ETHICS OF “OPPORTUNISTIC” WHOLE EXOME/GENOME SCREENING

As more and more patients with an indication for genetic testing are having their whole exome or whole genome sequenced, the possibility emerges to search for pathogenic variants that are not associated to the initial reason for the genetic test. Should such variants (so-called secondary findings) be actively looked for, because they could be of direct benefit for the health or reproductive choices of the patient and the patient’s family? Organizations such as the American College of Medical Genetics and Genomics, the French Society of Predictive and Personalized Medicine, and Genomics England state that “actionable” pathogenic variants should be routinely and systematically looked for and should be reported. In the March 2021 issue of the European Journal of Human Genetics, a committee on behalf of the ESHG, headed by Guido de Wert and Wybo Dondorp from the Department of Health, Ethics and Society of Maastricht University, the Netherlands, argues for a more cautious approach. In their publication, the authors discuss the wider ethical context of what they call “opportunistic genome screening” (OGS), with an emphasis on a balanced application of normative principles of proportionality, autonomy, economy and justice. They conclude that it is too early for OGS as a professional clinical standard, and point to cascade testing as an alternative to OGS. Please have a look at the paper for the details of this ongoing and important medical-ethical discussion.