Oculopharyngeal Muscular Dystrophy Research and Treatment Updates

Patients with oculopharyngeal muscular dystrophy (OPMD) typically experience a slow progression of three hallmark symptoms: dysphagia, ptosis, and limb weakness. Treatments for this rare genetic muscle disorder are mainly supportive.

Kara Stavros, MD, FAAN, associate professor of neurology at the Warren Alpert Medical School of Brown University and director of the neuromuscular division at Rhode Island Hospital, provides an overview of current treatment and the most promising research.

Current treatments for OPMD address the three main symptoms:

- **Dysphagia**. Speech and swallowing specialists help patients decrease the risk of choking and aspiration pneumonia. When dysphagia progresses, procedures – including cricopharyngeal myotomy – or feeding tubes may be offered.
- **Ptosis**. Drooping eyelids may impair a patient's vision and make them self-conscious about their appearance. Treatment options include eyeglass attachments that support eyelids or corrective surgery.
- **Limb weakness**. Leg weakness increases the risk of falling. Physical therapy may help with balance, improved gait, and fall prevention strategies. Some people with OPMD use assistive devices such as canes, walkers, or motorized scooters. Physical or occupational therapy may help with proximal arm weakness.

Researchers are investigating disease-modifying treatments for OPMD. A phase 1b/2a clinical trial for BB-301, an adeno-associated virus vector-delivered gene therapy treatment, recently dosed its first patient. The investigational drug is designed to silence and replace the mutated poly-A binding protein nuclear-1 (PABPN1) gene.

"It is in the very early stages and still under investigation," Dr. Stavros says. "We've seen advances in treatments for Duchenne muscular dystrophy and spinal muscular atrophy, which led to hopes that we could have breakthroughs for other neuromuscular conditions, like OPMD."

Other researchers are studying in mice the effects of monoclonal antibodies on myostatin, a negative regulator of muscle mass to investigate their effectiveness in slowing muscle atrophy by inhibiting myostatin.

"This is an interesting avenue of research," Dr. Stavros says, "but I think more studies are needed to better understand the mechanism for how this could impact the disease course."

OPMD Resources:

- Share Simply Stated: Updates in Oculopharyngeal Muscular Dystrophy (OPMD) with patients who'd like to learn more about the disease.
- Direct patients to MDA's Clinical Trials Finder to find opportunities to participate in research.
- The Neuromuscular Disease Center website of Washington University in St. Louis is a resource for providers.
- Dr. Stavros recommends the neurology journal "Continuum" as another resource for providers.