Journal of Cardiology Case Reports

Short Report ISSN: 2631-9934

Approach to the cascade retrospective study of heterozygous familial hypercholesterolemia: A statistical tool to better understand family dependence and the index case

Maria Christiane Valéria Braga Braile-Sternieri, Eliana Migliorini Mustafa, Victor Rodrigues Ribeiro Ferreira, Bethina Canaroli Sbardellini, Sofia Braile Sabino, Giovanni Braile Sternieri, Luiza Braile Verdi`, Idiberto José Zotarelli Filho* and Domingo Marcolino Braile

Domingo Braile Institute of Sao Jose do Rio Preto (SP), Rua Luiz Vaz de Camoes, 3111 - Vila Redentora, 15015-750, Brazil

Introduction

Familial hypercholesterolemia (FH) is an autosomal dominant genetic condition (LDL-C), in which apolipoprotein B or pro-protein is converted to subtilisin/kexin [1]. The diagnosis depends on factors such as family history and clinical presentation, genetic tests and severe elevations in plasma cholesterol levels [2]. Heterozygous HF (HeFH) is more common, occurring in approximately one in 500 people in the world. Homozygous FH, on the other hand, is apparently rare, occurring in approximately one in 1 million individuals [3].

Objective

To analyze by means of the multivariate analysis (Dendogram) the correlation of the HeFH of a family with 14 members by means of a retrospective study in genealogical cascade to elucidate the behavior of this pathology along the family descent.

Methods

The present study adopted the male patient AA as the *Index Case* (reference) of the entire study. Thus, 13 relatives, including his wife (non-FH), were correlated by a new statistical approach (Dendogram) to understand the genetic inheritance of HeFH by the degree of similarity of total cholesterol.

Results

The multivariate analysis allowed the qualitative and quantitative understanding of the family relationship of heterozygous hypercholesterolemia in terms of total cholesterol. The *Index Case* did not genetically influence their daughters and granddaughter, with 0.00% (p> 0.05) (Figure 1).

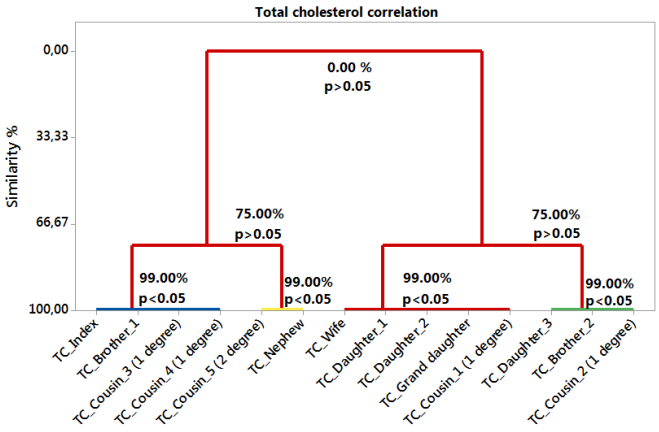

Figure 1. Multivariate analysis of the family relationship of heterozygous hypercholesterolemia in terms of total cholesterol. It was adopted p > 0.05 as statistically different (CI 95.0%).

Conclusion

The multivariate analysis (dendrogram) proved to be an important and more didactic tool to determine the degree of genetic dependence of FH on relatives with the *Index Case*.

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Correspondence to: Idiberto José Zotarelli Filho, Domingo Braile Institute of São José do Rio Preto (SP), Rua Luiz Vaz de Camoes, 3111 - Vila Redentora, 15015-750, Brazil, Tel: 5517-981666732; E-mail: m.zotarelli@gmail.com

Received: June 06, 2019; **Accepted:** June 20, 2019; **Published:** June 25, 2019