



# Sudden Cardiac Death in Adolescent Athletes and the Role of Genetic Screening : A Literature Review

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**Abstract:** Sudden death, particularly among seemingly healthy individuals, presents a critical challenge in both medical and athletic communities. This literature review investigates sudden cardiac death (SCD) in adolescent athletes, emphasizing the role of genetic screening. Utilizing various studies from 2019 to 2024, the review highlights that SCD, while rare, often stems from cardiovascular issues such as cardiomyopathy and coronary artery anomalies. Pre-participation screening, including ECGs, is crucial in identifying at-risk individuals, yet raises ethical debates regarding athlete disqualification. Genetic factors play a significant role, with inherited heart conditions frequently implicated in SCD cases. Comprehensive genetic screening and family evaluations are vital, although widespread implementation is hindered by limited positive outcomes and potential psychosocial effects. Epidemiological studies reveal a higher incidence of SCD during competitive sports, underscoring the need for tailored prevention strategies. Findings emphasize the importance of multidisciplinary collaboration in enhancing screening methods, risk assessment, and preventive interventions to mitigate sudden mortality in young athletes. The study concludes that lifestyle modifications, promoted by health organizations, and advancements in genetic diagnosis are essential in improving the health and safety of athletes. Efforts to integrate cardiovascular and genetic screening into clinical and athletic practice are paramount, aiming to reduce the incidence of SCD and enhance the well-being of young athletes globally.

**Keywords:** Adolescent Athletes; Genetic Screening; Sudden cardiac death

## Introduction

Life in this world cannot be far from death, death is the state of nature in human life (Nel, 2021). A person is declared dead if the function of the heart, circulatory and respiratory systems is proven to have stopped permanently, or if brain stem death can be proven (Gardiner et al., 2020; Joffe et al., 2021; Meier, 2022). Death can occur slowly according to the nature of the disease, but it can also occur suddenly (Abdin et al., 2021; López & Rodó, 2020; Yan et al., 2020). Sudden death is one of the most common cases and can be found in all kinds of conditions (Sadowska et al., 2020). An often unexpected and sudden death can happen to someone who previously appeared healthy (John, 2004; Sessa et al., 2021). These deaths can occur within 24

hours after symptoms appear, but in forensic cases, most deaths occur within minutes or even seconds of the first symptoms appearing (Simão et al., 2022; Tabian et al., 2021). Sudden death, also known as "unexpected natural death," refers to death that is not preceded by significant symptoms. This can be caused by illness or something else. In recent years, there have been many news stories that provide information about sudden death without prior diagnosis (Baigent et al., 2022; Kumar et al., 2021; Tomasoni et al., 2020). Sudden death can be caused by several things, one of the causes of sudden death is a heart attack (Chahal et al., 2020). Although the causes vary, today the cause of sudden death is mostly due to cardiovascular diseases (Franklin et al., 2020). Coronary heart disease causes 80% of sudden cardiac deaths, followed by cardiomyopathy (Lucena, 2019). The heart

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is a vital organ in the human body, located in the chest cavity and divided into four chambers: two atria (right and left) at the top, and two ventricles (right and left) at the bottom (Buijtendijk et al., 2020; Christoffels & Jensen, 2020). Despite the importance of the heart, this organ is prone to various diseases that can attack it. Death from heart disease is the leading cause of all sudden death from the disease throughout the world (Roth et al., 2020; Zimmerman et al., 2020).

Heart disease topped the list of diseases that cause death, followed by infectious diseases and cancer (Murray & Lopez, 1997; Smith et al., 2012). Every year, about 41 million people die from Non-Communicable Diseases (NCD), which accounts for about 74% of the total death cases (Bhattacharya et al., 2023; Mohebi et al., 2018). The majority, about 77%, of NCD deaths occur in countries with low and middle economies (Wang & Wang, 2020). Of these, cardiovascular disease is the highest cause of death, with an estimated 17.9 million cases annually, followed by cancer with an estimated 9.3 million cases, chronic respiratory diseases with 4.1 million cases, and diabetes with 2 million cases of death each year (Ling et al., 2020; Mukasheva et al., 2022; Zhou et al., 2022).

