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Knowledge, Research & Advocacy in PH



## PHenomenal Hope 2024

Knowledge, Research & Advocacy in PH

# Through the patient lens: the diagnostic journey from connective tissue disease to pulmonary arterial hypertension

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#### Introduction



- Connective tissue diseases (CTDs) are associated with a risk of developing pulmonary arterial hypertension (PAH); these include:<sup>1</sup>
  - Systemic sclerosis
  - Mixed CTD
  - Systemic lupus erythematosus





**8–12%** of patients with systemic sclerosis will develop PAH<sup>1</sup>



## Challenges to diagnosing PAH-CTD



 Early diagnosis and referral to a PAH specialist is important to improve survival, yet diagnosis is often delayed<sup>1</sup>



 The cause of dyspnea in patients with CTD can be difficult to determine, as it may be due to PAH, ILD, left heart disease, or their underlying autoimmune disease



 Simultaneous diagnosis of two rare diseases relies on coordination of care across a variety of specialist settings



#### Objective

- To gain insight into what support non-PAH specialist providers may need to successfully identify and manage people with PAH-CTD, we sought to understand from the patient perspective:
  - Which providers were involved in each stage of their diagnostic journey
  - What education they received from their provider about PAH risk
  - How patients feel about their diagnoses



 Individuals were invited to participate via Johnson & Johnson's existing Patient Engagement Research Council (PERC) program; eligibility criteria are below



#### **Inclusion criteria**

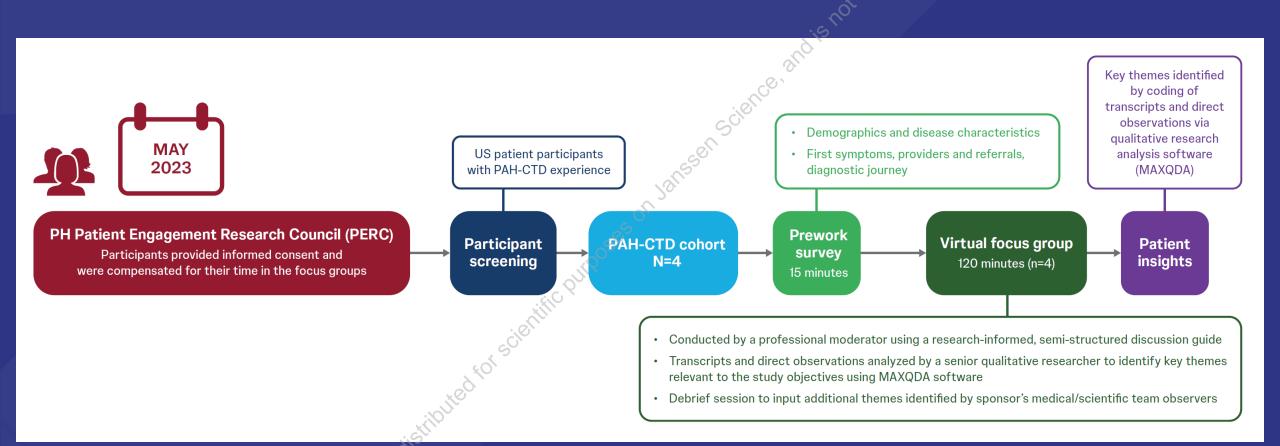
- Diagnosed with PAH-CTD in the past 5 years
- PAH functional class I-III

#### **Exclusion criteria**

- PAH functional class IV
- Diagnosed with sarcoidosis, pulmonary embolism, chronic thromboembolic pulmonary hypertension, interstitial lung disease, idiopathic pulmonary fibrosis, or pulmonary vascular occlusive disease
- Diagnosed with PAH-CTD over 5 years previously



#### Research design





## Focus group participant demographics



4 female participants





3 White

1 Hispanic or Latina





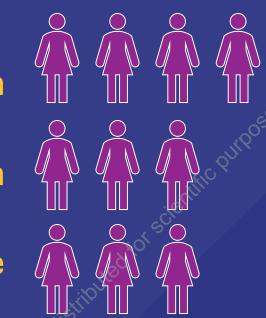
### Diagnostic journeys

**First CTD symptoms** 

Rash

Joint pain

Physical fatigue



Within 6 months of CTD symptoms, participants were referred to:

Rheumatologist (n=3)

Dermatologist (n=1)



Referrat **First CTD** Referral **CTD** PAH PAH PAH diagnosis diagnosis monitoring symptoms symptoms **Female** Aged 45 years<sup>a</sup> Experienced Cardiologist PCP referred to trouble taking diagnosed PAH a rheumatologist deep breaths PCP suspected Rheumatologist in 2021/2022 and a Could not PH and referred diagnosed mixed cardiologist Also diagnosed exercise or to a cardiologist **CTD** in 2021/2022 dermatomyositis. undertake (before mixed Presented to Little information MG, and strenuous CTD diagnosis **PCP** with rash **Ehlers-Danlos** on CTD provided activity without confirmed) History of syndrome in experiencing 2021/2022 seeing multiple dizziness **Suffers from PTSD** providers for due to repeated painful joints, dismissal from pneumonia, <u>=</u> and bronchitis multiple providers **PAH** <8 months <8 months



<sup>&</sup>lt;sup>a</sup>Age at time of the PERC engagement.

CTD, connective tissue disease; MG, myasthenia gravis; PAH, pulmonary arterial hypertension; PCP, primary care provider; PERC, Patient Engagement Research Council; PH, pulmonary hypertension; PTSD, post-traumatic stress disorder.

