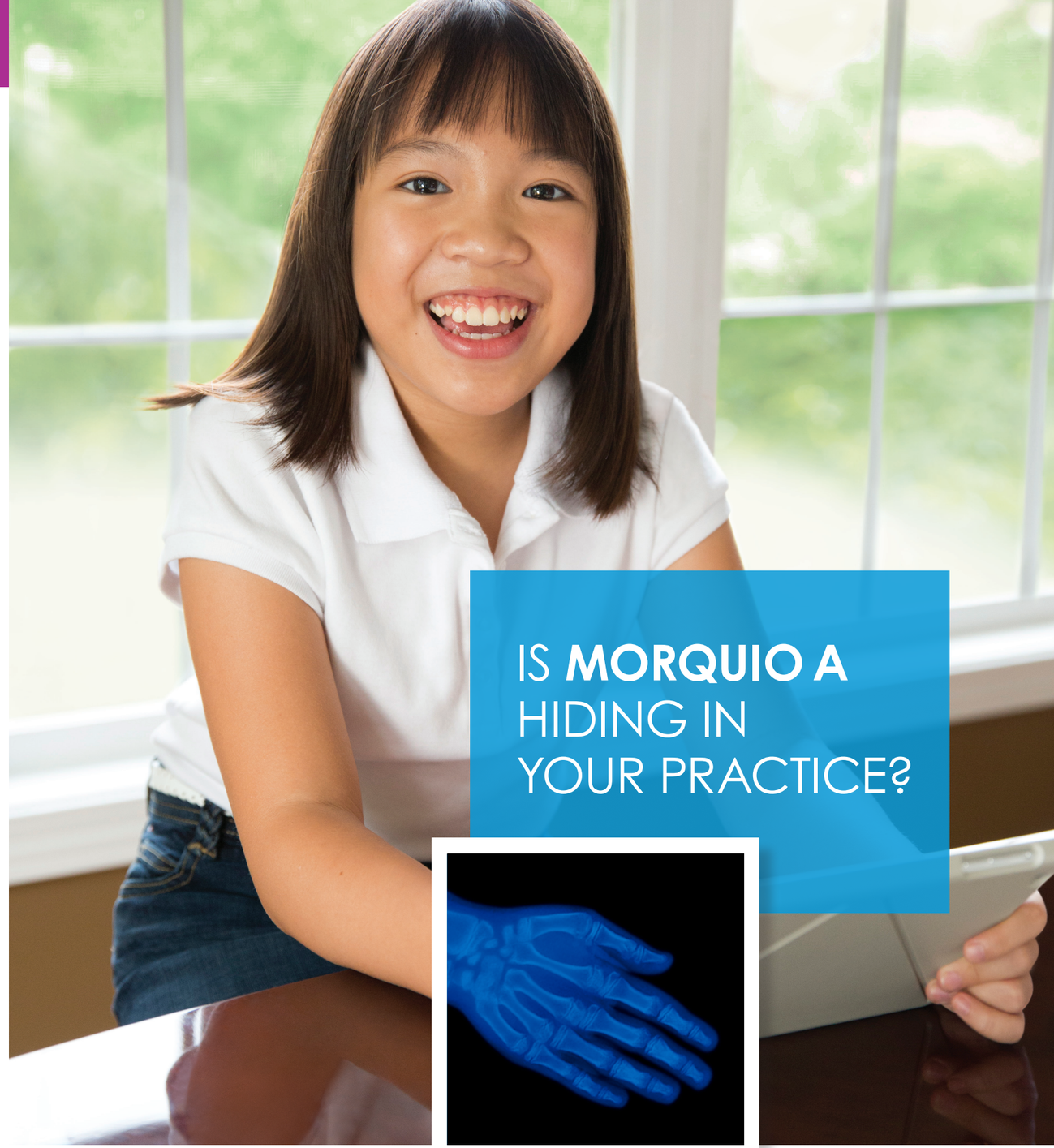
Refer patients to a geneticist for testing as soon as Morquio A is suspected[9]

Earlier diagnosis of Morquio A may improve long-term patient outcomes[2]

- The nonclassical phenotype of Morquio A can be easily missed, even among specialists[1]
- Nonclassical patients that present with joint pain, laxity, or contractures, and no systemic or local signs of inflammation may have Morquio A[9]
- A geneticist can perform a GALNS enzyme activity analysis to rule out or confirm a diagnosis of Morquio A[9]

Rule out or confirm Morquio A with an enzyme activity analysis?