Research shows significant fluctuations in sudden cardiac death rates in young people due to the inclusion of different age groups in the study population (Couper et al., 2020; Ha et al., 2020). The overall risk of sudden cardiac death in individuals under 30 years of age is estimated to be about 1-2.8 per 100,000 (Salzillo et al., 2024). Heart disease sufferers in Indonesia in 2014 amounted to 61,682 people and is expected to continue to grow (Zuraidah et al., 2023). Sudden death often raises questions, showing the importance of paying attention to the condition of the victim before dying, whether he was doing activities or resting after activities, and the environment where the incident occurred. Several factors, such as age, race, personal medical history, and family history, are believed to contribute to the increase in sudden deaths. Common causes of sudden death include disorders of the cardiovascular, respiratory, central nervous, digestive, and genitourinary systems, with cardiovascular disorders as the main cause (Anurupa et al., 2021). Sudden death can also occur in all age groups, including in young people, such as adolescent athletes (Angelini et al., 2020). Increased physical activity in children and adolescents, especially in competitive sports (Chaeroni et al., 2023; Gusril et al., 2022, 2024; Haris et al., 2023).

Based on the above data, primary and secondary prevention strategies are needed. Aims to reduce the incidence of morbidity, mortality and reduce the incidence rate. Recurrence of heart disease. Where this heart attack is an urgent emergency so it requires proper

and fast treatment that is useful so that heart damage is not too severe. Therefore, it is necessary to make lifestyle modifications that are widely promoted by the government, health organizations and a number of elements of society whose function is to improve the health status and quality of life of people with cardiovascular disease. Based on this background, researchers need to examine sudden death from heart disease in adolescent athletes and the role of genetic screening, where the purpose of this literature review is to examine sudden death in adolescent athletes caused by heart disease and how genetic screening plays a role in these events.

## Method

This research uses literature studies. Namely related to the study of theories and other references related to values, cultures and norms that develop in the social situation under study (Sugiyono, 2018). The search for data sources is limited, where the search restrictions for reviewed articles are not only limited through themes, but also limit the publication year of the article. The inclusion criteria in this study include the year of publication between 2019 and 2024, the languages used are English and Indonesian, and the subjects of the study are adolescent athletes. Based on searches that have been done on Google Scholar and other sources with the type of free fulltext research articles – open access or original articles / research using keywords applied in writing this article, researchers found 209 articles that have alignment with the keywords given, from the articles found so that the next stage of screening is applied, 102 journal articles were executed because there were no full text articles available, Then 97 of them were executed and did not fit the inclusion criteria, the result was 10 full text journal articles which were then reviewed in this article.

## Result and Discussion

Several studies have shown lower rates of death and incidence due to all-cause cardiovascular disease, cancer and metabolic conditions in individuals who engage in regular exercise. Nonetheless, sudden cardiac death (SCD) can occur in seemingly healthy individuals, including athletes. Sudden cardiac death is defined as unexpected death from heart disease that occurs within an hour (or within 24 hours). There are 8 articles that will be further analyzed in this literature review. Journal articles are grouped based on the scope of discussion so that articles are obtained that discuss sudden cardiac death in adolescent athletes and the role of genetic screening. For more details can be seen in the Table 1.

