PHenomenal Hope 2024

Knowledge, Research & Advocacy in PH

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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:[1]
  - Systemic sclerosis
  - Mixed CTD
  - Systemic lupus erythematosus

25% of all PAH cases are **PAH-CTD**[2]

8–12% of patients with systemic sclerosis will develop PAH[1]

1. Khangoora V, et al. *Pulm Circ*. 2023;13:e12276. 2. Humbert M, et al. *Eur Respir J*. 2023;61:2200879.

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Challenges to diagnosing PAH-CTD

- Early diagnosis and referral to a PAH specialist is important to improve survival, yet diagnosis is often delayed[1]

- 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

CTD, connective tissue disease; ILD, interstitial lung disease; PAH, pulmonary arterial hypertension.

1. Khangoora V, et al. *Pulm Circ*. 2023;13:e12276.

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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

CTD, connective tissue disease; PAH, pulmonary arterial hypertension.

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Participants

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

Inclusion criteria	Exclusion criteria
• Diagnosed with PAH-CTD in the past 5 years • PAH functional class I-III	• 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

CTD, connective tissue disease; PAH, pulmonary arterial hypertension.

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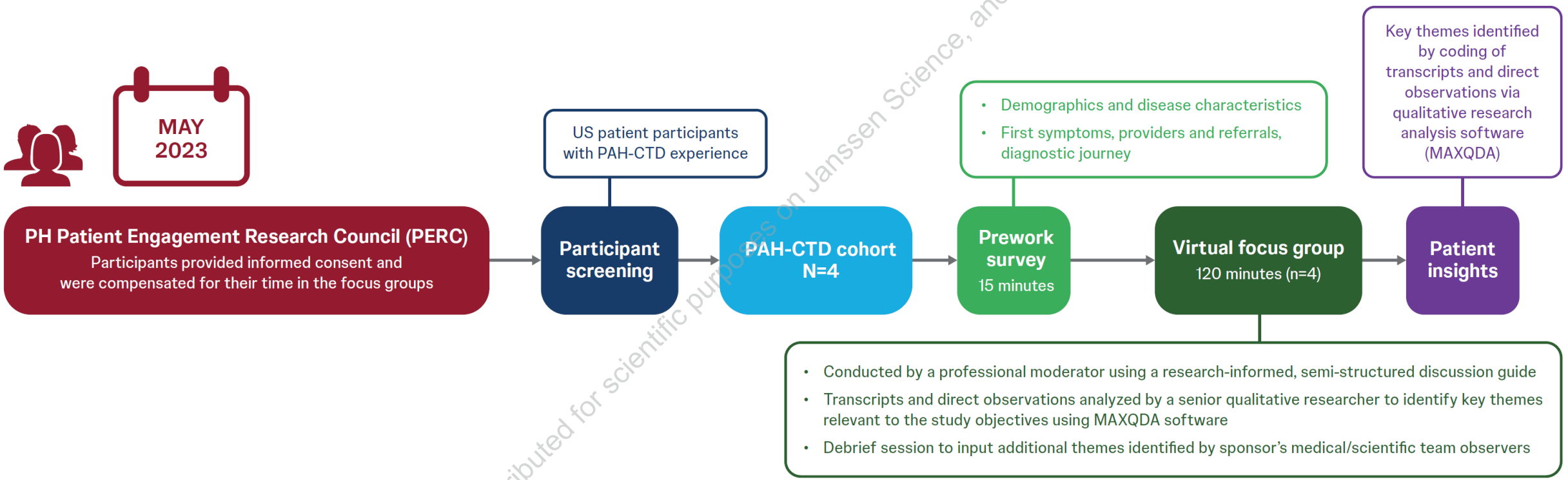
Research design



CTD, connective tissue disease; PAH, pulmonary arterial hypertension; PH, pulmonary hypertension; US, United States.

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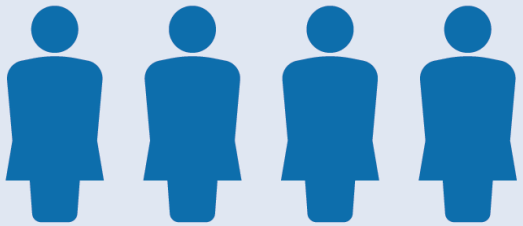
Focus group participant demographics



4 female participants

Age groups

18–34 years	1
34–54 years	2
55–64 years	1

Race/ethnicity

3 White

1 Hispanic or Latina

Education

Postgraduate degree	2
Bachelor's degree	1
Trade school	1

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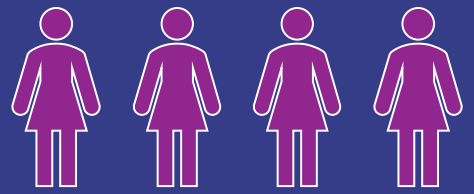
Diagnostic journeys

