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Spectrum of mutations in hypertrophic cardiomyopathy main genes among Tunisian patients

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Hypertrophic Cardiomyopathy (HCM) is a common genetic cardiac disorder, caused by mutations in genes encoding for sarcomere proteins and transmitted in an autosomal dominant form. Data about the mutational spectrum in HCM patients from North Africa is limited. We performed semiconductor ship (Ion Torrent PGM) next generation sequencing of the main sarcomeric genes (*MYH7, MYBPC3, TNNT2, TNNI3, ACTC1, TNNC1, MYL2, MYL3, TPM1)* in 45 Tunisian HCM patients. Overall, we found a total of 14 carriers (31%) with *MYH7*and *MYBPC3* presenting 75% of the mutations. A patient was homozygous for a new *MYL3* mutation and two patients were double mutation carriers (*MYBPC3+MYH7*). In conclusion, we did report the mutational spectrum of the main genes in Tunisian HCM patients and like studied other ethnic groups, mutations in *MYBPC3* and *MYH7* are the most frequent.

Biography

Nawel Jaafar has received her BSc degree in Medical Biotechnology and MSc degree in Genetics and Biodiversity in the High Institute of Biotechnology, Monastir, Tunisia. Currently, she is a PhD student in Biological Sciences and Biotechnology. She achieved several projects in Molecular Biology and Genetics fields.

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