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

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BioMarin Pharmaceutical Australia Pty Ltd
119 Willoughby Rd, Crows Nest, NSW 2065, Ph +61 2 9967 8662,
Medical Information 1800 387 876, medinfoasia@bmrn.com

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Morquio A is a progressive genetic disease with a multisystemic impact[1]

Morquio A (mucopolysaccharidosis [MPS] IVA) is an extremely rare autosomal recessive lysosomal storage disorder (LSD), caused by deficiency in the enzyme N-acetylgalactosamine-6-sulfatase (GALNS).[2]

- Deficient enzyme activity leads to an accumulation of glycosaminoglycans (GAGs), primarily in the bones and joints, causing progressive multisystemic and musculoskeletal complications[1,3]
- High degree of genetic heterogeneity may be responsible for the wide spectrum of phenotypic presentations[1]
  - Over 277 mutations have been identified encoding the *GALNS* gene[4]

Diverse presentations of Morquio A can complicate patient identification[2]

Nonclassical musculoskeletal presentation can include the following[1,2,5]:

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

In an international Morquio A registry, 25% of patients with Morquio A have a nonclassical phenotype[6]

Classical musculoskeletal presentation can include the following[2,7]:

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

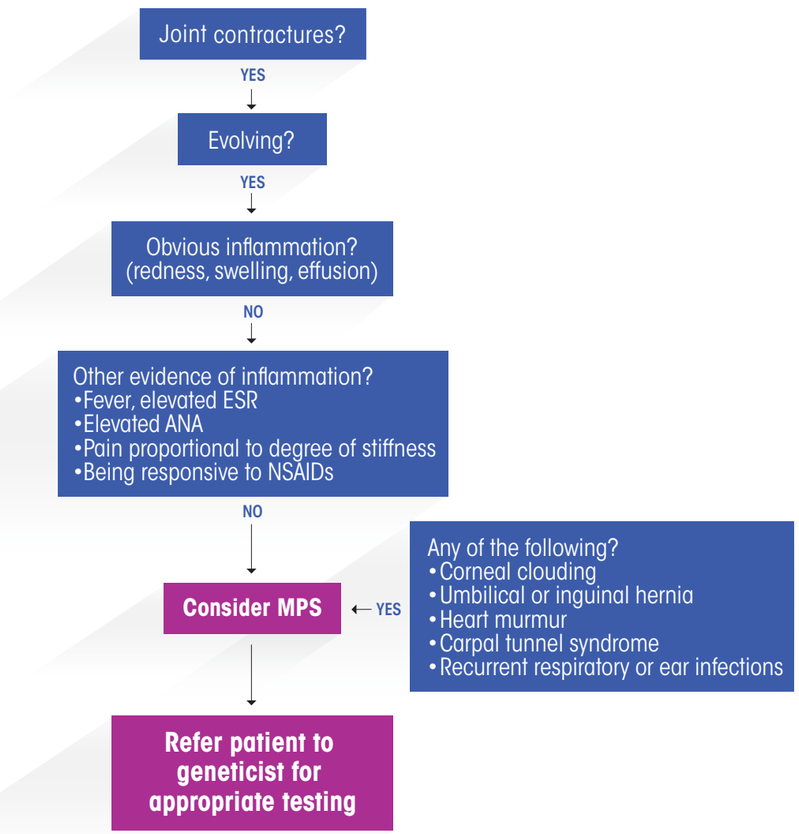
Rheumatologists are in a unique position to identify Morquio A earlier[8]

Bone and joint related manifestations are common in the very early stages of MPS, including Morquio A, and patients often visit a rheumatologist before being diagnosed[9]

Common bone and joint manifestations include[2,9]:

- Joint laxity
- Reduced hand function
- Carpal tunnel syndrome

The following diagnostic algorithm is recommended for the evaluation of any patient with joint pain, stiffness, or contractures, and no systemic or local signs of inflammation[9]:



Evolving joint pain in the absence of inflammation should raise the suspicion of an MPS disorder[9]

ANA, antinuclear antibodies; ESR, erythrocyte sedimentation rate; NSAIDs, nonsteroidal anti-inflammatory drugs.

Adapted from *Rheumatology.* 2011;50:v41-v48.