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Perspectives on the scientific approach to cystic fibrosis in paediatrics

Perspectivas en el abordaje científico de la fibrosis quística en la etapa pediátrica

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ABSTRACT

Objective: to analyse the perspectives on the scientific approach to cystic fibrosis in the paediatric stage. **Method**: descriptive documentary type, the search covered specialised databases, including 15 articles distributed in PubMed, Scielo, Scopus and Web of Science (Wos). **Results and conclusion**: The detection of the bacterium Segniliparus rugosus in a patient with CF raises questions about the microbiology associated with the disease in this context. This finding, together with the difficulties identified in the identification of this bacterium, underscores the need for further exploration of the microbiota in CF patients in Ecuador.

Descriptors: respiratory tract diseases; cystic fibrosis; lung diseases. (Source, DeCS).

RESUMEN

Objetivo: analizar las perspectivas en el abordaje científico de la fibrosis quística en la etapa pediátrica. **Método**: De tipo descriptivo documental, la búsqueda abarcó bases de datos especializadas, incluyendo 15 articulos distribuidos en PubMed, Scielo, Scopus y Web of Science (Wos). **Resultados y conclusión**: La detección de la bacteria Segniliparus rugosus en un paciente con FQ plantea interrogantes sobre la microbiología asociada a la enfermedad en este contexto. Este hallazgo, junto con las dificultades identificadas en la identificación de esta bacteria, subraya la necesidad de una exploración más profunda de la microbiota en pacientes con FQ en Ecuador.

Descriptores: enfermedades respiratorias; fibrosis quística; enfermedades pulmonares. (Fuente, DeCS).

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Original in brief

INTRODUCTION

In the contemporary medical landscape, there is a promising horizon when exploring current and future perspectives in the scientific approach to cystic fibrosis during the paediatric stage. This intriguing scenario not only reveals significant advances in the understanding of the disease, but also presents challenges that require careful consideration by the medical community. [1 2 3 4]

Recent developments in cystic fibrosis research in children have unravelled previously unknown complexities, opening new doors towards more precise and personalised intervention strategies. However, questions remain that require a multidisciplinary approach to fully unravel the mysteries of this childhood condition. [5 6 7]

This article embarks on an analytical journey, exploring not only recent achievements in the scientific understanding of paediatric cystic fibrosis, but also highlighting the challenges that remain. As the complexities are revealed, opportunities for innovation and collaboration open up, in the hope of significantly improving the quality of life for children affected by this genetic disease. [8 9 10]

We aim to explore perspectives on the scientific approach to cystic fibrosis in paediatrics.

METHOD

A meticulous methodological journey was undertaken to illuminate the perspectives in the scientific approach to cystic fibrosis in the paediatric stage, focusing specifically on the Ecuadorian context. This exploration was based on a descriptive documentary methodology, where the application of the analytical method constituted the compass that guided our research.

The ethical selection of 15 relevant research articles in the Ecuadorian field formed the starting point. These meticulously chosen studies represented an amalgam of existing scientific research on cystic fibrosis in children in this particular geographical context.

Content analysis, as a tool of scrutiny, emerged as the knight of the round table, unravelling meanings, patterns and correlations intrinsic to the selected texts. This rigorous exploration allowed us to extract crucial data and, more crucially, to identify gaps and areas of convergence in the Ecuadorian scientific literature on paediatric cystic fibrosis.

At every step of this methodological odyssey, ethical considerations remained guiding lights. Respect for confidentiality, integrity of information and equity in the representation of results remained at the epicentre of our actions, honouring the intrinsic responsibility of medical research.

As a result of this analytical journey, a solid and contextualised theoretical contribution was formed, providing a new nuance to the perspectives in the scientific approach to cystic fibrosis in the paediatric stage, specifically within the vibrant Ecuadorian research fabric. This amalgam of data, ethics and critical analysis constitutes the legacy of our research, aimed at enriching the scientific landscape and ultimately improving the care of young cystic fibrosis patients in Ecuador.

RESULTS

In this study, a comprehensive analysis of cystic fibrosis (CF) in the Ecuadorian population was conducted, merging epidemiological, genetic and clinical data. For contextualisation, information was collected on the incidence of CF in Ecuador, using as a reference an analysis from 2007 that revealed an incidence of 1 per 11,252 live births. [11 12]

A 16S rRNA gene sequencing analysis was applied to identify the presence of Segniliparus rugosus in an 8-year-old boy with CF. The detection of this bacterium was highlighted, along with the difficulties encountered in its identification, setting an important precedent in the understanding of the microbiology associated with CF in Ecuador. [13]

Continuing with the genetic approach, mutations in the CFTR gene were evaluated in Ecuadorian CF patients. New mutations were identified, including the H609R mutation, with an ancestry analysis revealing notable differences in genetic make-up compared to European populations. [14]

The research also revealed the high incidence of certain mutations in the Ecuadorian population, most notably the prevalence of G85E. The proposal of a mutation panel for initial screening in this population was based on these findings. [15]

Finally, clinical aspects, such as the symptomatic presentation of CF in the studied population, were addressed. Specific clinical combinations that might indicate the presence of CF were highlighted, and the predictive performance of the sweat test as a diagnostic tool was evaluated.

In summary, this comprehensive approach merged epidemiology, genetics and clinical to provide a detailed view of CF in the Ecuadorian population. The results obtained constitute a substantial contribution to scientific knowledge and guide towards more effective diagnostic and treatment strategies in the specific context of Ecuador and, by extension, in similar populations in Latin America.

CONCLUSION

At the conclusion of this comprehensive investigation of cystic fibrosis (CF) in the Ecuadorian population, a fascinating and complex picture is revealed that redefines our understanding of this hereditary disease. The convergence of epidemiological,

genetic and clinical data has shed light on key aspects that transcend conventional boundaries of medical knowledge.

The incidence of CF in Ecuador, aligned with that of other mestizo Latin American nations, has proven to be significant, placing the country in a crucial epidemiological context. Genetic analysis has highlighted the diversity of mutations, some unique to the Ecuadorian population, challenging previous paradigms and suggesting a unique aetiology compared to Caucasian populations.

The detection of the bacterium Segniliparus rugosus in a CF patient raises questions about the microbiology associated with the disease in this context. This finding, together with the difficulties identified in the identification of this bacterium, underscores the need for further exploration of the microbiota in CF patients in Ecuador.

Clinical analysis has revealed specific symptomatic combinations that could serve as early indicators of the presence of CF, offering valuable insights for early diagnosis and therapeutic intervention.

In the genetic context, the identification of new mutations and the proposal of a specific screening panel for the Ecuadorian population pose challenges and opportunities in the implementation of diagnostic strategies and genetic counselling.

In sum, this research has pushed the conventional boundaries of understanding CF in Ecuador, providing a solid platform for future research and clinical strategies. The lessons learned and findings highlighted not only enrich scientific knowledge in the field of CF, but also offer valuable insights for personalised medical care in the Ecuadorian context and, potentially, in populations with similar characteristics in the Latin American region. This scientific journey marks a significant milestone in the understanding and approach to CF, pointing to new horizons for research and clinical care in Ecuador and beyond.

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CONFLICT OF INTEREST

There is no conflict of interest with individuals or institutions involved in the research.

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