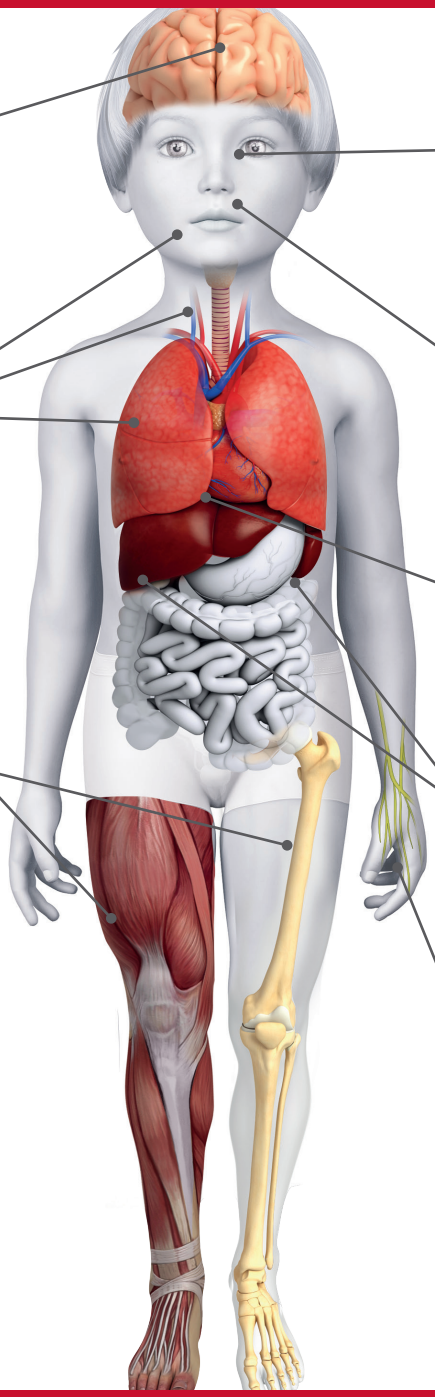
THINK OF **MPS I** (Mucopolysaccharidosis type I) IN YOUR **DIFFERENTIAL DIAGNOSIS**

It is a treatable disease

Common signs and symptoms[1]



NERVOUS SYSTEM

- Cognitive impairment **46%** **31%**

EAR-NOSE-THROAT

- Sleep disturbances/ snoring **52%** **33%** 27%
- Enlarged tongue **41%** **38%**
- Enlarged tonsils **29%** **33%**

MUSCULOSKELETAL

- Joint contractures **38%** **57%** 69%
- Dysostosis multiplex **44%** **38%** 35%
- Kyphosis/Gibbus Deformity **70%** **34%**
- Hip dysplasia **26%** 25%

MPS I SPECTRUM OF PHENOTYPES:

H : Hurler (the most severe, rapidly progressive)

H/S : Hurler-Scheie (slowly progressive)

S : Scheie (attenuated)

EYES

- Corneal clouding **71%** **68%** 70%

FACE

- Coarse facial features **86%** **73%** 48%

HEART

- Cardiac valve abnormalities **49%** **59%** 68%

VISCERAL

- Hepatomegaly **70%** **67%** 48%
- Hernia **59%** **60%** 54%
- Splenomegaly **51%** **47%** 28%

NERVOUS SYSTEM

- Carpal tunnel syndrome **28%** 51%

OTHER

- Respiratory tract infections[2]
- Premature cessation of skeletal growth[3]

1. Beck M, et al. Gen. Med. 2014;16:579-765.
2. Berger KI., et al. J Inherit Metab Dis . 2013;36:201-210.
3. Muenzer et al. Pediatrics 2009;123:19-29

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SUSPECT **MPS I** IN CASE OF...[1]

- Airway obstruction
- Regular Ear-Nose-Throat infections

- Carpal tunnel syndrome
- Cognitive impairment

- Hernia/Bulging abdomen
- Hepato-splenomegaly

AND/OR

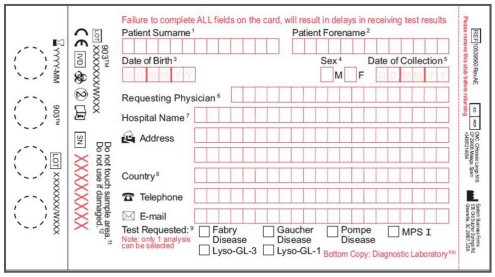
- Joint contractures
- Gait abnormalities
- Claw hands
- Trigger fingers
- Skeletal deformities

- Corneal clouding/vison impairment
- Cardiac valve abnormalities
- Facial coarse features
- Short stature
- Familial history

TEST THE PATIENTS & THEIR **SIBLINGS, TREAT EARLY**[4,5]

Brother(s) and/or sister(s) of a diagnosed patient may have MPS I as well, even if they don't show the same signs and symptoms.
Early diagnosis and early treatment are crucial to reduce disease progression before irreversible damage occurs.[4,5]

MPS I can be definitively diagnosed or ruled out with a simple blood-based enzymatic assay. It has to be confirmed by genotyping. Measurement of urinary glycosaminoglycan (GAG) levels is optional but nonspecific.[3]



A DBS testing kit is distributed through Sanofi Genzyme as an initial screening test.

Please contact your Sanofi Genzyme Product Specialist

Contact person in your lab:.....

Tel.:.....

4. Gabrielli et al. BMC Medical Genetics. 2016;17:19. 5. Bruni et al. MGM Reports. 2016; 8: 67-73

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