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Multidisciplinary Approach To Reveal Young Athlete Syncope

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Laboratory medicine, along with genetic investigations in sports medicine, is taking on an increasingly important role in monitoring athletes' health conditions. Acute or intense exercise can result in metabolic imbalances, muscle injuries or reveal cardiovascular disorders. This study aimed to monitor the health status of a basketball player with an integrated approach, including biochemical and genetic investigations and advanced imaging techniques, to shed light on the causes of recurrent syncope he experienced during exercise. Biochemical analyses showed that the athlete had abnormal iron, ferritin and bilirubin levels. Coronary Computed Tomographic Angiography highlighted the presence of an intra-myocardial bridge, suggesting this may be the cause of the observed syncopes. The athlete was excluded from competitive activity. In order to understand if this cardiac malformation could be caused by an inherited genetic condition, both array-CGH and whole exome sequencing were performed. Array-CGH showed two intronic deletions involving MACROD2 and COMMD10 genes, which could be related to a congenital heart defect; whole exome sequencing highlighted the genotype compatible with Gilbert syndrome. However, no clear pathogenic mutations related to the patient's cardiological phenotype were detected, even after applying machine learning methods. This case report highlights the importance and the need to provide exhaustive personalized diagnostic work up for the athletes in order to cover the cause of their malaise and for safeguarding their health. This multidisciplinary approach can be useful to create ad personam training and treatments, thus avoiding the appearance of diseases and injuries which, if underestimated, can become irreversible disorders and sometimes can result in the death of the athlete.

Biography

Dr. Brancaccio Mariarita. During her bachelor's degree in Health Biotechnology, she took part in study on the enhancement of new pulsed UV laser technologies for the development of immunosensors (March 2012) at the Physics department of the University of Naples "Federico II", in the laboratory of prof. Carlo Altucci.

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