



## **The Endocrinology Factor in Muscular Dystrophy**

Neuromuscular diseases can impact many processes and systems in the body, including the endocrine system. According to Sasigarn A. Bowden, MD, Professor of Pediatrics at The Ohio State University Division of Pediatric Endocrinology at Nationwide Children's Hospital, muscular dystrophy raises particular concerns from the endocrinologist's perspective.

"Duchenne muscular dystrophy (DMD) is the common focus, and the top endocrine concerns are bone fragility, fractures, delayed puberty, and short stature," Dr. Bowden says.

For providers, it can be challenging to find therapies that help reduce endocrine complications that come with the disease.

"Often, boys with DMD are on chronic corticosteroid treatment of either Prednisone or Deflazacort," she says. Taking these steroids long-term causes significant adverse effects such as weight gain, poor growth, delayed puberty, and low bone density.

In addition, as muscles deteriorate, patients become less physically active. “When they stop walking, there is no weight bearing to stimulate bone and being on chronic steroids can sometimes cause impairment to bone formation and increase bone absorption, so there is significant bone loss,” Dr. Bowden says.

This can lead to problems, such as compression fractures, that can cause significant morbidities.

## **Solutions may include:**

- Monitoring bone density
- Spinal X-ray to detect compression fractures
- Bone-strengthening treatments

## **Bone-strengthening treatment**

By monitoring bone health, providers can determine when a patient may need to start treatment to improve their bone density.

When Dr. Bowden sees early signs of fractures, she talks with patients and families about taking bisphosphonates, currently the only bone treatment for those with DMD with secondary osteoporosis.

“In the future, I think there will be new treatments, but right now bisphosphonates in either oral medication or IV is used,” Dr. Bowden says. “I’m more inclined to use the IV because the adverse effects from steroids are quite significant.”

Dr. Bowden also makes sure her patients have adequate Vitamin D intake.

## **Advances and challenges in Endocrinology**

Bisphosphonates have not been approved for pediatric use by the US Food and Drug Administration (FDA). “What we use right now is off label,” Dr. Bowden says.

Some in the Endocrinology field are moving toward giving bisphosphonates as prophylactic treatment.

“There is a study from Australia in a randomized controlled trial to assess prophylactic zoledronic acid for steroid-treated boys with DMD, and there were fairly good results that it helps improve bone density,” she says. “Without an intervention, patients suffer from severe osteoporosis, but the timing is debatable. We used to wait until the first fracture, but now we monitor spinal imaging either by X-rays or vertebral assessment by DXA scans, and once we see signs of compression fracture, even when the patient is asymptomatic, we start treatment.”

Dr. Bowden has observed that geography is a common barrier for patients to receive endocrine-related treatment. Many of her patients travel long distances to receive an infusion of zoledronic acid.

“I also work with local physicians by providing protocols and plans, so patients can receive the bisphosphonates infusion closer to home,” she says.

## Including Endocrinology in multidisciplinary care

Multidisciplinary teams are common in neuromuscular disease care, and endocrinologists often should be included in these teams for conditions like DMD. While very young patients may not have any endocrine complications, the care team should monitor bone health and make a referral to an endocrinologist if there are any abnormal results.

“A patient might have a team that includes a neurologist, cardiologist, pulmonologist, pediatric endocrinologist, gastroenterologist, genetic counselor, and physical therapists,” Dr. Bowden says. “It can be a lot, but communication helps in scheduling and in working well together.”

## Endocrinology resources

To find Endocrinology resources relevant for neuromuscular care providers, Dr. Bowden recommends being connected to the [Pediatric Endocrine Society Bone and Mineral](#) special interest group or [Parent Project Muscular Dystrophy](#). You can also find lectures and videos that are helpful for healthcare professionals at [Project ECHO for Duchenne Muscular Dystrophy](#).

