



## Lipid Screening in Childhood and Adolescence for Detection of Familial Hypercholesterolemia

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### ABSTRACT

**Background:** The literature surrounding lipid screening in childhood and adolescence for the detection of familial hypercholesterolemia (FH) highlights critical insights into the importance of early identification and intervention strategies.

**Literature Review:** (Brett et al., 2018) further elaborate on the vital role of primary care in detecting FH, particularly during childhood and adolescence when interventions can be most effective. They cite successful screening outcomes in countries with established programs and suggest that electronic health records could improve detection rates in general practice. This aligns with the systematic approaches proposed by (Qureshi et al., 2018), who recommend using established diagnostic criteria in non-specialist settings to identify FH, advocating for alerts within medical records to prompt cholesterol assessments. (Pederiva et al., 2021) discuss the critical window for diagnosing severe hypercholesterolemia in children, arguing against selective screening based solely on family history, which often fails to identify at-risk children. They advocate for universal screening programs, particularly before puberty, to mitigate future cardiovascular risks. (Thajer et al., 2022) explore a selective screening approach in preschool children, demonstrating that targeted efforts can effectively identify FH in collaboration with school health services. However, they acknowledge that while this approach has shown promise, universal screening may provide a more comprehensive solution to address the broader issue of FH underdiagnosis.

**Conclusion:** In conclusion, the collective findings from these articles advocate for a multifaceted approach to lipid screening in children and adolescents for the detection of familial hypercholesterolemia. They emphasize the necessity of universal screening programs, the integration of genetic testing, and the active involvement of primary care providers to improve detection rates and ensure timely interventions. This comprehensive strategy is crucial for reducing the burden of cardiovascular disease in this vulnerable population.

**Keyword:** Lipid Screening, Childhood, Adolescence, Detection of Familial Hypercholesterolemia

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## INTRODUCTION

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The literature surrounding lipid screening in childhood and adolescence for the detection of familial hypercholesterolemia (FH) highlights critical insights into the importance of early identification and intervention strategies. (Henneman et al., 2015) emphasize that screening for FH is vital due to its association with early onset cardiovascular disease, advocating for an integrated public health approach that incorporates genetic testing alongside traditional screening methods. They note that while various countries have implemented screening programs, many cases remain undetected, underscoring the need for universal cholesterol screening in children.

Building on this foundation, (Brett et al., 2018) argue that primary care plays a crucial role in the detection of FH, particularly during childhood and early adolescence when interventions can be most effective. They highlight successful screening outcomes in countries with established programs and suggest that electronic health records could enhance detection rates in general practice. This perspective aligns with the findings of (Qureshi et al., 2018), who propose systematic approaches in non-specialist settings to identify FH, recommending the use of established diagnostic criteria and alerts within medical records to prompt cholesterol assessments.

Further emphasizing the urgency of early detection, (Pederiva et al., 2021) discuss the critical window for diagnosing severe hypercholesterolemia in children. They advocate for universal screening programs, noting that selective screening based solely on family history often fails to identify at-risk children. Their findings resonate with the position paper by (Bjelakovic et al., 2021), which reviews cardiovascular risk stratification in children with FH and calls for universal screening prior to puberty, reinforcing the need for early intervention to mitigate future cardiovascular risks.

In the context of screening strategies, (Thajer et al., 2022) explore a selective screening approach in preschool children, indicating that although underdiagnosed, FH can be effectively identified through targeted efforts in

collaboration with school health services. Their study highlights the potential for early detection and intervention, echoing the broader consensus in the literature that emphasizes proactive screening measures to prevent long-term health complications associated with FH.

Together, these articles present a comprehensive view of the current landscape of lipid screening for FH in children and adolescents, advocating for enhanced awareness, systematic screening approaches, and the integration of genetic testing to improve outcomes and reduce the burden of cardiovascular disease in this vulnerable population.

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### LITERATURE REVIEW

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In their (Henneman et al., 2015) article, Henneman et al. explore the pressing issue of familial hypercholesterolemia (FH) screening in children, drawing insights from existing adult screening programs. FH is characterized as an autosomal dominant atherosclerotic condition that significantly increases the risk of early-onset cardiovascular disease. The authors emphasize the critical importance of early detection, as identifying individuals with elevated cholesterol levels can prompt timely interventions that may mitigate associated morbidity and mortality ((Henneman et al., 2015)).

