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Alpha-1 antitrypsin deficiency - Family study

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Introduction and Purpose: The World Health Organization (WHO) estimates that by 2020, chronic obstructive pulmonary disease (COPD) represents the 5th leading cause of death worldwide.

The Global Initiative for Chronic Obstructive Lung Disease (GOLD) draws attention to the need for systematic research of alpha-1 antitrypsin deficiency in a subset of patients with COPD at a young age or who have significant family history. The alpha-1 antitrypsin deficiency is a hereditary disease caused by a genetic mutation located in the long arm of chromosome 14 and although rarely diagnosed, it is one of the most prevalent genetic disorders.

Case Presentation: Female patient, Portuguese, 42 years old, married, unemployed. History of Chronic Obstructive Pulmonary Disease, with no known family history.

Come to an appointment with her family doctor because of shortness of breat, especially during physical activities, not totally responding to farmacology therapy with Fluticason + Salmeterol, 500+50ug/dose; Salbutamol, 100 ug/dose; Glycopyrronium Bromide + Indacaterol, 43ug+85ug. Physical examination was normal, included a normal pulmonary auscultation, with any signs of respiratory difficulty.

Blood tests to check alpha1-antitrypsin deficiency were asked and on the next appointment the diagnose of alpha-1 antitrypsin deficiency was confirmed. It was explained to the patient that this is a family genetic disorder and that all family members should be investigated.

All her consenting first degree relatives were seen and had a history taken, an examination, measurement of serum a, antitrypsin concentration. All had Alpha antitrypsin concentrations, so as they were oriented to a specific appointment in the hospital to a better genetic orientation and therapy.

Conclusion: In the case of the patient we presented first the diagnosis was performed later in life and in which other factors could be also responsible for clinical manifestations. However in relation to the descendants the study was conducted in a timely manner, even before any symptoms and at a very young age what is good prognostic indicator because either changes in lifestyle, such as smoking cessation, or therapy replacement can be initiated at once and avoid the early onset of the disease.