

Possible Symptoms by Organ System

OPHTHALMIC SYMPTOMS

- Macular cherry-red spot
- Corneal clouding
- Nystagmus

PULMONARY SYMPTOMS

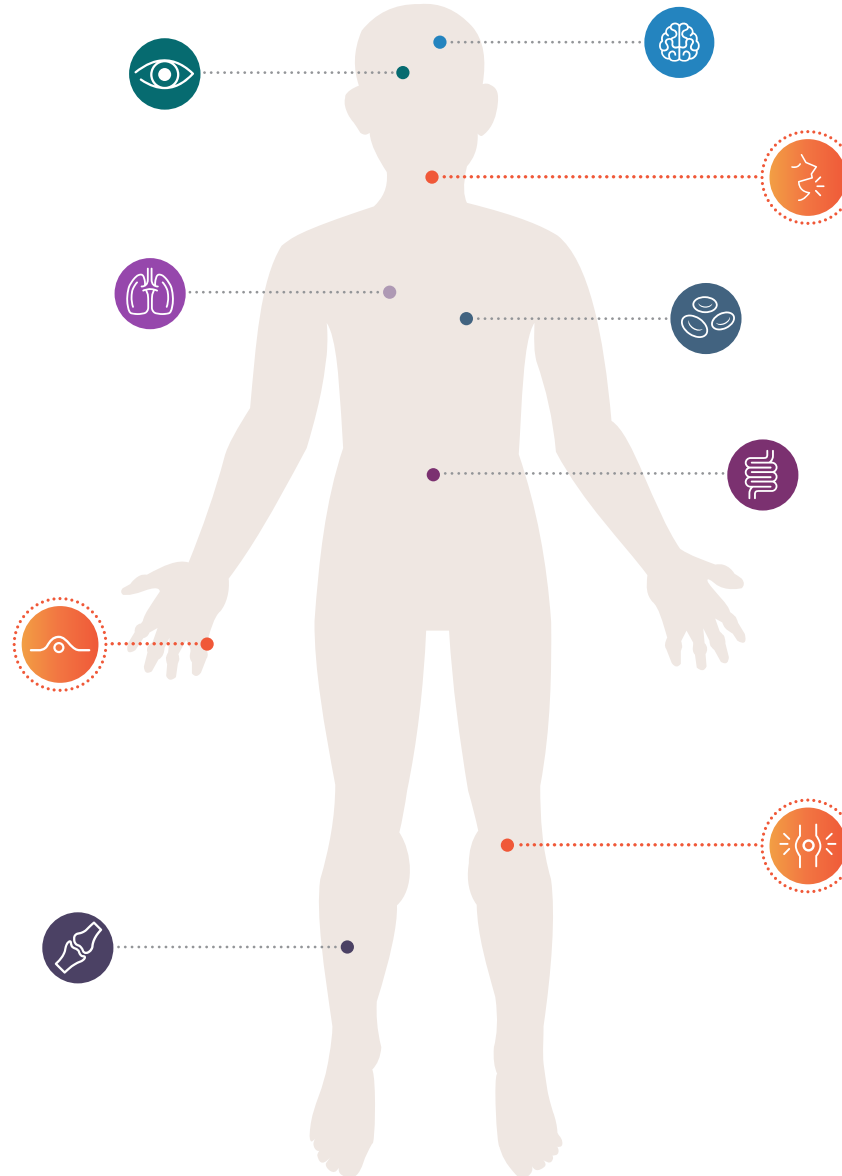
- Respiratory distress
- Aspiration
- Infection and pneumonia
- Lung inflammation and infiltration

CARDINAL SYMPTOM

- Bumps (nodules) under the skin

ORTHOPEDIC SYMPTOMS

- Bone erosion
- Progress osteoporosis
- Peripheral osteolysis



NEUROLOGIC SYMPTOMS

- Developmental delay/cognitive impairment
- Seizures
- Hypotonia and muscle weakness
- Lower motor neuron disease

CARDINAL SYMPTOM

- Hoarse or weak voice (due to nodules in the larynx)

HEMATOLOGIC SYMPTOMS

- Systemic inflammation with or without fever
- Foamy histiocytes and macrophages

GASTROINTESTINAL SYMPTOMS

- Failure to thrive
- Hepato-/splenomegaly
- Jaundice
- Liver Failure

CARDINAL SYMPTOM

- Painful and swollen joints, or joint disease