Case Study

Diagnosis of Duchenne Muscular Dystrophy (DMD) via Telemedicine: *A Physiatrist Referral*

Patient Characteristics and History

- 4-year-old male referred with hyperCKemia (CK: 17,000 U/L)
- Walked at 18 months (preferred to crawl)
- Diagnosed with an ASD
- No family history of DMD
- Lived 2 hours away from the neuromuscular clinic

Telemedicine in Practice

The initial neuromuscular clinic consult was held virtually due to the family's comfort level and distance from the clinic.

Virtual physical exam was suggestive of muscular dystrophy.

- Calf pseudohypertrophy
- Gowers' maneuver
- Trendelenburg gait
- Lordosis
- Toe walking
- Lack of eye contact
- Difficulties with verbal communication

Watching the patient's functional movements (eg, walking down a hallway and upper extremity ROM) can help identify patterns of muscle weakness.

Plan included genetic testing for DMD.

Educational materials were provided through the screen share function.

- The family was referred to appropriate educational resources (eg, exon map, ClinicalTrials.gov, Parent Project Muscular Dystrophy)

Advantages to telemedicine use

The patient can be observed holistically in his natural environment

- Observation of daily activities
- Assessment of the home environment
- Increased comfort level for patients
- Convenience and safety benefits (eg, reduced travel and exposure to illness)

Challenges to telemedicine use

Fundoscopic and pupil eye exams, individual muscle strength, and reflexes are difficult to assess virtually

Camera angles may need to change for different analyses (eg, walking versus ROM)

Outcome

A Decode Duchenne genetic testing kit was ordered and sent to the patient's home

Additional lab tests were ordered locally through EPIC

The DMD diagnosis was confirmed

A steroid regimen was initiated (weight was checked on a family scale at home to determine the dose)

The patient continued with virtual follow-up but will come in for an in-office visit in the future

Goryeb Children's Hospital
ATLANTIC HEALTH SYSTEM

Image for illustration purposes only and not representative of the actual patient case.

ASD, autism spectrum disorder; CK, creatine kinase; ROM, range of motion.

SAREPTA THERAPEUTICS