Questionare: Diagnosis, screening, and follow-up of patients with familial interstitial lung disease: Results from an international survey

q1: Are you currently a pulmonologist working with patients suffering from Interstitial lung diseases?
q1a1: Yes
q1a2: No

q2: Which country are you from?

q3: Where is your working place?(Multiple answers possible)
q3a1: Public Academic Center
q3a2: Public non-academic Center
q3a3: Private Academic Center
q3a4: Private non-academic Center
q3a5: Specialized ILD center
q3a6: Pulmonology department (non-university center)
q3a7: Intensive care unit
q3a8: Other

q4: How many years of experience do you have as a physician?

q5: How many years of experience do you have as a pulmonologist?

q6: How many years of experience do you have as an ILD physician?

q7: How many ILD patients do you have in your practice or center in total?

q8: How many new ILD referrals do you have in your practice or center each year?

q9: What percentage of your ILD patients have a history of familiar interstitial lung disease?

q10: How often (percentage) do you ask newly referred patients about their family history with respect to interstitial lung diseases?

q11: How often (percentage) do you draw a pedigree when asking newly referred patients about their family history with respect to interstitial lung diseases and diseases associated with a risk for developing interstitial lung diseases?

q12: How often (percentage) do you draw a pedigree in patients with a known family history of interstitial lung disease?

q13: How often (percentage) do you ask about a family history of early grey hair, in general in your patients with ILD(Early gray hair: Before the age of 30)?

q14: How often (percentage) do you ask about a family history of early grey hair, if a family history of ILD is present(Early gray hair: Before the age of 30)?

q15. How often (percentage) do you ask about a family history of liver cirrhosis, in general in your patients with ILD?

q16: How often (percentage) do you ask about a family history of liver cirrhosis, if a family history of ILD is present?

q17: How often (percentage) do you ask about a family history of bone marrow disease, in general in your patients with ILD?

q18: How often (percentage) do you ask about a family history of bone marrow disease if a family history of ILD is present?

q19: How often (percentage) do you ask about a family history of lung cancer (specific lung adenocarcinoma) if a family history of ILD is present?

q20: How often (percentage) do you ask about a family history of premature osteoporosis if a family history of ILD is present?

q21: How often (percentage) do you ask about a family history of skin diseases if a family history of ILD is present?

q22: How often (percentage) do you ask about a family history of nail dystrophy if a family history of ILD is present?

q23: How often (percentage) do you ask about a family history of Hermansky-Pudlak Syndrome (characterized by oculocutaneous albinism and bleeding diathesis) if a family history of ILD is present?

q24: How often (percentage) do you ask newly referred patients with an idiopathic ILD < 50 years of age about early grey hair?

q25: How often (percentage) do you ask newly referred patients with an idiopathic ILD < 50 years of age about unexplained liver cirrhosis?

q26: How often (percentage) do you ask newly referred patients with an idiopathic ILD < 50 years of age about bone marrow disease?

q27: Do you have access to genetic testing?
q27a1: yes
q27a2: no

q28: If yes to the question above: When will you refer a patient with a family history for genetic testing?
q28a1: at any age
q28a2: < 40 years of age
q28a3: < 50 years of age
q28a4: < 60 years of age
q28a5: < 70 years of age
q28a6: Age depends on the patients personal or family history of liver cirrhosis or bone marrow disease

q29: Do you have access to genetic ILD multidisciplinary meetings?
q29a1: yes
q29a2 no

q30: If yes, to the question above:Which medical specialties are a part of the ILD multidisciplinary conference?(Multiple answers possible)
q30a1: Pulmonologist

q30a2: Clinical geneticist
q30a3: Genetic counselor
q30a4: Hepatologist
q30a5 Hematologist
q30a6: Other (please specify)

q31: How often (percentage) do you refer ILD patients with ILD < 50 years of age without a family history of an interstitial lung disease for genetic testing?

q32: How often (percentage) do you refer ILD patients with ILD < 60 years of age without a family history of an interstitial lung disease for genetic testing?

q33: In case a pathogenic variant in the telomere-related gene or surfactant protein gene is found in the index patient, do you suggest genetic testing and counselling for first degree relatives?

q33a1: yes
q33a2: No

q34: In case a pathogenic variant in a telomere-related gene or surfactant protein gene is found in a first degree relative do you recommend screening for ILD among their relatives?
q34a1: yes
q34a2 no

q35 In some cases of familial pulmonary fibrosis, a mutation is not detected but the pedigree is indicative of familial pulmonary fibrosis. Do you, in such cases, recommend screening for ILD among their first degree relatives?

q35a1: yes
q35a2: no

q36: At what age do you recommend to start the screening program in a patient with a proven genetic background?

q36a1: 30 years of age
q36a2: 40 years of age
q36a3: 45 years of age
q36a4: 50 years of age
q36a5: 55 years of age
q36a6: 60 years of age
q36a7: 10 years before the index patient was diagnosed (genetic anticipation)
q36a8: Not applicable, I don't have access to genetic screening

q37: If a pathogenic variant in a telomere-related gene is found in a first degree relative, do you recommend screening for liver disease in this individual?
q37a1: Yes
q37a2: No

q38:If a pathogenic variant in a telomere-related gene is found in a first degree relative, do you recommend screening for bone marrow disease in this individual?
q38a1: Yes
q38a2: No

q39: If a pathogenic variant in a telomere-related genes is found in a first degree relative, do you recommend screening for other organ involvement in this individual?
q39a1: Yes
q39a2: No

q40: If yes, to the question above, please specify organs:

q41: Do you have a standardized screening program for a first degree relative with a pathogenic variant?
q41a1: Yes
q41a2: No

q42: Do you perform/recommend a HRCT as a part of the screening program?
q42a1: yes
q42a2: no

q43: Do you perform/recommend pulmonary function test as a part of the screening program?
q43a1: yes
q43a2: no

q44: If yes, to the question above: Do you perform spirometry as part of the screening program?

