Points to remember for Clinical Practice

Rare Lung Diseases associated with Down Syndrome

- The association of Down syndrome with rare or uncommon lung diseases such as pulmonary haemosiderosis or cystic fibrosis is worth noting.

- Due to an often atypical presentation or lack of knowledge of the existence of these diseases, positive diagnosis is often delayed, meaning that the actual prevalence is underestimated and, most importantly, there is a delay in setting up treatment.

- Though rare, pulmonary haemosiderosis has an incidence that is 75 times higher among children with DS than in the neurotypical population. The diagnosis is suggested by the triad of haemoptysis, iron-deficiency anaemia and presence of alveolar and/or interstitial opacities on lung imaging. In people with DS, the onset is earlier, and outcomes more severe, frequently exacerbated by pulmonary arterial hypertension, and the incidence of recurrence is higher. As the symptoms of alveolar bleeding can be inconspicuous, it is suggested to perform a chest X-ray on all children with DS presenting with chronic anaemia and/or chronic and unexplained dyspnoea.

- Cystic fibrosis should be suggested in the context of current respiratory infections. The gold-standard diagnostic test is the sweat test, which should be used as a first-line approach. Genotyping is required to tailor the treatment.

- Once diagnosed with these diseases, people with DS should benefit from the same treatment opportunities as neurotypical patients.