First CTD symptoms

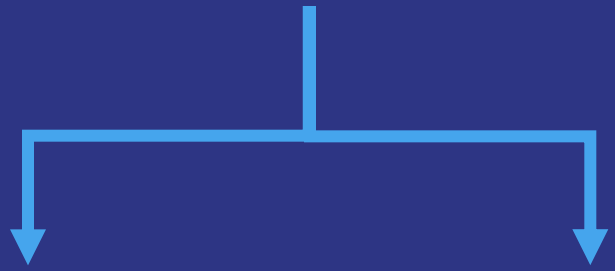
Rash



Joint pain

Physical fatigue

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



Rheumatologist (n=3)

Dermatologist (n=1)

CTD, connective tissue disease.

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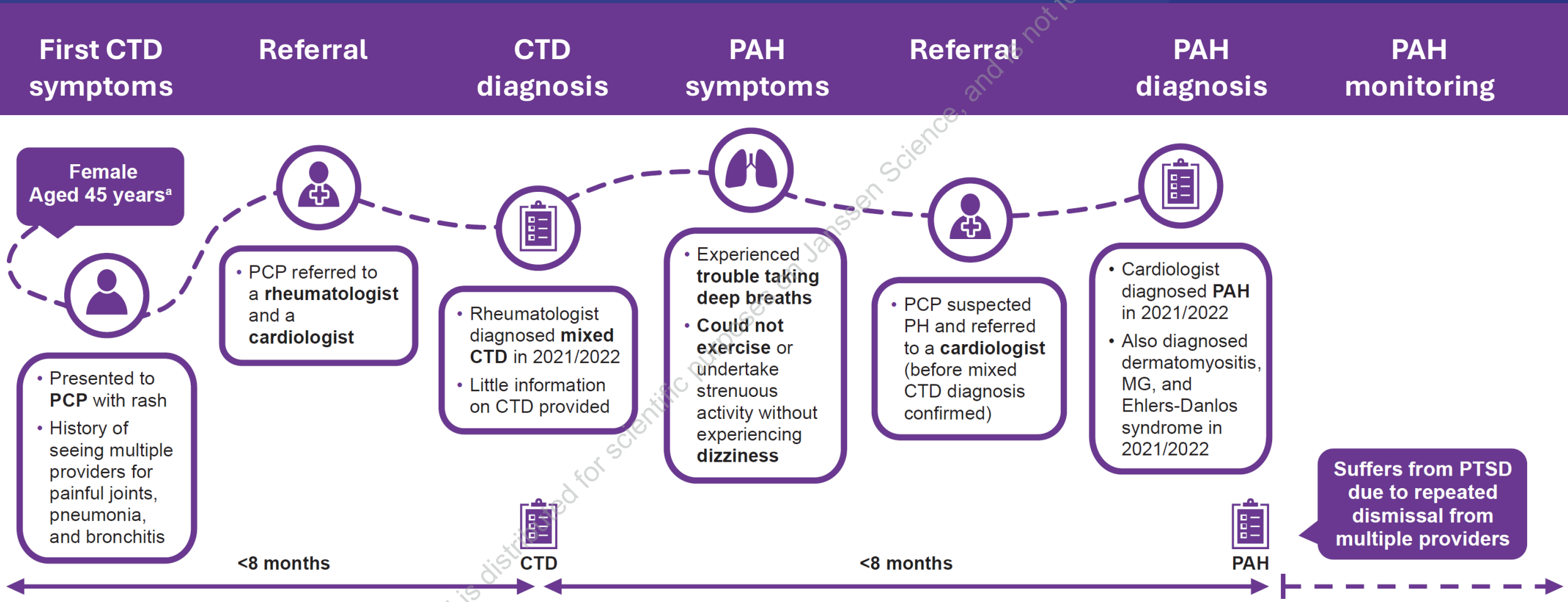
Participant 1



[a]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.

