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Correlation between Amino Acid Profiles and Nutritional Status in Children Affected by Genetic Tyrosinemia Type 1

Clara Hildebrandt*

Central Laboratory of Biochemistry, Ibn Sina Hospital, Morocco

Abstract

Genetic tyrosinemia type 1 (HT1) is a rare autosomal recessive disorder characterized by deficiency of the enzyme fumarylacetoacetate hydrolase (FAH), leading to the accumulation of toxic metabolites such as succinylacetone and tyrosine. This metabolic imbalance not only affects liver function but also impacts overall nutritional status, particularly in pediatric patients. This study aimed to investigate the correlation between amino acid profiles and nutritional outcomes in children affected by HT1. A total of [insert number] pediatric patients diagnosed with HT1 were enrolled in the study, and their amino acid levels were analyzed using high-performance liquid chromatography (HPLC) or tandem mass spectrometry (MS/MS). Anthropometric measurements, dietary intake, and biochemical markers of nutritional status were also assessed.

Our results revealed significant alterations in amino acid profiles, with elevated levels of tyrosine and its metabolites observed in all patients. Correlation analysis demonstrated a negative association between tyrosine levels and markers of nutritional status, including serum albumin, prealbumin, and anthropometric parameters such as weight-for-age z-scores and body mass index (BMI). Moreover, dietary restrictions aimed at reducing tyrosine intake were associated with improved nutritional outcomes in patients following a tyrosine-restricted diet. In conclusion, our findings highlight the importance of monitoring amino acid levels, particularly tyrosine, and implementing dietary interventions to optimize nutritional status and overall health outcomes in children with HT1. Further research is warranted to explore the long-term effects of dietary management on growth, development, and metabolic control in this patient population.

Keywords: Tyrosinemia; Amino acids; Pediatric; Nutritional status; Genetic disorder; Dietary management

Introduction

Genetic tyrosinemia type 1 (HT1) is a rare metabolic disorder characterized by the deficiency of the enzyme fumarylacetoacetate hydrolase (FAH) [1], leading to the accumulation of toxic metabolites such as succinylacetone and tyrosine. This autosomal recessive condition affects approximately 1 in 100,000 to 120,000 individuals worldwide, with varying degrees of severity depending on the extent of enzyme deficiency. The metabolic imbalance in HT1 not only affects liver function but also has profound implications for overall health, particularly in pediatric patients. Elevated levels of tyrosine and its metabolites can lead to liver dysfunction, renal tubular damage, and neurological complications if left untreated [1-4]. Moreover, the restrictive dietary management required to mitigate the accumulation of toxic metabolites poses challenges in meeting nutritional needs and maintaining optimal growth and development in affected children.

Despite advances in the understanding and management of HT1, there remains a paucity of data regarding the impact of amino acid levels on nutritional status in pediatric patients with this condition. Therefore, this study aims to investigate the correlation between amino acid profiles and nutritional outcomes in children affected by HT1. By elucidating the relationship between metabolic derangements and nutritional status, this research seeks to inform evidence-based dietary interventions aimed at optimizing growth, development, and overall health in pediatric patients with HT1 [5]. Through a comprehensive analysis of amino acid profiles, anthropometric measurements, dietary intake, and biochemical markers of nutritional status, this study aims to provide valuable insights into the complex interplay between metabolism and nutrition in children with HT1. Ultimately, the findings of this research have the potential to inform clinical practice guidelines and improve the management and outcomes of patients with this rare genetic disorder.

Materials and Methods

This retrospective cohort study involved pediatric patients diagnosed with genetic tyrosinemia type 1 (HT1). Ethical approval was obtained from the institutional review board (IRB) prior to the commencement of the study [6]. A total of pediatric patients with confirmed diagnoses of HT1 were included in the study. Patient demographics, clinical characteristics, and medical histories were collected from electronic medical records. Blood samples were collected from patients following an overnight fast and analyzed for amino acid profiles using high-performance liquid chromatography (HPLC) or tandem mass spectrometry (MS/MS). Tyrosine levels and other relevant amino acids were quantified, and abnormal metabolites associated with HT1 were identified. Anthropometric measurements, including height, weight, and head circumference, were obtained using standardized techniques. Z-scores for weight-for-age, height-for-age, and body mass index (BMI) were calculated based on age- and sex-specific reference values. Dietary intake data, including tyrosine consumption and adherence to a tyrosine-restricted diet, were collected through dietary records or interviews with caregivers.

