Alpha-1 antitrypsin deficiency study released

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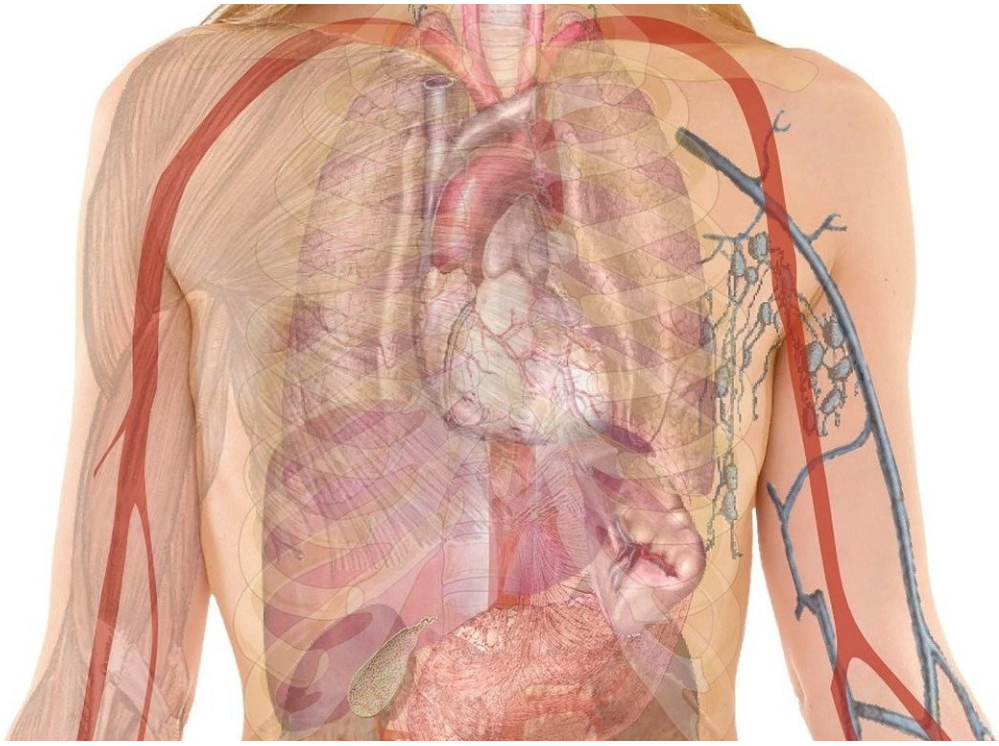

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Alpha-1 antitrypsin deficiency consists of a production deficit of a protein that protects the lungs from degradation or inflammation caused by lung infections, as well as from external agents such a tobacco or pollution. From the liver, Alpha-1 antitrypsin travels to the lungs through the bloodstream. However, if this transfer does not take place properly, its accumulation can cause liver diseases, as well as lung problems such

as emphysema. This disease, according to the study, is under-diagnosed to the point that almost 90 percent of those who have it are unaware.

The goal of the study is to help researchers and medical specialists to better understand this disease, which would facilitate research, prevent cases of erroneous diagnosis and increase treatment efficiency.

This compilation includes a specific chapter on how the disease affects children of paediatric age, as well as relating the latest research on epigenetics and biomarkers, and future therapeutic approaches, including research into gene editing.

Furthermore, the study also highlights the clinical efficiency of replacement therapy, which has been called into question in some countries due to the low number of clinical trials, which makes national health agencies reluctant to pay for replacement therapy. This forces patients to pay for their own medication.

One of the main treatment difficulties that the deficit of Alpha-1 antitrypsin raises, according to the study, is the clinical variability of patients. While some never show any symptoms throughout their lifetime, others develop symptomatology in their childhood, during puberty or when they are young adults.

For this reason, in accordance with the group that carried out the study, there is a need for biomarkers that prognosticate and facilitate the prediction of the disease's progression or its treatment. This will increase understanding of the affected molecular mechanisms.

More information: María Torres-Durán et al. Alpha-1 antitrypsin deficiency: outstanding questions and future directions, *Orphanet Journal of Rare Diseases* (2018). DOI: 10.1186/s13023-018-0856-9