**First CTD** Referral **CTD** PAH Referral PAH PAH diagnosis diagnosis monitoring symptoms symptoms **Female** Aged 29 years<sup>a</sup> PCP referred to Shortness of Diagnosed with a dermatologist breath when **PAH** in 2018 walking Dermatologist Undergoes RHC Little information Rheumatologist encouraged Difficulty about PAH every couple of years Rheumatologist patient to see a diagnosed breathing when provided Presented to **lupus** in 2016 referred to a rheumatologist lying flat pulmonologist **PCP** with joint X-ray showed Lack of education pain, loss of Also referred to fluid in lungs about PAH caused appetite, cardiologist and later anxiety when feeling drained, nephrologist limitations of butterfly rash disease discovered <8 months **PAH** 2 years

<sup>a</sup>Age at time of the PERC engagement.

CTD, connective tissue disease; PAH, pulmonary arterial hypertension; PCP, primary care provider; PERC, Patient Engagement Research Council; RHC, right heart catheterization.



First CTD symptoms

Referral

CTD diagnosis

PAH symptoms

Referrat

PAH diagnosis

PAH monitoring

Female Aged 61 years<sup>a</sup>



- Presented to PCP with psoriasis in 2013/2014
- History of pulmonary issues

PCP referred to

a dermatologist

 Dermatologist noticed spots on face and fingernails, suspected scleroderma (not followed up)

~2 years

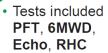
- A second dermatologist diagnosed scleroderma and lupus in 2015
- Detailed education on CTD provided



- Shortness of breath during walks and when climbing stairs
- Coughing
- Poorer running performance



- Referred to a rheumatologist in 2017
- Rheumatologist referred on to a pulmonologist and cardiologist



- Pulmonologist and cardiologist diagnosed PAH, ILD and mixed CTD in 2018
- Detailed education on PAH provided



 Undergoes 6MWD test every 6 months and PFTs every year

PAH diagnosis caused severe anxiety warranting medication



CTL

3 years

rs

PAH

==

6MWD, 6-Minute Walk Distance; CTD, connective tissue disease; Echo, echocardiogram; ILD, interstitial lung disease; PAH, pulmonary arterial hypertension; PFT, pulmonary function test; PCP, primary care provider; PERC, Patient Engagement Research Council; RHC, right heart catheterization.



<sup>&</sup>lt;sup>a</sup>Age at time of the PERC engagement.

Referrat **First CTD** Referral **CTD** PAH PAH **PAH** diagnosis symptoms diagnosis monitoring symptoms **Female** Aged 49 years<sup>a</sup> Rheumatologist Diagnosed with conducted PFT lupus in 2004 Experienced Pulmonologist PCP referred to and Echo in Subsequently shortness of diagnosed PAH a rheumatologist 2009 (results • RHC initially every diagnosed with breath and in 2015 not followed up) 6 months, now less mixed CTD chest pain Presented to Admitted through in 2007 frequently Unable to **PCP** with joint **ER** with chest Little information exercise as pain in 2015 pain, rash, and on CTD provided previously symptoms of · Referred to a **PAH diagnosis** Ravnaud's pulmonologist increased anxiety, phenomenon warranting Family history medication of RA PAH <8 months ~11 years



<sup>&</sup>lt;sup>a</sup>Age at time of the PERC engagement.

CTD, connective tissue disease; Echo, echocardiogram; ER, emergency room; PAH, pulmonary arterial hypertension; PCP, primary care provider; PERC, patient engagement research council; RA, rheumatoid arthritis; RHC, right heart catheterization.

# Participant quotes about their experience with PAH-CTD

#### Feeling dismissed



My relationship with my PCP was never great but felt I didn't have a choice, and I felt dismissed, and [they] attributed everything to my weight. [It was] narrow-minded.
... There was completely a lack of knowledge [about CTD].

– Patient, age 29

#### Feeling validated



66 [My PCP] listened to me and sent me where I needed to go. Before that, I never went to see a doctor. 9 9

– Patient, age 49



# Participant quotes about their experience with PAH-CTD

# Struggling to understand diagnoses

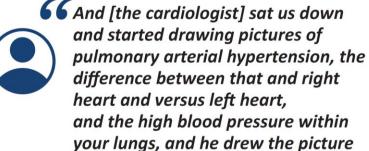


Everything has been a learning process for me because [my pulmonologist] never really described what PH was or what it does. All they told me is, 'Your artery has extra pressure.' No one explained to me what the symptoms are for PH.

- Patient, age 29

# Receiving PAH education

and explained it extremely well.



- Patient, age 61



# Recommendations for providers from people with PAH-CTD



Increase education for providers to improve screening, referrals, and diagnoses of this high-risk population



**Primary care and advanced practice providers** improve overall education around CTD and PAH, including signs and first symptoms important in recognizing CTD



**Rheumatologists:** encourage providers to spend time assessing cardiopulmonary morbidities associated with exercise intolerance in their patients who have CTD and improve education about the risks of developing PAH in CTD



Dermatologists: encourage referral to PAH specialist after diagnosing a patient with a CTD



#### Conclusions



 The convergence of rare, complicated diseases brings a unique set of diagnostic challenges for physicians



People with CTD described marked variability in the care they received, provider knowledge, and time between CTD and PAH diagnoses



 Equipping providers with adequate knowledge to recognize CTD, confidence in evaluating PAH risk will enable timely referral to PAH specialists



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#### Disclosures

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- RC has nothing to disclose
- SG and MH are employees of Actelion Pharmaceuticals US, Inc., a Johnson & Johnson Company, Titusville, NJ, USA



## Thank you!

https://www.janssenscience.com/media/attestation/congresses/pulmonary-hypertension/2024/team-phenomenal-hope/through-the-patient-lens-the-diagnostic-journey-from-connective-tissue-disease-to-pulmonary-arterial.pdf

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