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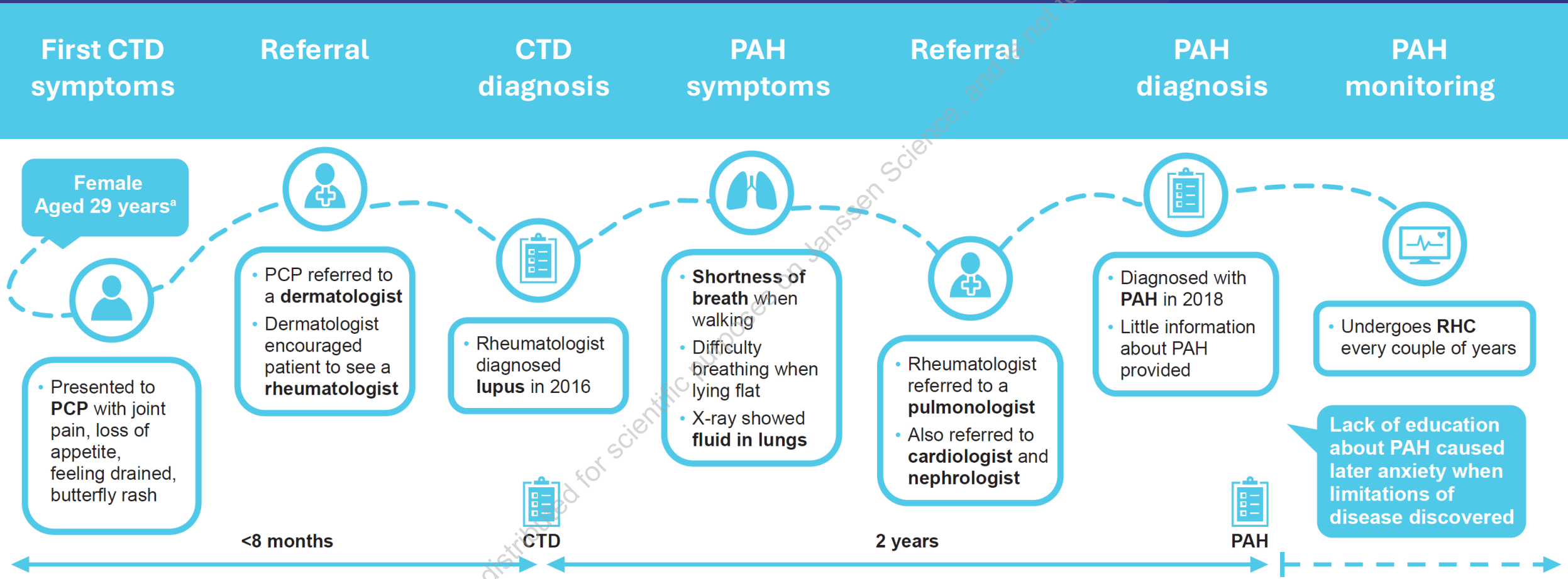
Participant 2



[a]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.

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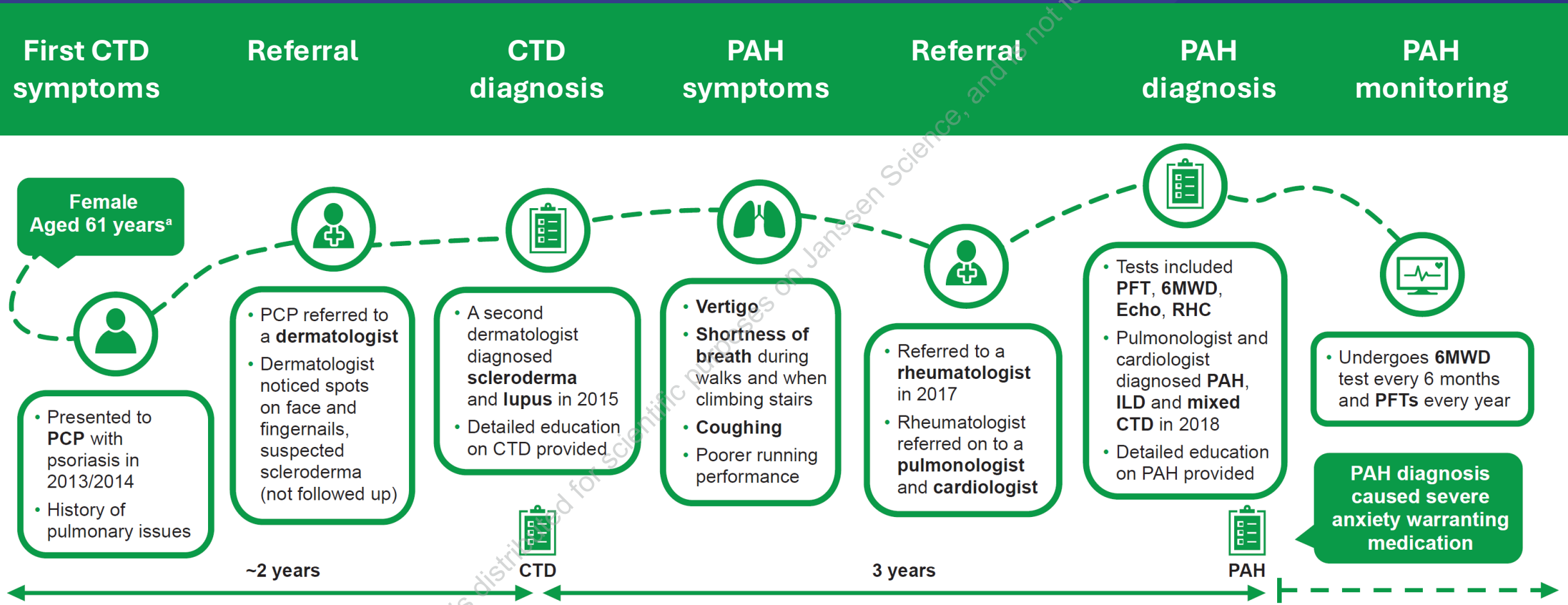
Participant 3