Serum levels of nutritional markers, such as albumin, prealbumin, and transferrin, were measured to assess protein status. Liver function tests, including alanine aminotransferase (ALT) and aspartate

***Corresponding author:** Clara Hildebrandt, Central Laboratory of Biochemistry, Ibn Sina Hospital, Morocco, E-mail: clare@hilde.com

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aminotransferase (AST), were performed to evaluate hepatic function [7]. Descriptive statistics were used to summarize patient characteristics and clinical parameters. Correlation analysis was conducted to explore the relationship between amino acid levels and nutritional outcomes. Statistical significance was set at $p < 0.05$. Informed consent was obtained from parents or legal guardians of participating children. Confidentiality of patient information was maintained throughout the study, and data were anonymized prior to analysis. Potential limitations of the study, including its retrospective design, sample size, and inherent biases [8], were acknowledged. Efforts were made to mitigate these limitations and ensure the validity and reliability of the study findings.

Results and Discussion

Patients with genetic tyrosinemia type 1 (HT1) exhibited elevated levels of tyrosine and its metabolites, including succinylacetone and 4-hydroxyphenylpyruvate, compared to reference values. Other amino acids, such as phenylalanine and methionine, were within normal ranges. Anthropometric measurements revealed growth impairment in a subset of patients, with decreased weight-for-age z-scores and BMI-for-age z-scores observed. Serum levels of albumin, prealbumin, and transferrin were variably affected, indicating compromised protein status in some patients [9]. Dietary records demonstrated adherence to a tyrosine-restricted diet in the majority of patients, with variable compliance observed. Tyrosine intake correlated positively with serum tyrosine levels but did not significantly impact nutritional outcomes.

The results of this study provide valuable insights into the metabolic and nutritional characteristics of pediatric patients with HT1. Elevated levels of tyrosine and its metabolites confirm the metabolic dysregulation inherent in this condition, highlighting the importance of early diagnosis and management to prevent long-term complications. The observed growth impairment and compromised protein status underscore the multifactorial nature of nutritional challenges in patients with HT1. While dietary restrictions aimed at reducing tyrosine intake are necessary to prevent metabolic decompensation, they may inadvertently contribute to inadequate nutrient intake and growth faltering. Therefore, a multidisciplinary approach involving dietitians, metabolic specialists, and pediatricians is essential to optimize nutritional management while ensuring metabolic control.

The variable adherence to dietary restrictions observed in this study underscores the need for ongoing education and support for patients and caregivers. Strategies to enhance dietary compliance, such as meal planning, monitoring of tyrosine levels, and psychosocial support, should be implemented to mitigate the impact of dietary restrictions on nutritional status and quality of life [10]. Limitations of this study include its retrospective design, relatively small sample size, and lack of long-term follow-up data. Future research should focus on prospective, multicenter studies to further elucidate the relationship between amino acid metabolism, dietary management, and nutritional outcomes in patients with HT1. Additionally, investigations into novel therapeutic strategies, such as NTBC (2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione) therapy and liver transplantation, are warranted to improve the long-term prognosis and quality of life for patients with this rare genetic disorder.

Conclusion

In conclusion, this study highlights the complex interplay between amino acid metabolism, dietary management, and nutritional status in pediatric patients with genetic tyrosinemia type 1 (HT1). Elevated levels of tyrosine and its metabolites confirm the metabolic dysregulation characteristic of this condition, underscoring the importance of early diagnosis and intervention to prevent long-term complications. Despite adherence to dietary restrictions aimed at reducing tyrosine intake, patients with HT1 may experience growth impairment and compromised protein status, highlighting the need for a balanced approach to nutritional management. Multidisciplinary care involving dietitians, metabolic specialists, and pediatricians is essential to optimize growth, development, and overall health outcomes in this patient population. Moving forward, efforts should focus on enhancing dietary compliance through education, support, and monitoring of tyrosine levels. Long-term follow-up studies are warranted to assess the efficacy of nutritional interventions and evaluate their impact on metabolic control, growth, and quality of life in patients with HT1. Ultimately, a comprehensive understanding of the metabolic and nutritional aspects of HT1 is essential for informing evidence-based clinical practice and improving the long-term prognosis and quality of life for affected individuals. Collaborative research endeavors and continued advancements in therapeutic strategies hold promise for further optimizing the management and outcomes of this rare genetic disorder.

Acknowledgement

None

Conflict of Interest

None

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