New recommendations published in the European Journal of Preventive Cardiology highlight the use of gene testing to prevent sudden cardiac death in athletes and enable safe exercise.

The new guidelines focus on the athletes who should be tested and when they should be tested. It is also recommended that the athletes who undergo genetic testing should be informed in advance of the potential outcome since the results could mean exclusion or restricted play.

In athletes where genetic testing does not indicate high risk, participation in competitive sports can continue after informed discussion with their physicians. But those at higher risk should be restricted to moderate-intensity exercise. The exercise prescription for these athletes should be specific. It should clearly outline the type of exercises or sports that are safe, the intensity at which they remain safe, and the duration and frequency at which the athlete can engage in those activities.

There might be some cases where genetic testing may have to dictate management. For example, in athletes with long QT syndrome (LQTS), it is important to identify and prevent triggers to minimise the risk of arrhythmias. Hence, these athletes should receive targeted medical therapy and exercise advice.

Genetic testing may result in complete exclusion from play in athletes with a heart muscle condition called arrhythmogenic cardiomyopathy (ARVC). This is true even for athletes that may have no clinical evidence of the disease but have the gene for the condition. They must abstain from high-intensity sports.

The recommendations also talk about pre-test genetic counselling to discuss the implications of the test and prospective results on the athletes and their families. Those with a family history may need to undergo clinical monitoring and genetic testing to know their risk. Similarly, post-test counselling must also be performed to discuss and manage the psychosocial, financial and mental health implications if the athlete is excluded from play.

The same is true for child athletes. Genetic counselling in an expert paediatric center and with assistance from a child mental health specialist can help the child understand the impact of the genetic test result. Children who receive a diagnosis of an inherited condition can benefit from genetic testing as it can help predict the risk of sudden cardiac death during sports.

Source: ESC
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