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Genetic spectrum of idiopathic restrictive cardiomyopathy uncovered by new generation sequencing

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Background: cardiomyopathies represent a rare group of disorders often of genetic origin. The genetic spectrum of cardiomyopathies include more then 50 genes. While approximately 50% of genetic causes is known for hypertrophic and arrhythmogenic cardiomyopathy, the genetic spectrum of restrictive cardiomyopathy (RCMP) is widely unknown. Sanger sequencing approach revealed that sarcomeric protein gene mutations and mutations in structural proteins can be associated with idiopathic restrictive cardiomyopathy. The aim of the present study was to identify genetic background of idiopathic RCMP using new generation sequencing.

Patients and Methods: we applied new generation sequencing approach (Ion Torrent) to perform a genetic screening for 62 cardiomyopathy-associated genes in 19 patients with idiopathic RCMP after exclusion of infiltratiove and storage discorders.

Results: in 70% of patients more then 1 pathogenic variant was identified. Among all identified pathogenic substitutions only 27% represented variants in sarcomeric protein genes being the only pathogenic variant only in 21% of patients. Pathogenic variants in cytoplasmic and intrasarcomeric cytoskeletal protein genes represented 43%, the rest 30% of the variants were detected in membrane-associated cytoskeletal and matrix-interacting proteins.

Conclusions: using new generation sequencing we identified more then one pathogenic variant in most of patients with RCMP. Most of the variants were identified not in sarcomeric, but in cytoskeletal and matrix-associated protein genes.