The Alpha-1 antitrypsin deficiency is a genetic disorder characterized by a deficiency of Alpha-1 antitrypsin, a protein that helps protect the lungs from damage. This deficiency can lead to severe lung disease, as well as liver disease and other complications. The disease is inherited in an autosomal recessive pattern, meaning that an individual must inherit two copies of the defective gene, one from each parent, to develop the disorder.

Similar to the Pan-Italian Antitrypsin Deficiency Registry (PIAD) in Italy, the Mut-Alpha-1 Project was organized in Portugal to study the prevalence of the disease in the country. The study included 2,000 patients with Alpha-1 antitrypsin deficiency from 10 centers in Portugal, and the diagnostic criteria for severe disease were based on the most recent guidelines from the Pan-Italian Antitrypsin Deficiency Registry.

The study found that the prevalence of the disease in Portugal was 1 in 1,000 individuals, which is consistent with previous studies in other European countries. The most common genotype was PI*Z, which is associated with severe lung disease, and the most common severe phenotype was AA TD.

The study also showed that the prevalence of the disease was higher in men than in women, and that the disease was more common in individuals of northern European descent.

In conclusion, the study in Portugal confirmed the prevalence of the disease in the country and provided important information for the management of the disease. Further studies are needed to better understand the genetics of the disease and to improve the care of affected individuals.
filled all criteria for augmentation therapy. However, the phenotype was reported as PI*MS. Due to the inconsistency between the plasma levels and phenotyping, genotyping was performed by SERPINA1 gene sequencing, showing a PI*SMailton genotype.

This was an example of a patient with severe, early-onset emphysema with severe AATD defined by a serum level of AAT < 57 mg/dL, considered as the protective threshold, but with a PI*MS phenotype, consistent with a mild deficiency.

The patient was referred for evaluation for lung transplantation, and in the meantime was considered for augmentation therapy with intravenous AAT.

In conclusion, among the rare deficient variants of AAT, PI*SMailton is probably the most frequent on the Iberian Peninsula, although it still represents a challenge because it is not detected by the first line diagnostic tests (phenotyping/allele-specific genotyping). An PI*SMailton allele-specific genotyping assay has been developed for faster and cheaper diagnosis, but it is not universally available. A larger registry database is needed for a better understanding of the characteristics and natural history of carriers of this rare variant.

Conflicts of interest

Teresa Martin has received speaker fees from Menarini and GlaxoSmithKline. Marc Miravitlles has received speaker fees from AstraZeneca, Boehringer Ingelheim, Chiesi, Cipla, Menarini, Rovi, Bial, Sandoz, Zambon, CSL Behring, Grifols and Novartis, consulting fees from AstraZeneca, Boehringer Ingelheim, Chiesi, GlaxoSmithKline, Bial, Gebro Pharma, Kamada, CSL Behring, Laboratorios Esteve, Ferrer, Mereo Biopharma, Spin Therapeutics, Verona Pharma, TEVA, pH Pharma, Novartis, Sanofi and Grifols and research grants from GlaxoSmithKline and Grifols. Sofia Tello Furtado has received speaker fees from AstraZeneca, Boehringer Ingelheim, Bial, Novartis, Boehringer Ingelheim, and GlaxoSmithKline.

References


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Acute colonic pseudo-obstruction causing Acute Respiratory Failure in Duchenne Muscular Dystrophy

To the Editor,

Duchenne Muscular Dystrophy (DMD) is the most common inherited muscle disease diagnosed in children, with a prevalence ranging between 1.3 and 2.1 per 10,000 live male births. Caused by a mutation of the dystrophin encoding gene located at Xp21, the disease results in a relentless progression of muscle weakness and wasting of the skeletal and cardiac muscle cells. Even though implementing nocturnal and daytime long-term ventilation and cough assistance has reduced the risk of respiratory complications, Acute Respiratory Failure (ARF) is still a common occurrence in DMD patients and a leading cause of death in the very advanced stages of the disease. The pathogenesis of ARF has been attributed to an imbalance between increased respiratory load and reduced diaphragmatic capacity. Well-known aetiologies include pneumonia, otherwise benign