4.01 Experience of a National Rare Lung Disease Clinic

Andres Clarke[1], Marissa O'Callaghan[1], Evelyn Lynn[1], Lindsay Brown[1], Cormac McCarthy[1]

[1]Department of Respiratory Medicine, St Vincent's University Hospital, Dublin 4, Ireland

Background: Rare lung disease is defined by prevalence of less than 5 per 10,000[1]. Specialty expertise is scarce. Specialised clinics help ensure access to appropriate healthcare for patients with rare diseases.[1] Many rare diseases if diagnosed in a timely manner and managed appropriately are compatible with a normal life.[2]

The aim of this study was to identify the cohort of patients who attend the National Rare Lung Disease clinic and their underlying disease.

Methods: Single centre observational study looking at the patients who attended the Rare Lung Disease clinic from April 2019 to August 2023. Data collected from clinic letters identified age, gender, underlying diagnosis, and those who were under investigation for suspected rare lung disease.

Results: 180 patients attended the clinic from April 2019 to August 2023. 47 were male and 131 were female. 76.4 % of patients had a formal diagnosis. 24.4% of patients attending were under investigation for suspected rare lung disease. Table 1 shows the variety of diagnosed rare lung disease currently attending the National Rare Lung Disease Clinic (NRLDC).

Conclusion: Specialised clinics allow patient access to expertise for diagnosis and management of rare lung disease, and allow for opportunities for identification, international collaboration and research for future avenues for treatment.[3]

Disclosures:

Table 1. Patient cohort attending National Rare Lung disease clinic from April 2019 –August 2023

Diagnosis	**Percentage (%)**
Under investigation	24.4 (N=44)
Birt-Hogg-Dubé syndrome (BHD)	17.8 (N=32)
Lymphangioleiomyomatosis (LAM)	16.1 (N=29)
Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia (DIPNECH)	13.3 (N=24)
Lymphoid interstitial pneumonia (LIP)	6.1 (N=11)
Pulmonary Langerhans cell histiocytosis (PLCH)	4.4 (N=8)
Pulmonary alveolar proteinosis (PAP)	3.9 (N=7)
Tuberous sclerosis (TSC) LAM	2.8 (N=5)
Yellow nail syndrome (YNS)	1.7 (N=3)
Alpha-1 antitrypsin deficiency (A1AT)	1.1 (N=2)
Lymphangiomatosis (GLA)	1.1 (N=2)
Respiratory bronchiolitis–associated interstitial lung disease (RBILD)	1.1 (N=2)
Tuberous sclerosis Multifocal micronodular pneumocyte hyperplasia (TSC MMPH)	1.1 (N=2)
Cryptogenic Organising Pneumonia (COP)	0.6 (N=1)
Catamenial Pneumothorax	0.6 (N=1)
IGG4 Disease	0.6 (N=1)
MEN1 associated nodules	0.6 (N=1)
Nodular Lymphoid Hyperplasia	0.6 (N=1)
Pulmonary Alveolar Microlithiasis (PAM)	0.6 (N=1)
Primary ciliary dyskinesia (PCD)	0.6 (N=1)
Pulmonary Light Chain Deposition Disease (PLCDD)	0.6 (N=1)
Pulmonary Meningoendothelial Nodules	0.6 (N=1)
	100 (N=180)

References:

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