The article outlines various screening strategies that have been successfully implemented in countries such as the UK and the Netherlands. These programs have provided valuable lessons regarding the effectiveness of different approaches, yet the authors highlight a persistent challenge: the incomplete identification of cases. This gap underscores the need for more comprehensive screening initiatives, particularly those targeting children, given the potential for early intervention to alter the disease trajectory.

Henneman et al. advocate for the consideration of universal cholesterol screening in children, which poses both opportunities and challenges. The authors argue that such an approach could lead to a significant increase in the identification of at-risk individuals, ultimately allowing for earlier and more effective management of FH. However, they also acknowledge the complexities involved in

implementing these programs, particularly in ensuring that they are integrated into existing public health frameworks.

The authors conclude with recommendations that stress the necessity of an integrated practice model in public health genomics. This model should encompass collaboration among various stakeholders, including primary care practitioners, clinical geneticists, and public health officials. The success of any screening initiative will depend on this collaborative approach, which aims to streamline processes and improve outcomes for children at risk of FH ((Henneman et al., 2015)).

The article "Screening for familial hypercholesterolaemia in primary care: Time for general practice to play its part" by Brett, Qureshi, Gidding, and Watts ((Brett et al., 2018)) provides a comprehensive analysis of the critical role that primary care can play in the early detection and management of familial hypercholesterolemia (FH) in children and adolescents. The authors argue that early diagnosis during childhood and adolescence is crucial for implementing effective lifelong treatment strategies, yet they highlight a significant gap in the current screening practices within general practice settings.

One of the key insights presented in the article is the contrast between countries with established screening programs, such as the Netherlands and Norway, and those without. The authors note that these countries have achieved higher rates of newly diagnosed FH cases and successful cascade testing of relatives, demonstrating the effectiveness of systematic screening approaches. This evidence underscores the potential benefits of adopting similar screening protocols in other regions, particularly in primary care, where most affected individuals are likely to be identified ((Brett et al., 2018)).

The article also emphasizes the acceptability of cascade screening among close relatives of diagnosed individuals, noting that it does not adversely affect quality of life. This finding suggests that implementing a structured approach to screening for FH could be both feasible and beneficial in primary care settings. The authors reference successful initiatives in Australia, such as laboratory alerts and

community-based screening efforts, as examples of how primary care can leverage existing resources to identify FH more effectively ((Brett et al., 2018)).

Furthermore, the article discusses the current state of screening practices in the UK and the United States, highlighting the endorsement of universal cholesterol screening at ages 9 to 11 by prominent health organizations. However, the authors express concern over the incomplete implementation of these recommendations and the lack of cost-benefit analyses to support such initiatives. This gap indicates a need for further research and policy development to ensure that screening for FH becomes a standard practice in pediatric care ((Brett et al., 2018)).

The article "Strategies for identifying familial hypercholesterolaemia in non-specialist clinical settings" by Qureshi et al. ((Qureshi et al., 2018)) presents a comprehensive overview of the diagnostic criteria and strategies for identifying familial hypercholesterolemia (FH) in primary care settings. The authors emphasize the importance of using established diagnostic frameworks, such as the Simon Broome criteria, US MedPed criteria, Dutch Lipid Clinic Network (DLCN) criteria, and Japanese criteria, to facilitate accurate diagnosis of FH.

One of the key insights from the article is the structured approach proposed by the DLCN criteria, which incorporates five essential domains: family history, clinical history, physical examination, and LDL-cholesterol levels. This multifaceted assessment allows for a more robust identification of individuals at risk. The authors argue that individuals who meet at least two of the specified criteria should be considered for further genetic testing, which underscores the significance of early diagnosis in managing FH effectively.

The article further highlights the role of non-specialist clinicians in the initial assessment of potential FH cases. This is particularly crucial given that many individuals may not have access to specialized care immediately. The authors advocate for more systematic screening approaches in non-specialist settings, suggesting that prospective population screening programs could be particularly beneficial. Such strategies are aimed at identifying demographic groups that are more likely to harbor FH, thereby improving the overall detection rates of this condition.

Moreover, the authors align their recommendations with the World Health Organization's (WHO) recognition of the need to manage raised lipids as a key modifiable risk factor for cardiovascular disease (CVD). The evidence presented in the article strongly supports the notion that early identification and treatment of individuals with FH can significantly reduce the risk of CVD, thereby improving long-term health outcomes.