[a]Age at time of the PERC engagement.
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.

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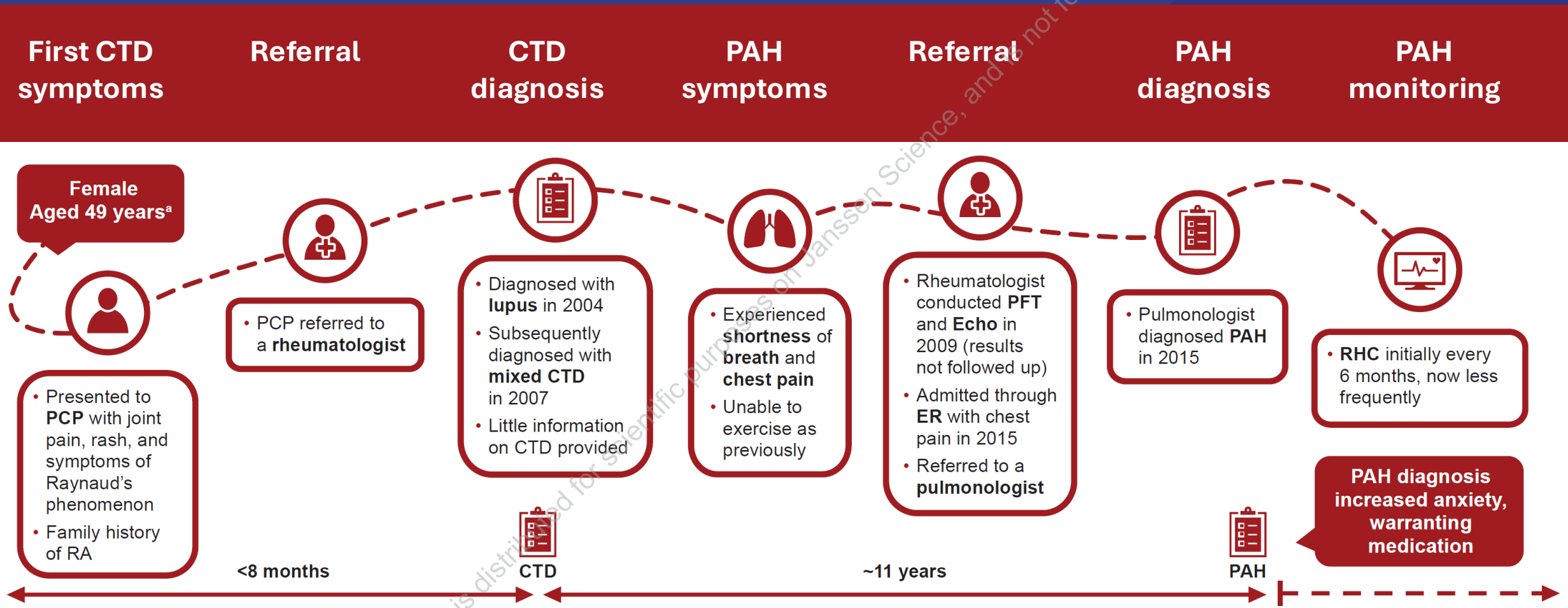
Participant 4



[a]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.

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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

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

– Patient, age 49

CTD, connective tissue disease; PAH, pulmonary arterial hypertension; PCP, primary care provider.

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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 [the cardiologist] sat us down and started drawing pictures of pulmonary arterial hypertension, the difference between that and right heart and versus left heart, and the high blood pressure within your lungs, and he drew the picture and explained it extremely well."

– Patient, age 61

PAH, pulmonary arterial hypertension; PH, pulmonary hypertension.

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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

CTD, connective tissue disease; PAH, pulmonary arterial hypertension.

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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

CTD, connective tissue disease; PAH, pulmonary arterial hypertension.

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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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