

**Recommended schedule of assessments for patients with MPS VI<sup>1</sup>**

| <b>Imaging study</b>  | <b>Initial assessments</b> | <b>Every 3 months</b> | <b>Every 12 months</b> | <b>As clinically indicated <sup>a</sup></b> |
|---|----------------------------|-----------------------|------------------------|---|
| <b>Confirmation of MPS VI</b>   | ●                          |                       |                        |   |
| <b>Medical history<sup>b</sup></b>  | ●                          |                       | ●                      |   |
| <b>Physical examination</b>   | ●                          |                       | ●                      |   |
| <b>Neurologic examination</b>   | ●                          |                       | ●                      |   |
| <b>Height, weight</b>   | ●                          |                       | ●                      |   |
| <b>Head circumference<sup>c</sup></b>   | ●                          |                       | ●                      |   |
| <b>Tanner stage<sup>d</sup></b>   | ●                          |                       | ●                      |   |
| <b>Photographs</b>  | ●                          |                       |                        | ●   |
| <b>Endurance<sup>e</sup></b>  |                            |                       |                        |   |
| 12-minute walk test   | ●                          |                       | ●                      |   |
| 3-minute stair climb  | ●                          |                       | ●                      |   |
| <b>Ophthalmology</b>  |                            |                       |                        |   |
| Visual acuity   | ●                          |                       | ●                      |   |
| Corneal examination   | ●                          |                       | ●                      |   |
| Fundoscopy examination  | ●                          |                       | ●                      |   |
| Intraocular pressure  | ●                          |                       | ●                      |   |
| Refraction  | ●                          |                       | ●                      |   |
| <b>Audiometry</b>   | ●                          |                       | ●                      |   |
| <b>Cardiology</b>   |                            |                       |                        |   |
| Echocardiogram  | ●                          |                       | ●                      |   |
| Electrocardiogram   | ●                          |                       | ●                      |   |
| Blood pressure  | ●                          |                       | ●                      |   |
| <b>Electrophysiology</b>  |                            |                       |                        |   |
| Nerve conduction <sup>f</sup>   | ●                          |                       |                        | ●   |
| <b>Pulmonary function</b>   |                            |                       |                        |   |
| Forced vital capacity, forced expiratory volume in 1 second, maximum voluntary ventilation <sup>g</sup> | ●                          |                       | ●                      |   |
| Sleep study   | ●                          |                       |                        | ●   |
| <b>Imaging studies</b>  |                            |                       |                        |   |
| Hip films <sup>h</sup>  | ●                          |                       |                        | ●   |
| Skeletal survey   | ●                          |                       |                        | ●   |
| Flex/ext radiograph of cervical spine   | ●                          |                       |                        | ●   |
| MRI of brain and spine <sup>i</sup>   | ●                          |                       |                        | ●   |
| <b>Laboratory assessments</b>   |                            |                       |                        |   |
| uGAG levels   | ●                          |                       | ●                      |   |
| <b>Supplemental assessments for patients on ERT<sup>j</sup></b>   |                            |                       |                        |   |
| Total anti-ASB antibody <sup>k</sup>  | ●                          | ●                     | Yearly after 24 months |   |

Adapted from Giugliani, *Pediatrics*, 2007.

Abbreviations: MPS, mucopolysaccharidosis; MRI, magnetic resonance imaging; uGAG, urinary glycosaminoglycan.

<sup>a</sup> “As clinically indicated” generally means every 2 to 3 years depending on the rate of disease progression and clinical symptoms.

<sup>b</sup> For infants, more frequent examinations are necessary.

<sup>c</sup> Monitored until head growth has stopped.

<sup>d</sup> Continue assessments until pubertal maturation is completed.

<sup>e</sup> Endurance-testing paradigm before and after Enzyme replacement therapy (ERT): distance walked in 12 minutes (or 6-minute walk test per American Thoracic Society guidelines, but preferably same minute length as completed in previous test); number of stairs climbed in 3 minutes.

<sup>f</sup> Median nerve conduction measured to evaluate carpal tunnel syndrome.

<sup>g</sup> Pulmonary function tests are to include forced vital capacity, forced expiratory volume in 1 second, and maximum voluntary ventilation.

<sup>h</sup> Anteroposterior and “frog-leg” lateral views of pelvis.

<sup>i</sup> MRI of brain and spinal cord may require sedation or general anesthesia depending on patient age and cooperation. General anesthesia carries substantial risk for patients with MPS VI.

<sup>j</sup> For patients on ERT, results should be obtained at baseline, then at months 3, 6, 12, 18, and 24, and then yearly.

<sup>k</sup> Anti-ASB antibody testing is only available for US patients enrolled in the clinical surveillance program.

**Reference: 1.** Giugliani R, Harmatz P, Wraith JE. Management guidelines for mucopolysaccharidosis VI. *Pediatrics*. 2007;120:405-418. doi:10.1542/peds.2006-2184.