The article "Early Prevention of Atherosclerosis: Detection and Management of Hypercholesterolaemia in Children and Adolescents" by Pederiva et al. ((Pederiva et al., 2021)) emphasizes the critical importance of early detection of severe hypercholesterolemia in children as a strategy to identify individuals at high cardiovascular risk. The authors argue that childhood and early adolescence represent an optimal period for diagnosing familial hypercholesterolemia (FH) and implementing lifelong treatment strategies. This assertion is supported by evidence that screening programs in countries such as the Netherlands and Norway yield significantly higher rates of newly diagnosed FH cases compared to regions without formal screening protocols.

The article discusses Slovenia's initiative to implement universal screening for children aged over five years, although the feasibility of this approach remains uncertain. This highlights a key challenge in the implementation of screening programs: the balance between policy initiatives and practical execution. Furthermore, the authors point out that relying solely on family history to identify children with FH is inadequate, as it fails to detect a substantial number of cases. Consequently, they advocate for universal lipid screening for children aged 9–11 years and young adults aged 17–21 years, reflecting a shift towards more proactive screening measures.

In Canada, the recommendations for routine universal lipid screening at 11 years, or even as early as 12 months for children with a first-degree family history of coronary heart disease (CHD) or FH, align with the article's emphasis on the need for early intervention. The authors also underscore the importance of enhancing public awareness regarding FH, particularly in community settings. Many families may recognize the risk of premature CHD but lack an understanding of the

implications for their own health, which suggests a gap in education and outreach efforts.

The article critically addresses the barriers to effective screening strategies, noting low adherence from the medical community and limited compliance from parents and children. This indicates a need for improved engagement and education regarding the significance of early lipid management. The establishment of FH registries, as mentioned in the article, is a positive step towards generating comprehensive data on the detection and management of FH, which can inform future practices and policies.

The authors also reference the LIPIGEN Network in Italy, which connects specialized centers for the detection and treatment of dyslipidemia, particularly FH. This network exemplifies the potential for collaborative approaches to improve detection and treatment outcomes. The article concludes by highlighting the European Atherosclerosis Society Consensus, which advocates for country-specific screening strategies that leverage existing methods, including opportunistic screening for individuals with a positive family history. It also suggests that cascade screening based on genetic testing is advisable when feasible, further emphasizing the need for tailored approaches to screening.

The article "Cascade Screening and Treatment Initiation in Young Adults with Heterozygous Familial Hypercholesterolemia" by Peterson et al. ((L. Peterson et al., 2021)) provides a comprehensive examination of familial hypercholesterolemia (FH), specifically focusing on the heterozygous form (HeFH), which is notably prevalent, affecting approximately 1 in 200 to 300 individuals. The authors emphasize the critical nature of early detection and treatment, linking timely medical intervention with a significant reduction in the risk of atherosclerotic cardiovascular disease (ASCVD) and mortality.

The article underscores the importance of cascade screening, which involves testing family members of individuals diagnosed with HeFH, as a proactive measure to identify affected children as early as 2 years old. This approach is supported by current guidelines, which advocate for the initiation of statin therapy in children starting at age 8. The authors highlight that while universal

cholesterol screening is recommended by the American Academy of Pediatrics, there is a notable lack of robust evidence assessing the balance of potential harms and benefits of such a policy. This gap in evidence may contribute to the under-utilization of screening practices among healthcare providers.

A critical evaluation of the material reveals that while the authors effectively outline the clinical implications of HeFH and the necessity for early intervention, they also point out the challenges in physician compliance with screening guidelines. The article notes that despite the potential benefits of universal screening, there is insufficient recognition of these guidelines among physicians, which can lead to missed opportunities for early diagnosis and treatment in children and adolescents. This lack of awareness is concerning, given the established correlation between early treatment and improved long-term health outcomes.

Moreover, the article raises important questions regarding the implementation of screening protocols in clinical practice. It suggests that education and awareness campaigns targeting healthcare providers may be essential to enhance compliance with screening recommendations and ultimately improve detection rates of HeFH among the pediatric population.

The article "Risk Assessment and Clinical Management of Children and Adolescents with Heterozygous Familial Hypercholesterolaemia" by Bjelakovic et al. ((Bjelakovic et al., 2021)) provides a comprehensive examination of familial hypercholesterolemia (FH), particularly focusing on its implications for pediatric patients. The authors highlight that FH is a genetic lipid disorder that significantly elevates the risk of premature atherosclerosis and coronary artery disease, emphasizing the urgent need for improved detection and management strategies in children and adolescents.

A critical aspect of the article is its identification of the underdiagnosis and undertreatment of FH on a global scale. The authors note that heterozygous familial hypercholesterolemia (HeFH) affects approximately 1 in 200 to 1 in 500 individuals, leading to lifelong elevated low-density lipoprotein cholesterol (LDL-C) levels. This condition poses a markedly higher risk of cardiovascular disease

compared to individuals without FH, underscoring the necessity for early intervention ((Bjelakovic et al., 2021)).