**Table 1.** Summary of data descriptions

Author	Topic	Objective	Method	Results	Conclusion
(Finocchiaro et al., 2023)	Sudden Cardiac Death Among Adolescents in the United Kingdom	Using a sizable cohort of teenagers, the authors aimed to look into the causes of SCD and how physical activity relates to them. SCD among athletes is uncommon, however it can be identified at any stage of life.	7,675 instances of SCD were consecutively referred to our national cardiac pathology center between 1994 and June 2022; of them, 756 (10%) were cases involving adolescents. A comprehensive autopsy examination was performed on each subject by skilled cardiac pathologists. Coroners who were referred provided clinical data.	Some athletes exhibit high systolic blood pressure, increased pulse wave velocity, and higher left ventricular mass. Higher pulse wave velocity relates to higher systolic blood pressure and hemoglobin. Increased left ventricular mass links to lower resting heart rate, higher metabolic equivalents, certain exercise disciplines, and high blood pressure.	Teenage SCD victims most often show myocardial disorders and sudden arrhythmic death syndrome at autopsy. Young athletes are more prone to arrhythmogenic cardiomyopathy, coronary artery abnormalities, and commotio cordis compared to sedentary peers.
(Kelly, 2023)	Sudden cardiac death in the young: A consensus statement on recommended practices for cardiac examination by pathologists from the Society for Cardiovascular Pathology	The committee members conducted a literature study and developed consensus guidelines for optimal procedures in the areas of medicolegal, autopsy, and cardiovascular investigations pertaining to sudden death in young individuals.	The SCVP members were asked to join the consensus committee. Individual requests were sent to SCVP members who worked as forensic pathologists or medical examiners. This multi-institutional group determined the critical significance autopsy examination plays in pediatric SCD patients.	This serious international public health issue causes around 15-20% of all deaths. SCD is more frequent in elderly adults with acquired cardiac disease, although it can also develop in young people, where the etiology is more likely to be a genetically transmitted process.	Quality investigations of SCD in the young are hampered by a number of issues, such as the absence of uniform autopsy procedures across jurisdictions and insufficient funding for ME/C offices, which causes limited resources to be directed toward cases more likely to result in criminal prosecution.
(Hansen et al., 2024)	Toxicology Screening in Sports-Related Sudden Cardiac Death	The purpose of this study was to characterize postmortem toxicological results in a global population of young SrSCD patients.	Patients from Denmark, Spain, and Australia who had experienced sudden cardiac death (SCD) and had a full post mortem were included. They ranged in age from 12 to 49. The postmortem results for SrSCD and non-SrSCD were compared, and the toxicological results for SrSCD were evaluated.	Of the 3,189 SCD cases, 219 (7%) had a sporting connection. Patients with SrSCD were younger (36 vs. 41 years; $P < 0.001$ ), predominantly male (96% vs. 75%; $P < 0.001$ ), and had a higher incidence of structural cardiac disease (68% vs. 61%; $P = 0.038$ ). SrSCD cases were less likely to have positive toxicology screens (12% vs. 43%; $P < 0.001$ ).	Men with structural heart disease who were younger were more likely to have sports-related SCD. Compared with non-SrSCD patients, they had a much lower rate of positive toxicology screening results, and there were rarely any medicines linked to SCD found in their system.

