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Sommario/riassunto

The field of cardiovascular genetics has tremendously benefited from the recent application of massive parallel sequencing technology also referred to as next generation sequencing (NGS). However, along with the discovery of additional genes associated with human cardiac diseases, the analysis of large dataset of genetic information uncovered a much more complex and variegated landscape, which often departs from the comfort zone of the monogenic Mendelian diseases image that clinical molecular geneticists have been well acquainted with for many decades. It is now clear that, in addition to highly penetrant genetic variants, which in isolation are able to recapitulate the full clinical presentation when expressed in animal models, we are now aware that a small but significant fraction of subjects presenting with cardiac muscle diseases such as cardiomyopathies or primary arrhythmias such as long QT syndrome (LQTS), may harbor at least two deleterious variants in the same gene (compound heterozygous) or in different gene (double heterozygous). Although the clinical presentation in subjects with more than one deleterious variant appears to be more severe and with an earlier disease onset, it somehow changes the viewpoint of clinical molecular geneticists whose aim is to identify all possible genetic contributors to a human condition. In this light, the employment in clinical diagnostics of the NGS technology, allowing the simultaneous interrogation of a DNA target spanning from large panel of genes up to the entire genome, will definitely aid at

uncovering all such contributors, which will have to be tested functionally to confirm their role in human cardiac conditions. The uncovering of all clinically relevant deleterious changes associated with a cardiovascular disease would probably increase our understanding of the clinical variability commonly occurring among affected family relatives, and potentially provide with unexpected therapeutic targets for the treatment of symptoms related to the presence of “accessory” deleterious genetic variants other than the key molecular culprit. The objective of this Research Topic is to explore the current challenges presenting to the cardiovascular genetics providers, such as clinical geneticists, genetic counselors, clinical molecular geneticists and molecular pathologists involved in the diagnosis, counseling, testing and interpretation of genetic tests results for the comprehensive management of patients affected by cardiovascular genetic disorders.