Transcript Details
This is a transcript of an educational program. Details about the program and additional media formats for the program are accessible by visiting: https://reachmd.com/programs/clinicians-roundtable/telomere-disorders-in-familial-pulmonary-fibrosis/67683/

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Recognizing Telomere Biology Disorders in Familial Pulmonary Fibrosis

Announcer:
Welcome to *Clinician's Roundtable* on ReachMD. On this episode, we'll hear from Dr. Pilar Rivera Ortega. Not only is she a consultant in respiratory medicine at the Royal Devon University and Exeter Hospital, but she also serves as an Associate Research and Development Director for Royal Devon University Healthcare NHS Foundation Trust in the United Kingdom. She'll be discussing her recent presentation at the 2026 European Respiratory Society Congress that explored when we should suspect a telomere biology disorder in patients with familial pulmonary fibrosis. Let's hear from Dr. Ortega now.

Dr. Ortega:
When I am in my clinic in front of a patient with interstitial lung disease and suddenly the patient is saying that there is another relative in his or her family with interstitial lung disease or with pulmonary fibrosis, that should be the first red flag. So if we have two or more members in the same family—could be a first-degree relative or second-degree relative with interstitial lung disease, and it doesn't necessarily have to be exactly the same type of interstitial lung disease that your patient that you have in front of you has—that should be the first feature to make you think that this patient possibly has a genetic predisposition.

Other features that are more, I would say, tailored or specific to telomere biology disorders are, for example, if your patient appears to have pulmonary fibrosis as well as some hematological abnormalities, particularly myelodysplasias, aplastic anemia, or certain types of hematological malignancies like leukemia and so on. Other organs like the liver could be affected, particularly if we have liver cirrhosis, or if we have early gray hair. We define the cutoff of early if there is some gray hair at age 30 or before that. If we have this combination of factors—they are not the only ones—but if there's liver abnormalities, hematological abnormalities, early gray hair, and pulmonary fibrosis alongside other organs that could be affected, these are the features that make me think that this patient could have a telomere biology disorder.

To diagnose patients or to confirm our clinical suspicion of telomere biology disorder, we have some tests that can be done during our usual consultations or in dedicated clinics for familial pulmonary fibrosis. Genetic testing could be a panel or whole genome sequencing. Most of the time—and there is not a consensus yet about which tests should be chosen in the first instance—in most of the countries, they have a genetic panel that includes telomere-related genes, surfactant-related genes, and sometimes some common variants that are called polymorphisms or SNPs, such as MUC5B and TOLLIP. In parallel, we can have another blood test or part of the same test using a different technique to measure the length of the telomere. And we have different techniques to be able to have a reliable report.

When we have the ILD genetic panel and also the telomere length, we can discuss the information with the clinical aspects, like context and family history. We can discuss all of these reports ideally in a familial pulmonary fibrosis MDT with other disciplines, such as hematologists, hepatologists, immunologists, geneticists, genetic counselors, lung transplant physicians, and others.

Announcer:
That was Dr. Pilar Rivera Ortega sharing her insights on when we should suspect a telomere biology disorder in patients with familial pulmonary fibrosis. To access this and other episodes in our series, visit *Clinician's Roundtable* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!