Refer patients to a geneticist for an enzyme activity analysis if they are/have been[1]:

- Diagnosed with any of these disorders without confirmatory testing
- Currently undergoing evaluation for any of these disorders

- MED
- SED
- Bilateral Legg–Calvé–Perthes disease
- Pseudoachondroplasia
- Undiagnosed skeletal dysplasia

Rule out or confirm Morquio A with an enzyme activity analysis

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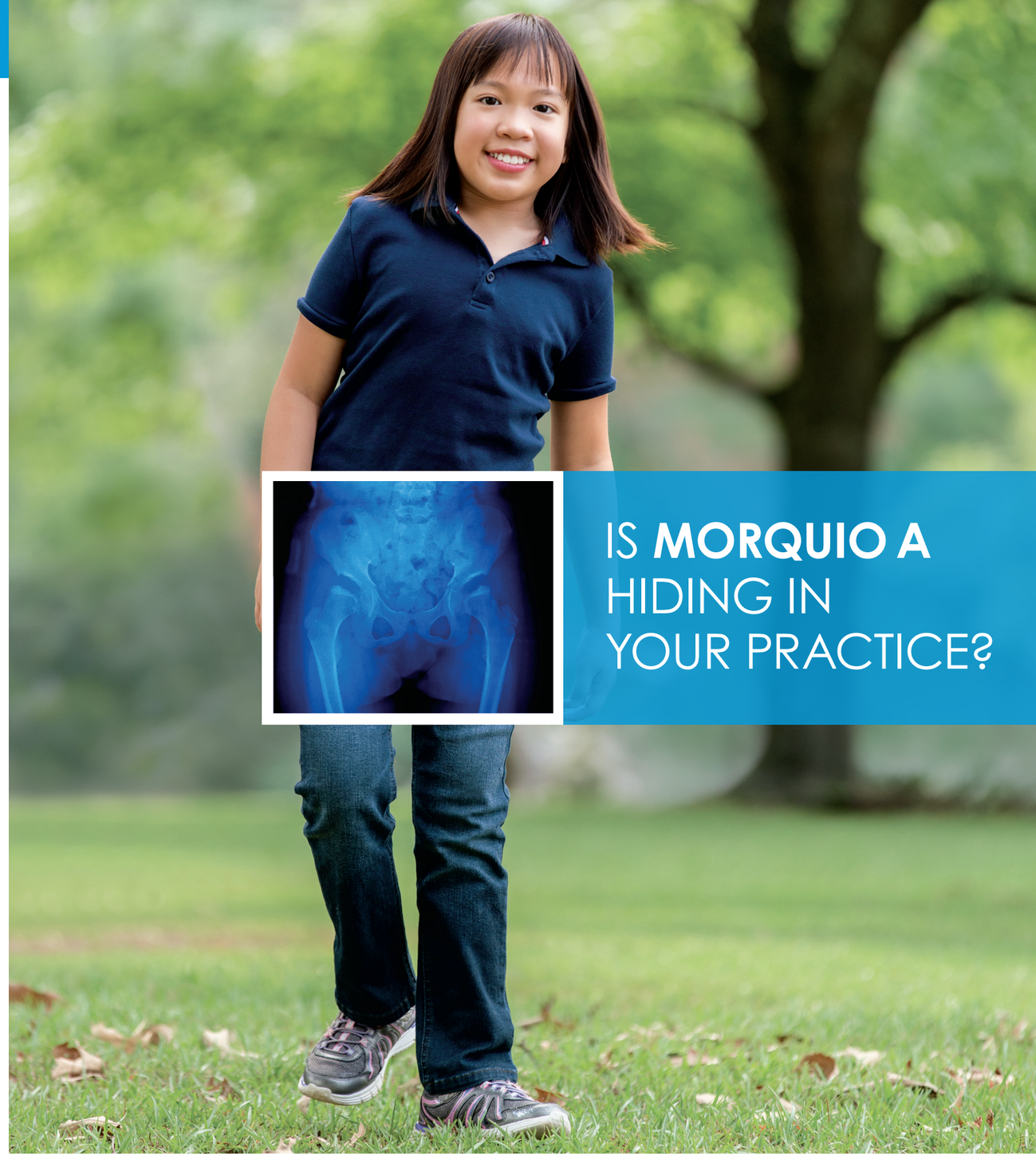

IS **MORQUIO A** HIDING IN YOUR PRACTICE?

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Diverse presentations of Morquio A can complicate patient identification

Morquio A presents similarly to other skeletal conditions[1]

- These include multiple epiphyseal dysplasia (MED), spondyloepiphyseal dysplasia (SED), pseudoachondroplasia, and bilateral Legg–Calvé–Perthes disease[1]
- Signs of Morquio A include skeletal dysplasia combined with nonskeletal organ system involvement, dystosis multiplex, and/or bilateral hip disease[2,*]
- Diagnosis of Morquio A is often delayed over 2 years due to misdiagnosis[3,4]

Nonclassical musculoskeletal presentation can include the following[1,3,5]:

- Potential height of > 140 cm
- Hip stiffness and pain
- Lack of overt skeletal presentation

In an international registry, 1 out of 4 Morquio A patients has a nonclassical phenotype[4]

Classical musculoskeletal presentation can include the following[1,6]:

- Height of < 120 cm
- Overt spinal and skeletal abnormalities
- Joint laxity
- Genu valgum
- Abnormal gait
- Chest abnormalities

*Patients may present with nonskeletal manifestations including respiratory, cardiovascular, visual, auditory, muscular, dental, and oral abnormalities.[7]

Patients with Morquio A require disease-specific surgical plans

SURGERY CAN BE LIFE THREATENING IN PATIENTS WITH MORQUIO A[8]

≈ 75% of patients < 12 years of age and more than 95% of patients ≥ 12 years of age require surgical or medical interventions[9]

Life-threatening complications may include paralysis, heart failure, and respiratory failure[10]

There is high risk of anesthesia-related morbidity and mortality due to[10]:

- Cervical instability and myelopathy
- Compromised airways and respiratory function
- Cardiac abnormalities

Surgical complications may account for mortality in 1 out of 10 patients with Morquio A[8]

Patients with Morquio A benefit from a team of multidisciplinary specialists: a metabolic specialist or physician, a paediatrician, an orthopaedic surgeon, an anesthesiologist, a cardiologist, a respiratory physician, an ENT specialist, a neurosurgeon, and a radiologist.[2,10]