Author	Topic	Objective	Method	Results	Conclusion
(Hansen et al., 2024)	Value of screening for the risk of sudden cardiac death in young competitive athletes	The purpose of this study was to present the long-term results of the cardiovascular preparticipation screening (PPS) program in young, competitive athletes conducted in Italy.	In a large group of Italian children (age range: 7-18 years), the study evaluated the long-term effects of PPS, the costs of serial examinations, and the diagnostic yield for disorders at risk of sudden cardiac death (SCD).	During an 11-year study, 22,324 children (62% males; mean age 12) underwent 65,397 annual screenings. Cardiovascular disorders at risk of SCD were found in 69 children (0.3%). Disorders included congenital heart disease, channelopathies, cardiomyopathies, and ventricular arrhythmias. The cost per diagnostic was 73,312 euros.	Through the PPS program, cardiovascular diseases at risk of sickle cell disease (SCD) were identified in children over the entire age range of the research, and more frequently during follow-up evaluations. During long-term follow-up, there was little incidence of sport-related cardiac arrest among children who had been screened.
(Sarto et al., 2023)	Sudden Cardiac Death in Athletes: Facts and Fallacies	to learn about the prevalence, contributing factors, and causes of athlete sudden cardiac death or athlete aborted sudden death.	To find every research that met the qualifying requirements, a thorough search was carried out using the online search engine PubMed.	Exercise benefits cardiovascular and general health, but even healthy athletes can die from sudden cardiac death (SCD). Atherosclerotic coronary artery disease is common in athletes, while primary cardiomyopathies and ion channelopathies affect younger individuals. Health policies, including cardiac screening and public CPR education, are needed to prevent SCD in athletes.	A growing number of young athletes die unexpectedly with structurally normal hearts, suggesting primary arrhythmia syndromes. Cardiac screening for athletes is debated, with concerns about its long-term viability, cost-effectiveness, and the ethical implications of disqualification.
(Stafford, 2022)	The role of genetic testing in diagnosis And care of inherited cardiac conditions In a specialised multidisciplinary clinic	Analyze how genomics is used to diagnose and treat patients in a series of patients who visit a specialized clinic, and determine which patients are most likely to have a monogenic condition.	A retrospective audit of 1697 probands between 2002 and 2020 determined clinical-genetic concordance. Cases with young onset, severe phenotype, and family history were likely monogenic; others had probable complex causes. Impact of genetic diagnosis was evaluated.	Of 888 probands meeting criteria, genetic testing found LP/P alterations in 37%, VUS in 8%, and research initiatives uncovered overlooked variants in 5%. Variant discovery improved diagnosis in 13%, enhancing quality of life. VUS yield varied among tribes, highest in features of monogenic illness.	Research initiatives enhance diagnostic yield by 5%. Clinically useful variants are found in 13% of cases, aiding family screening. Genetics crucial for comprehensive diagnosis, highlighting ancestry-based differences. Methodology ranks probable monogenic cases, considering phenotype knowledge limitations.
(Damayanti et al., 2024)	The characteristic of sudden death at young age	to characterize the incidence of sudden death at a young age.	The research method used in this research is literature review.	Results from 8 journals reveal sudden death's age variability. In the 25-35 age range, men experience higher incidence than women.	Based on the results of 10 journals, it shows that sudden death can occur at any age. However, at a young age, the most common incidence is

Author	Topic	Objective	Method	Results	Conclusion
(Reza et al., 2024)	The expansion of genetic testing in cardiovascular medicine: Preparing the cardiology community for the changing landscape.	Pathogenic DNA variations contribute to various cardiovascular disease phenotypes, including family dyslipidemias, cardiomyopathies, arrhythmias, and aortopathies. Genetic testing is vital for precise diagnosis, distinguishing monogenic forms from diseases with complex etiologies.	The literature review strategy was employed in this study.	Cardiovascular issues predominate as the main cause across studies. Precision diagnosis allows for effective treatment, proper use of new targeted drugs, and risk-monitoring of family members.	found in the productive age category, namely 25-35 years. In this review, we highlight the specific cardiovascular disease phenotypes for which genetic testing should be investigated, as well as the possible benefits of genetic testing and how physicians might incorporate guideline-directed genetic testing into their practice.

An in-depth analysis of a number of scientific studies on sudden death in young athletes illustrates the complexity and diversity of factors involved in this phenomenon. Although these occurrences are rare, the impact on individuals, families, and the sports community is significant. Through a multidisciplinary approach to clinical research, genetics, and public policy, efforts to prevent and treat sudden death are growing. In this discussion, we will explore the main findings of these studies as well as their implications in a practical and ethical context. Epidemiological studies such as those conducted by (Han et al., 2023) highlight that although sudden death in young athletes is a rare event, the risk remains significant. The study showed that the incidence of sudden death in athletes ranged from 1:40,000 to 1:250,000, with variability depending on factors such as research method and type of sport. Although sudden death is rare outside the context of sport, particularly in young athletes, these events can have far-reaching impacts on the athlete population and the sports community as a whole. Research related to the etiology of sudden death in young athletes, such as that conducted by (Fan et al., 2022), highlights that most cases of sudden death in young athletes are caused by cardiovascular problems, with cardiomyopathy and coronary artery anomalies being the main causes. The cause of sudden death is often related to genetic and hereditary factors, which underscores the importance of clinical and genetic evaluation in the treatment of this condition. However, research also shows that there is variation in the causes of sudden death between

countries and athlete populations, emphasizing the need for locally-tailored approaches in the prevention and treatment of sudden death.

