Clinical Description: Systemic Sclerosis-associated Interstitial Lung Disease (SSc-ILD) – Incident

Synonyms: Scleroderma Lung Disease; SSc-ILD; New-onset ILD secondary to Systemic Sclerosis.

1. Clinical Case Definition

This phenotype specifically captures the *initial onset* or *first clinical recognition* (incidence) of Interstitial Lung Disease in a patient with either established or concurrently diagnosed Systemic Sclerosis (SSc). SSc-ILD is a manifestation of SSc, an autoimmune connective tissue disease, characterized by inflammation and progressive fibrosis of the pulmonary parenchyma (the interstitium).

The diagnosis requires confirmation of both components:

- **Systemic Sclerosis** is diagnosed based on established clinical features (e.g., skin thickening/sclerodactyly, Raynaud's phenomenon) and supportive autoantibodies (e.g., anti-Scl-70/topoisomerase I, anti-centromere, or anti-RNA polymerase III).
- **Incident Interstitial Lung Disease** is identified by new evidence of pulmonary involvement. This is typically established using High-Resolution Computed Tomography (HRCT) of the chest, demonstrating findings such as ground-glass opacities, reticulation, traction bronchiectasis, or honeycombing, often in patterns consistent with Non-Specific Interstitial Pneumonia (NSIP) or Usual Interstitial Pneumonia (UIP). Pulmonary Function Tests (PFTs) support the diagnosis by revealing a restrictive ventilatory defect (reduced Forced Vital Capacity (FVC)) and/or impaired gas exchange (reduced Diffusion Capacity for Carbon Monoxide (DLCO)).

2. Phenotype Scope & Granularity

- **Temporal Context:** Incident. Intended to identify the new onset or first clinical recognition of ILD in the context of SSc.
- **Severity:** Broad spectrum. Inclusive of all severities present at the time of initial diagnosis, ranging from subclinical radiographic findings to symptomatic and physiologically significant disease.
- **Acuity / Chronicity:** Represents the onset of a chronic pulmonary manifestation (ILD) within the context of an underlying chronic systemic disease (SSc).
- **Etiology:** Autoimmune; specifically secondary to Systemic Sclerosis.
- **Population Context:** Primarily the adult population.

3. Related Conditions & Scope Boundaries

This phenotype specifically requires the identification of new-onset ILD secondary to Systemic Sclerosis.

- **Prevalent SSc-ILD / Prior History of SSc-ILD:** Patients with a pre-existing diagnosis of SSc-ILD before the observation period are not within the scope of this *incident* phenotype.
- **Idiopathic Pulmonary Fibrosis (IPF):** IPF is a distinct form of ILD occurring without an associated systemic autoimmune disease and is not within the scope of this phenotype.
- **Other Connective Tissue Disease-associated ILD (CTD-ILD):** ILD associated with other systemic diseases (e.g., Rheumatoid Arthritis, Myositis, Sjögren's Syndrome) are distinct etiologies and are not within the scope of this SSc-specific phenotype.
- **Exposure-related or Drug-induced ILD:** ILD caused primarily by environmental exposures (e.g., hypersensitivity pneumonitis) or medications are etiologically distinct and not within the scope of SSc-ILD.
- **Systemic Sclerosis without ILD:** Patients with SSc who have not yet developed interstitial lung involvement are not the target population for this phenotype definition.
- **Other Pulmonary Manifestations of SSc:** While SSc can cause Pulmonary Arterial Hypertension (PAH) or aspiration pneumonia, these are distinct pathophysiological processes (vasculopathy or aspiration vs. parenchymal fibrosis) and are not sufficient alone to meet this phenotype definition.

4. Key Complications & Common Comorbidities

These conditions frequently co-occur with SSc-ILD or are complications of SSc, but they are distinct clinical entities from the interstitial lung pathology itself.

- **Pulmonary Hypertension (PH) / Pulmonary Arterial Hypertension (PAH):** A frequent and significant vascular complication/comorbidity.
- **Gastroesophageal Reflux Disease (GERD) and Esophageal Dysmotility:** Highly prevalent in SSc and may exacerbate lung injury via aspiration.
- **Raynaud’s Phenomenon and Digital Ulcers:** Common vascular manifestations of the underlying SSc.
- **Scleroderma Renal Crisis:** A specific renal complication of SSc.
- **Cardiac Involvement:** Including myocarditis, pericardial effusion, or heart failure.

5. Intended Data Sources

This clinical description is intended for building concept sets against real-world administrative claims, electronic health record (EHR) databases, and patient registries. The target data should be standardized to the OMOP Common Data Model. Examples of licensable data sources where this concept set would be applied include:

- **Optum®** (Clinformatics® Data Mart, SES, Pan-Therapeutic)
- **Merative™** (MarketScan® Commercial Claims and Encounters)
- **Veradigm** (Health Verity, Practice Fusion EHR)
- **IQVIA** (AmbEMR, PharMetrics Plus)
- **HealthVerity**