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REVIEW

Diagnosis and Prevention of Long QT Syndrome in Sports

Diagnóstico y Prevención del el Síndrome de QT Largo en el Ámbito Deportivo

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ABSTRACT

The relationship between sports and cardiovascular health, highlighting that, although exercise offered significant benefits, it could also trigger serious events in individuals with hereditary heart conditions, such as Long QT Syndrome (LQTS). This genetic channelopathy affected ventricular repolarization, increasing the risk of arrhythmias and sudden cardiac death, especially in athletes. LQTS was caused by mutations in specific genes and could manifest as syncope, palpitations or sudden death during physical exertion. Diagnosis required a detailed electrocardiographic evaluation, complicated by physiological changes induced by training. The use of genetic testing and the importance of family screening were also mentioned. Finally, it was highlighted that international guidelines allowed for individualized assessment to determine sports eligibility, promoting personalized medicine that protected the lives of athletes.

Keywords: Long QT Syndrome (LQTS); Arrhythmia; Diagnosis; Genetics; Athletes.

RESUMEN

La relación entre la práctica deportiva y la salud cardiovascular, destacando que, aunque el ejercicio ofrecía beneficios significativos, también podía desencadenar eventos graves en individuos con condiciones cardíacas hereditarias, como el Síndrome de QT Largo (SQTL). Esta canalopatía genética afectaba la repolarización ventricular, aumentando el riesgo de arritmias y muerte súbita cardíaca, especialmente en atletas. El SQTL se debía a mutaciones en genes específicos y podía manifestarse con síncope, palpitaciones o muerte súbita durante el esfuerzo físico. El diagnóstico requirió una evaluación electrocardiográfica detallada, complicada por los cambios fisiológicos inducidos por el entrenamiento. También se mencionó el uso de pruebas genéticas y la importancia del cribado familiar. Finalmente, se destacó que las guías internacionales permitieron una evaluación individualizada para determinar la elegibilidad deportiva, promoviendo una medicina personalizada que protegiera la vida de los atletas.

Palabras clave: Síndrome de QT Largo (SQTL); Arritmia; Diagnóstico; Genética; Deportistas.

INTRODUCTION

Physical exercise is indisputably associated with multiple cardiovascular health benefits. However, in some instances, it can trigger serious adverse events, especially in individuals with underlying heart conditions. One of the phenomena that has raised the most concern in this context is sudden cardiac death (SCD) in athletes, particularly those with hereditary channelopathies such as Long QT Syndrome (LQTS). This genetic condition, which alters the ventricular repolarization of the heart, poses significant diagnostic and therapeutic

challenges, especially in high-performance sports. Throughout this analysis, we will explore the main clinical, diagnostic, and preventive aspects of LQTS to contribute to a more accurate understanding of its impact on athletes' health.

DEVELOPMENT

Sudden cardiac death (SCD) in athletes has been a widely studied phenomenon in recent decades, particularly because of its implications for public health and high-performance sports. The literature establishes that regular physical activity has beneficial effects on the cardiovascular system, such as improving lipid, glucose, and blood pressure levels and stabilizing the heart's electrical activity.[1] However, specific individuals, especially those with hereditary channelopathies such as Long QT Syndrome (LQTS), may be at increased risk of fatal cardiac events during exercise.[2]

LQTS is part of a group of channelopathies that alter cardiac ion channels, interfering with normal electrical conduction and predisposing individuals to the development of malignant arrhythmias, such as torsades de pointes.[3,4] This condition is due to genetic mutations in genes such as KCNQ1, KCNH2, and SCN5A, which affect the duration of ventricular repolarization.[5,6] The prevalence of SQTL in the general population is estimated to be between 1:2000 and 1:2500,[7] although this frequency may be higher in elite athletes.[8]

From a clinical perspective, SQTL can manifest as syncope, palpitations, or even sudden death, especially during times of physical or emotional stress.[9,10] Electrocardiographic evaluation, an exceptionally accurate measurement of the corrected QT interval (QTc), is essential for diagnosis, although it may be influenced by bradycardia and physiological adaptation to training in athletes.[11,12] Methods such as the Bazett formula or QT/RR diagrams have been proposed to improve diagnostic accuracy.[13,14]

In sports practice, distinguishing between exercise-induced physiological cardiac remodeling and structural or channelopathic heart disease is a diagnostic challenge.[15,16] For example, endurance sports tend to induce eccentric remodeling, while strength exercises favor concentric hypertrophy, which can alter electrocardiographic parameters.[17,18,19,20]

Given the hereditary nature of SQTL, genetic testing is essential in diagnosis and risk stratification. However, its use should be guided by strict clinical criteria, given the possibility of finding genetic variants of uncertain significance.[21,22] Leading international associations, such as the Heart Rhythm Society (HRS) and the European Heart Rhythm Association (EHRA), recommend cascade genetic screening of family members in confirmed cases.[23,24,25,26,27]

Recommendations regarding sports eligibility have evolved.[28,29,30] The most recent guidelines from both the European Society of Cardiology (ESC) and the American Heart Association (AHA) propose individualized assessment, allowing asymptomatic athletes with a negative phenotype to participate under strict medical supervision and precautionary measures.[31,32,33]

This body of theory establishes the basis for understanding the challenges in the early detection of LQTS in athletes and the need for accurate diagnostic methodologies and individualized protocols to allow safe participation in competitive activities, in line with the principle of personalized medicine.

CONCLUSIONS

Long QT syndrome represents an important risk factor for sudden cardiac death in athletes, especially in physically demanding contexts. Due to the overlap between physiological adaptations to training and pathological signs, the complexity of its diagnosis requires a comprehensive clinical approach that includes detailed electrocardiographic evaluation and, in some instances, genetic testing. Individualization in medical decision-making, supported by updated international guidelines, allows us to move toward more personalized and safer medicine, where the protection of life is balanced with the right of athletes to participate in sports. This approach is essential to prevent tragedies and promote responsible, evidence-based sports practice.

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CONFLICT OF INTEREST

None.

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