencing also leads to centres identifying new cases of highly rare conditions. Often clinician researchers seek to publish these. But what type of "case report" is of interest to *EJHG*? Simply adding a clinical description of a rare condition is not enough. The report of WNT2B [8] is a great example of a modern "case report". First, published clinical descriptions are extremely few with a clear need to expand documentation of the phenotype. The clinical evaluation is exhaustive, adding novel features to the phenotype. The report documents the expression of WNT2B in human gut and adds to our understanding of disease mechanisms. We hope you enjoy this month's issue and find it useful for your clinical and research work. This month the *EJHG* editorial office relocates from Leiden to the University of Sheffield. All submissions will continue to be handled by the existing online manuscript submission system.

✉ Alisdair McNeill
a.mcneill@sheffield.ac.uk

[1] Department of Neuroscience, The University of Sheffield, Sheffield, UK

[2] Sheffield Clinical Genetics Department, Sheffield Children's Hospital NHS Foundation Trust, Sheffield, UK

References

1. McDermott JH, Burn J, Donnai D, Newman WG. The rise of point-of-care genetics: how the SARS-CoV-2 pandemic will accelerate adoption of genetic testing in the acute setting. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-021-00816-x.
2. Tønne E, Due-Tønnessen BJ, Mero IL, Wiig US, Kulseth MA, Vigeland MD, et al. Benefits of clinical criteria and high-throughput sequencing for diagnosing children with syndromic craniosynostosis. Eur J Hum Genet. 2020. https://doi.org/10.1038/s41431-020-00788-4.
3. Ghosh SG, Breuss MW, Schlachetzki Z, Chai G, Ross D, Stanley V, et al. Biallelic hypomorphic mutations in *HEATR5B*, encoding HEAT repeat-containing protein 5B, in a neurological syndrome with pontocerebellar hypoplasia. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-021-00832-x.
4. Abu-Libdeh B, Mor-Shaked H, Atawna AA, Gillis D, Halstuk O, Shaul-Lotan N, et al. Homozygous variant in *MADD*, encoding a Rab guanine nucleotide exchange factor, results in pleiotropic effects and a multisystemic disorder. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-021-00844-7.
5. Bourinaris T, Athanasiou A, Efthymiou S, Wiethoff S, Salpietro V, Houlden H. Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-021-00866-1.
6. Forzano F, Genuardi M, Moreau Y; European Society of Human Genetics. ESHG warns against misuses of genetic tests and biobanks for discrimination purposes. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-020-00786-6.

7. Tibben A, Dondorp W, Cornelis C, Knoers N, Brilstra E, van Summeren M, et al. Parents, their children, whole exome sequencing and unsolicited findings: growing towards the child's future autonomy. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-020-00794-6.

8. Zhang YJ, Jimenez L, Azova S, Kremen J, Chan YM, Elhusseiny AM, et al. Novel variants in the stem cell niche factor WNT2B define the disease phenotype as a congenital enteropathy with ocular dysgenesis. Eur J Hum Genet. 2021. https://doi.org/10.1038/s41431-021-00812-1.