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FIP. Familial Interstitial Pneumonia

NIHR BioResource – Rare Diseases study project

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Summary

Familial interstitial pneumonia refers to an association of two or more cases of idiopathic interstitial pneumonias in the same family.

Dr Pilar Rivera-Ortega, FIP project Lead

Idiopathic interstitial pneumonias, or IIPs, are conditions associated with lung scarring (pulmonary fibrosis) of an unknown cause. The best known example is idiopathic pulmonary fibrosis (IPF). There are however many others, including: non-specific interstitial pneumonia (NSIP), cryptogenic organizing pneumonia (COP), lymphocytic interstitial pneumonia (LIP) among others. Further research is urgently needed to answer questions regarding their cause, factors influencing prognosis, early diagnosis and new targets for treatment.

IIPs have been reported in closely related family members in 2–20% of cases. Certain genetic mutations are associated with ~20% of all familial interstitial pneumonia cases. Critical shortening in the length of telomeres (fragments of DNA at the end of chromosomes) is also believed to be strongly associated with the development of familial interstitial pneumonia and can influence prognosis (shorter telomeres believed to be linked to a more aggressive disease).

There is a significant unmet need to investigate the genetic determinants of IIPs. The NIHR BioResource project offers a fantastic opportunity to study how genetic targets are linked to the clinical features of IIPs. We hope this project will help to identify family members who are at risk of developing these rare and often more aggressive forms of IIPs in order to provide early diagnosis and more effective treatment.

Recruitment Criteria

Inclusion

Patients who meet one or more of the following 3 criteria are eligible for recruitment:

1. Two or more relatives with interstitial lung disease (ILD) from the same family
2. ILD associated with family history of aplastic anaemia or dyskeratosis congenita
3. Sporadic IIPs that present one of the following features:
    a) Early onset of disease, especially those younger than 60 years of age
    b) Presence of non-specific immunological abnormalities
    c) Presence of non-specific haematological abnormalities
    d) Personal or family history of:
        i. Bone-marrow failure or myelodysplasia
        ii. Cryptogenic cirrhosis
        iii. Premature hair graying (under the age of 25)
        iv. Premature menopause (under the age of 45), non-surgical aetiology
        v. Frequent cancer (keeping in mind that the identification of telomere shortening in sporadic cases may represent the first case of a familial form).