

# Single-center experience with genetic testing for cardiomyopathies using next-generation sequencing: results, challenges and the future

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**Introduction:** Cardiomyopathies represent an important cause of heart failure and genetic testing for cardiomyopathies has become an established care pathway in contemporary cardiology practice<sup>1</sup>, as it is valuable for risk stratification, treatment decisions, and family screening. Since 2020 we have established in Dubrava University Hospital a genetic testing program for cardiomyopathies using next-generation sequencing.

**Patients and Methods:** We are reporting on the results of genetic testing performed on patients with cardiomyopathies as well challenges and the future direction of the program. From June 2020 to June 2024 selected patients were subjected to genetic testing. We used standard Illumina TruSight Cardio Sequencing Kit, a panel covering 174 genes most associated with inherited cardiac conditions. Results were uploaded and analyzed using Variant Interpreter Illumina, cloud-based interpretation and reporting platform for genomic data.

**Results:** 77 patients underwent genetic testing in UHD (58 males, 40.4±14.4 years). Of those patients, 8 had previously undergone heart transplantation, and one analysis was postmortem. According to the phenotype, 39 patients were classified as having dilated cardiomyopathy, 31 had hypertrophic cardiomyopathy, 6 arrhythmogenic cardiomyopathy and 1 restrictive. Pathogenic or likely pathogenic mutation was identified in 40 patients.

**Conclusion:** Genetic testing provides insight into diagnosis, treatment, and prognosis of patients with non-ischemic cardiomyopathies, and directs screening which allows the identification of relatives at risk and initiation of appropriate medical and device therapies<sup>1</sup>. For a successful genetic testing program, a multidisciplinary team and close collaboration of different specialties are necessary, as well as a good patient selection. All this accompanied by a close follow up, genetic counseling and family screening enables a complete program of genetic monitoring.

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## LITERATURE

1. Yogasundaram H, Alhumaid W, Dzwiniel T, Christian S, Oudit GY. Cardiomyopathies and Genetic Testing in Heart Failure: Role in Defining Phenotype-Targeted Approaches and Management. *Can J Cardiol.* 2021 Apr;37(4):547-559. <https://doi.org/10.1016/j.cjca.2021.01.016>