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PCD. Primary Ciliary Dyskinesia

NIHR BioResource – Rare Diseases study project

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V1 26/05/2022

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Summary

PCD is a rare inherited disorder of ciliary function with an estimated incidence of 1:10,000 to 1:20,000. Prevalence varies significantly within populations. This multisystem disease is characterised by progressive sino-pulmonary disease with the development of bronchiectasis, fertility problems and disorders of organ laterality in around 50% of individuals. Abnormal structure of the sperm flagella and the cilia lining the fallopian tubes can lead to infertility.

Clinical presentation can be variable, ranging from mild to severe phenotype. Symptoms typically start in early infancy, with upper and lower respiratory tract infections and the development of bronchiectasis. The diagnosis is based on clinical phenotype and a combination of nasal nitric oxide, high-speed video microscopy to analyse ciliary beat pattern and frequency, and electron microscopy to assess ciliary ultrastructure.

Symptoms include chronic rhinorrhoea, daily wet cough, recurrent sinusitis, recurrent otitis media, bronchiectasis, infertility, disorders of organ laterality, and rarely hydrocephalus.

Ongoing international collaboration aims to find treatments that can directly improve or normalise ciliary function.

Recruitment Criteria

Inclusion

Confirmed diagnosis of Primary Ciliary Dyskinesia.