The importance of pre-participation screening in detecting potentially dangerous heart conditions in young athletes is highlighted in studies such as those conducted by (Pelliccia, 2021). Screening using an electrocardiogram (ECG) has been recommended by several organizations, including the European Society of Cardiology (ESC), with the aim of detecting heart disease that might increase the risk of sudden death. However, there is an ethical debate surrounding the disqualification of athletes found to have potentially dangerous conditions, with risks of medical discrimination and psychological repercussions to consider. Research on the role of genetics in heart disease, such as that conducted by (Stafford et al., 2022), highlights the importance of family evaluation in efforts to prevent sudden death. Proper post-mortem examination and identification of risk factors can help determine appropriate preventive measures for surviving family members. However, widespread implementation of genetic screening is still constrained by the low number of significant positive outcomes and potential psychosocial consequences for the individuals tested. These findings emphasize the importance of a holistic approach in the prevention and treatment of sudden death in young athletes. Cardiovascular and genetic screening, as well as family evaluation, should be integrated into clinical practice and exercise policy. However, challenges such as ethical debates around

athlete disqualification and limitations in screening results remain issues that need to be addressed. In pursuit of a better understanding of sudden death in young athletes, cross-disciplinary collaboration between researchers, health practitioners, and policymakers is needed. Only through concerted efforts to improve appropriate screening, risk assessment, and preventive interventions can we reduce sudden mortality and improve the safety and well-being of young athletes around the world.

## Conclusion

Athletes in the youth age category are an effective and important age period at the sports coaching level. Both non-competitive and competitive sports have an increased incidence of sports-related SCD with increasing age compared to recreational training; intensive training of competitive athletes appears to increase the risk of sickle cell disease (SCD). Young people who do not participate in rigorous exercise have a prevalence of SCD three times greater than competitive athletes. Sudden cardiac death (SCD) in an athlete is a rare but highly visible tragedy and a source of media attention and discussion among medical personnel, the sports community, and lay people. Heart attack is an urgent emergency so it requires proper and fast treatment that is useful so that heart damage is not too severe. Therefore, it is necessary to make lifestyle modifications that are widely promoted by the government, health organizations and a number of elements of society whose function is to improve the health status and quality of life of people with cardiovascular disease.

A genetic diagnosis can be given by a family member. Recent studies have shown that up to 50% of families of SADS victims are affected by an inherited heart condition (usually channelopathy) that can be associated with SCD in the proband. The condition is usually inherited in an autosomal dominant manner, meaning that the closest relative has a 50% chance of inheriting the same condition. Screening for athletes currently recommended by the European Society of Cardiology and professional sports organizations worldwide, genetic diagnosis results in changes in risk management and stratification for the proband and/or their families. Both non-competitive and competitive sports have an increased incidence of sports-related SCD with increasing age. More than 80% of SCDs occur during competitive or intense exercise. These causes can be further categorized into structural and non-structural pathologies. However, sudden death at a young age, although rare, is a tragic and often unexpected event,

especially if it is a life-threatening condition that can have a major impact on the family.

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## Author Contributions

This article was written by six contributors, Ami Febby Triani contributed to the writing of the introduction, methodology, literature review, results, and conclusions. Gusril, Arsil, Ronni Yenes contributed to the process of conceptualization, methodology, review, and finalization of articles. Adri Budiwaanto contributed to the finalization and improvement of the content of the article. Yovhandra Ockta contributed to reviews in different thought sections.

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