



# SCLERODERMA, THE DISEASE THAT MUMMIFIES



Scleroderma is a little-known and incurable autoimmune disease of as yet unknown cause that involves four main abnormalities: dysregulation of the immune system, inflammation, microvascular damage, and fibrosis. Diagnosed in thousands of Quebecers, scleroderma is characterized by an overproduction of collagen and damage to the blood vessels that causes excessive scarring within the various organs. This imbalance leads to a hardening of the skin (fibrosis) and an alteration of the affected organs. Moreover, in most cases, scleroderma limits the motor skills and dexterity of those affected and causes great physical and psychological suffering since it produces disfiguring appearance-related changes.



## SCLERODERMA IN BRIEF:

- ▶ Of the thousands of Quebecers affected (diagnosed in about 4 out of every 10,000 inhabitants), women are six times more likely than men to be affected by the disease.
- ▶ First symptoms usually appear in the early forties, but can also occur in children and people of all ages.
- ▶ Significant decrease in quality of life and life expectancy.
- ▶ Very difficult to diagnose: symptoms vary greatly from person to person, so patients will not develop all the complications of the disease.
- ▶ The skin, digestive system, heart, lungs, and kidneys are the organs most commonly affected by the potentially serious complications of scleroderma.
- ▶ Current medical treatments are used to limit disease-related complications and improve quality of life and longevity. However, there is not yet a definitive cure.



# HOW SCLERODERMA CAN AFFECT THE HUMAN BODY

The symptoms of scleroderma vary greatly from person to person, so that patients will not necessarily develop all the complications of the disease. The symptoms of the disease may be visible, as is the case when the skin is affected, or the symptoms may be invisible, as when internal organs are affected.

## SYMPTOMS AND MANIFESTATIONS OF SCLERODERMA

### SKIN HARDENING

Thickening and loss of elasticity of the skin on different parts of the body. Hence the name «scleroderma», which means hard skin.

### PULMONARY FIBROSIS

A potentially serious complication where normal lung tissue is gradually replaced by scarred fibrotic tissue, making it difficult to breathe and deliver needed oxygen to the body.

Pulmonary fibrosis causes shortness of breath and also sometimes a dry cough.

### RENAL CRISIS

A renal crisis, which is due to an acute obstruction of arterioles and capillaries in the kidneys, leads to a sudden and sharp increase in arterial blood pressure. The symptoms are those of a hypertensive crisis: new and severe headaches, marked shortness of breath (left heart failure), and even epileptic seizures (convulsions). This is a very serious complication which requires urgent medical attention. Often during a scleroderma renal crisis, the kidneys stop functioning and dialysis (filtering the blood to avoid uremia) is then needed.

### BLOOD VESSELS

The narrowing of the arteries, small blood vessels, and capillaries, can lead to many complications, including the development of pulmonary arterial hypertension (PAH), digital ulcers, and other conditions.

### PULMONARY ARTERIAL HYPERTENSION (PAH)

Increased pressure in the pulmonary arteries due to the narrowing of small arteries in the lungs. Blood flow to the lungs is significantly restricted, making the heart work harder to pump blood through the lungs.

As arterial blood pressure rises in the pulmonary arteries, small pulmonary vessels slowly become clogged (a process which may take several years). This occurs through fibrosis of the small vessels, eventually leading to thrombosis, and the blood can no longer reach all parts of the lungs. Thus, it becomes more difficult for the lungs to supply enough oxygen to the body.

Sustained high blood pressure in the arteries of the lungs puts a strain on the heart, making it more difficult to circulate the blood through the lungs. Over time, this can eventually lead to congestive heart failure, particularly the right side, what is referred to as right heart failure (RHF). Right heart failure is indicative of significant PAH and is a serious complication of scleroderma.

PAH results in one or more of the following symptoms:

- Shortness of breath on exertion and at rest
- Palpitations (heart rhythm disorder)
- Fatigue
- Chest pain • Dizziness
- Temporary loss of consciousness (syncope)
- Swelling of the ankles and legs

### SCLERODERMA FACES

Hollow eyes, pinched nose, thin pursed lips, mask-like face, small puckered mouth (microstomia), and peri-oral folds. Thinning lips and facial muscle atrophy can make the teeth appear more prominent.

### EYES

Dry eyes caused by a decrease in tear production.

### TELANGIECTASIA

Small dilated capillaries visible on the face and hands, sometimes referred to as «spider veins».

### RAYNAUD'S PHENOMENON

Raynaud's is present in up to 95% of people with scleroderma. Whitening of fingers and/or toes triggered by cold or severe stress. The whiteness phase can be followed by a blue phase and then a red phase.

### SCLERODACTYL

The skin of the fingers, which have become infiltrated with collagen (fibrosis), may look full and sausage-like. Functional loss or decreased range of motion.

### CALCINOSIS

Calcium deposits under the skin that may require antibiotics to cure occasional infections and sometimes surgery to drain calcium deposits and relieve pain.

### DIGITAL ULCERS

Ulcers occur on the fingertips or on the top of the fingers. They are painful and difficult to heal. In the most severe cases, it can lead to necrosis and amputation may be needed.

### SKIN PIGMENTATION

Dark or pale spots occurring in one-third of patients.

### DIGESTIVE SYSTEM

Gastrointestinal disorders affect the vast majority of patients. Gastric reflux is a common symptom that manifests itself by a burning sensation radiating up to the throat after meals and may cause inflammation of the lining of the esophagus (esophagitis reflux) if left untreated.

### MUSCLE AND JOINT PAINS

Joint pain is common. It is caused by inflammation of the joints and tendons, which quite often leads to joint swelling and stiffness that can become quite debilitating.

Muscular pain (myalgia) can be intermittent or continuous. It can also be associated with muscle weakness (myositis). Symptoms include difficulty in climbing stairs, lifting objects and getting up, and also difficulty swallowing.