WHAT WOULD YOU SUSPECT?




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Diverse presentations of Morquio A can complicate patient identification[1]

Morquio A is a multisystemic disorder and patients may present with different subsets of symptoms[1,2]

- Deficient enzyme activity leads to an accumulation of glycosaminoglycans (GAGs) primarily in bones, joints, and organs, causing progressive multisystemic and musculoskeletal complications[2,3]
- Nonclassical phenotype patients may lack pronounced skeletal manifestations that would lead to suspicion of Morquio A[1,2]

Nonclassical musculoskeletal presentation can include[1,2,4]:

- Potential height of >140 cm
- Hip stiffness and/or pain may be the primary presenting symptoms
- Lack of overt skeletal manifestations

In an International Registry,
1 out of 4
patients with Morquio A has a nonclassical phenotype[6]

Classical musculoskeletal presentation can include[1,5,6]:

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

Morquio A can progress to serious complications regardless of phenotype[2]

Patients with Morquio A are at risk for surgical and non-surgical complications

SURGICAL RISKS

Surgery can be life-threatening in patients with Morquio A.[7,8]

- Almost 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 during surgery include paralysis, heart failure, and respiratory failure[8]
- Patients are at high risk of anesthesia-related morbidity and mortality due to[7,8]:
  - Cervical instability and myelopathy
  - Compromised airways and respiratory function
  - Cardiac abnormalities

OTHER RISKS

All patients are at risk for progressive organ damage due to N-acetylgalactosamine-6-sulfatase (GALNS) enzyme deficiency.[1,5,9,10]

- Cardiovascular[9]
  - Mitral and aortic valve stenosis and regurgitation, tricuspid regurgitation, hypertrophy
- Pulmonary[1]
  - Respiratory failure, recurrent respiratory infections, obstructive sleep apnea, progressive endurance insufficiency
- Ear, Nose, Throat[3]
  - Airway obstruction, conductive and neurosensory hearing loss

Surgical complications may account for mortality in 1 out of 10 patients with Morquio A[7]

MORQUIO A: WHO TO TEST?

Earlier diagnosis of Morquio A may improve long-term patient outcomes[1]

Diagnosis of Morquio A is often delayed, sometimes for years, due to misdiagnosis[2,6]

Perform enzyme activity analysis if patients[1]:

- Have been diagnosed with any of these disorders without confirmatory testing
- Are currently undergoing evaluation for any of these disorders

> – Multiple epiphyseal dysplasia (MED)
> – Spondyloepiphyseal dysplasia (SED)
> – Bilateral Legg-Calvé-Perthes disease
> – Pseudoachondroplasia
> – Undiagnosed skeletal dysplasia

If you suspect Morquio A, confirm with enzyme testing

Gabe, age 11

Chief Complaints:

- Deteriorating muscle weakness
- Increasing loss of ambulation
- Increasing fatigue
- Pain

Original Diagnosis:

- SED diagnosis by paediatric orthopedist at age 5 (2009)
- Referred for genetic counseling, SED molecular screen negative; SED diagnosis maintained
- Seen by paediatric orthopedist 2009-2015 for ongoing complications

Morquio A Diagnosis:

- Referred back to geneticist for worsening joint pain and decreasing stamina, December 2015
- Diagnosed with Morquio A using whole exome sequencing, April 2016

Raul, age 18

Chief Complaints:

- Pain in legs, knees, and back
- Frequent falls

Original Diagnosis:

- Referred to orthopaedic specialist in 2010
- Multiple X-rays revealed deformities in lower extremities
- Diagnosed with SEDT (SED tarda)

Morquio A Diagnosis:

- Orthopaedic team reassessed patient's history and realised the **SEDT diagnosis had never been confirmed by a laboratory**
- Mucopolysaccharide (MPS) enzyme activity analysis was ordered which led to Morquio A diagnosis, December 2015

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