q44a1: yes
q44a2: no

q45: Do you perform diffusing capacity test of the lungs as part of the screening program?

q45a1: Yes
q45a2: No

q46: Do you perform a somatic evaluation as part of the screening program according to the nature of the mutation/syndrome (cutaneous examination, premature greying of hair, nail dystrophy, oral leukoplakia and abnormal skin pigmentation, signs of liver cirrhosis)?
q46a1: Yes
q46a2: No

q47: Does your local Department of Clinical Genetics use next-generation sequencing (NGS) for genetic testing?
q47a1: yes
q47a2: no
q47a3: Don’t know

q48: Does your local Department of Clinical Genetics use whole-genome sequencing (WES) for genetic testing?
q48a1: yes
q48a2: no
q48a3: Don’t know

q49: Does your local Department of Clinical Genetics measure peripheral blood leukocyte telomere length as part of the genetic testing?
q49a1: yes
q49a2: no
q49a3: Don’t know

q50: Does your Department of Clinical Genetics test for the MUC5B promoter polymorphism?
q50a1: yes
q50a2: no
q50a3: Don’t know

q51: Does your Department of Clinical Genetics test for pathogenic variants in surfactant protein genes?
q51a1: yes
q512: no
q51a3: Don’t know

q52: Does your Department of Clinical Genetics test for pathogenic variants in telomere-related genes?
q52a1: yes
q52a2: no
q52a3: Don’t know

q53: In patients with a family history of interstitial lung disease, going through lung transplant work-up, do you measure peripheral blood leukocyte telomere length before lung transplantation at your center?
q53a1: yes
q53a2: no
q53a3: Don’t know

q54: In patients with a family history of interstitial lung disease is undergoing lung transplant work-up, will you refer patients for genetic testing?
q54a1: yes
q54a2: no
q54a3: Don’t know

q55: Will identification of a telomere-related gene mutation prevent the patient from lung transplantation at your center/collaborating transplant center?
q55a1: yes
q55a2: no
q55a3: sometimes

q56: If telomere length is below the 10th percentile, will this prevent the patient from lung transplantation at your center/collaborating transplant center?
q56a1: yes
q56a2: no
q56a3: sometimes

q57: Will identification of a surfactant protein mutation prevent the patient from lung transplantation at your center/collaborating transplant center?
q57a1: yes

q57a2: no
q57a3: sometimes

q58: Will identification of peripheral blood leukocyte telomere length below the 10th percentile, or a telomere-related gene mutation modify the protocol of treatment after lung transplantation at your center/collaborating transplant center?

q58a1 yes
q58a2: no

q59: Do you treat patients with a pathogenic variant in the telomere-related genes with immunomodulatory drugs?
q59a1: yes
q59a2: no
q59a3: sometimes

q60: Do you treat patients with a pathogenic variant in the telomere-related or surfactant protein genes with antifibrotic therapy?
q60a1: yes
q60a2: no
q60a3: sometimes

q61: If yes, when will you start? (Multiple answers possible)
q61a1: At diagnosis of the genetic background
q61a2: If there are symptoms assigned to pulmonary fibrosis?
q61a3: If the pulmonary function is reduced due to pulmonary fibrosis?
q61a4: If there is sign of fibrosis on (high resolution) computed tomography?
q61a5: Only if sign of progressive fibrosis? (worsening of symptoms/pulmonary function/radiology)?

q62: In which ways do you find genetic testing of ILD patients useful from a clinical perspective?(Multiple answers possible)
q62a1: I do not find it useful from a clinical perspective
q62a2: To determine if familial screening is necessary
q62a3: In determining treatment options
q62a4: As part of lung transplant work-up
q62a5: To predict the course of the disease with higher precision
q62a6: To better evaluate the risk for extra-pulmonary health issues
q62a7: For diagnostic purposes

q63: How often (percentage) do your patients with interstitial lung disease ask for genetic testing?

q64: How often (percentage) do first degree relatives of a patient with interstitial lunge disease ask for genetic testing?

q65: How often (percentage) do first degree relatives of a patient with interstitial lung disease ask for screening in form of a HRCT?

q66: How often (percentage) do first degree relatives of a patient with interstitial lung disease ask for screening in form of pulmonary function testing?

q67: How often (percentage) do patients with interstitial lung disease decline genetic testing when this is suggested by their physician and/or genetics counsellor?

q68: How often (percentage) do first degree relatives of a patient with interstitial lung disease decline genetic testing when this is suggested by their physician and/or genetics counsellor?

q69: If they decline, please specify:

q70: How often (percentage) do you discuss ethical concerns with patients before referral for genetic testing?

q71: How often (percentage) do you discuss ethical concerns with first degree relatives before referral for genetic testing?

q72: Do you have ethical concerns with genetic testing of first degree relative?

q72a1: yes

q72a2: no

q73; if yes, please specify

q74: What is in your opinion the biggest challenge at the moment (multiple answers possible)

q74a1: Lack of evidence-based specific treatment options for familial fibrosis
q74a2; Lack of trials focusing on patients with familial pulmonary fibrosis
q74a3: Lack of guidelines for diagnosis of patients with familial fibrosis
q74a4: Lack of guidelines for screening for familial fibrosis
q74a5: Other (please specify)