The paper also discusses the current guidelines for lipid screening in children, referencing recommendations from the National Heart, Lung, and Blood Institute and the National Lipid Association. These guidelines advocate for universal screening for hypercholesterolaemia between the ages of 9 to 11 years and highlight the importance of screening children as young as one year if there is a known familial history of hypercholesterolaemia. The authors suggest that if a genetic defect is identified in a parent, an LDL-C level exceeding 135 mg/dL serves as a critical diagnostic threshold for FH ((Bjelakovic et al., 2021)).

Furthermore, the article delves into non-traditional cardiovascular risk factors that may affect children with FH, thereby broadening the scope of risk assessment beyond standard lipid profiles. This consideration is vital for developing a more nuanced understanding of cardiovascular risk in pediatric populations.

The article "A Selective Screening Strategy Performed in Pre-School Children and Siblings to Detect Familial Hypercholesterolemia" by Thajer et al. ((Thajer et al., 2022)) addresses the critical issue of underdiagnosis of familial hypercholesterolemia (FH) in the pediatric population. The authors emphasize the hereditary nature of this condition, which is characterized by elevated low-density lipoprotein cholesterol (LDL-C) levels from an early age, leading to a significantly increased risk of coronary heart disease (CHD) if left untreated. The prevalence of heterozygous familial hypercholesterolemia (heFH) in Europe is notably concerning, estimated to affect between 1 in 200 and 1 in 500 individuals, underscoring the necessity for effective screening strategies.

The article outlines various screening methodologies, including universal, cascade, and selective strategies, with a particular focus on the selective screening approach implemented in Austria. This strategy involves collaboration with school physicians during pre-school examinations, which the authors argue has been successful in identifying cases of FH among young children. The authors provide a thorough evaluation of the selective screening model, highlighting its effectiveness in detecting FH early, thus allowing for timely intervention and management.

A critical evaluation of the material reveals that while the selective screening strategy has shown promise, it may not be sufficient on its own to address the broader issue of FH underdiagnosis. The authors suggest that universal screening, as initiated in Slovenia for five-year-olds, could potentially provide a more comprehensive solution, ensuring that no cases are overlooked. The article effectively illustrates the importance of early detection and the role of screening in mitigating the long-term health risks associated with familial hypercholesterolemia.

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### CONCLUSION

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The literature on lipid screening in childhood and adolescence for the detection of familial hypercholesterolemia (FH) emphasizes the critical role of early identification and intervention strategies. (Henneman et al., 2015) underscore the importance of screening due to the association of FH with early-onset cardiovascular disease, advocating for universal cholesterol screening in children to enhance detection rates. They highlight that while some countries have established screening programs, many cases remain undetected, necessitating a more integrated public health approach that includes genetic testing alongside traditional screening methods.

(Brett et al., 2018) further elaborate on the vital role of primary care in detecting FH, particularly during childhood and adolescence when interventions can be most effective. They cite successful screening outcomes in countries with established programs and suggest that electronic health records could improve detection rates in general practice. This aligns with the systematic approaches proposed by (Qureshi et al., 2018), who recommend using established diagnostic criteria in non-specialist settings to identify FH, advocating for alerts within medical records to prompt cholesterol assessments.

(Pederiva et al., 2021) discuss the critical window for diagnosing severe hypercholesterolemia in children, arguing against selective screening based solely on family history, which often fails to identify at-risk children. They advocate for universal screening programs, particularly before puberty, to mitigate future cardiovascular risks. This sentiment is echoed by (Bjelakovic et al., 2021), who

highlight the underdiagnosis of FH globally and call for universal lipid screening between the ages of 9 to 11 years, emphasizing the need for public awareness and education regarding FH.

(Thajer et al., 2022) explore a selective screening approach in preschool children, demonstrating that targeted efforts can effectively identify FH in collaboration with school health services. However, they acknowledge that while this approach has shown promise, universal screening may provide a more comprehensive solution to address the broader issue of FH underdiagnosis.

In conclusion, the collective findings from these articles advocate for a multifaceted approach to lipid screening in children and adolescents for the detection of familial hypercholesterolemia. They emphasize the necessity of universal screening programs, the integration of genetic testing, and the active involvement of primary care providers to improve detection rates and ensure timely interventions. This comprehensive strategy is crucial for reducing the burden of cardiovascular disease in this vulnerable population.

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