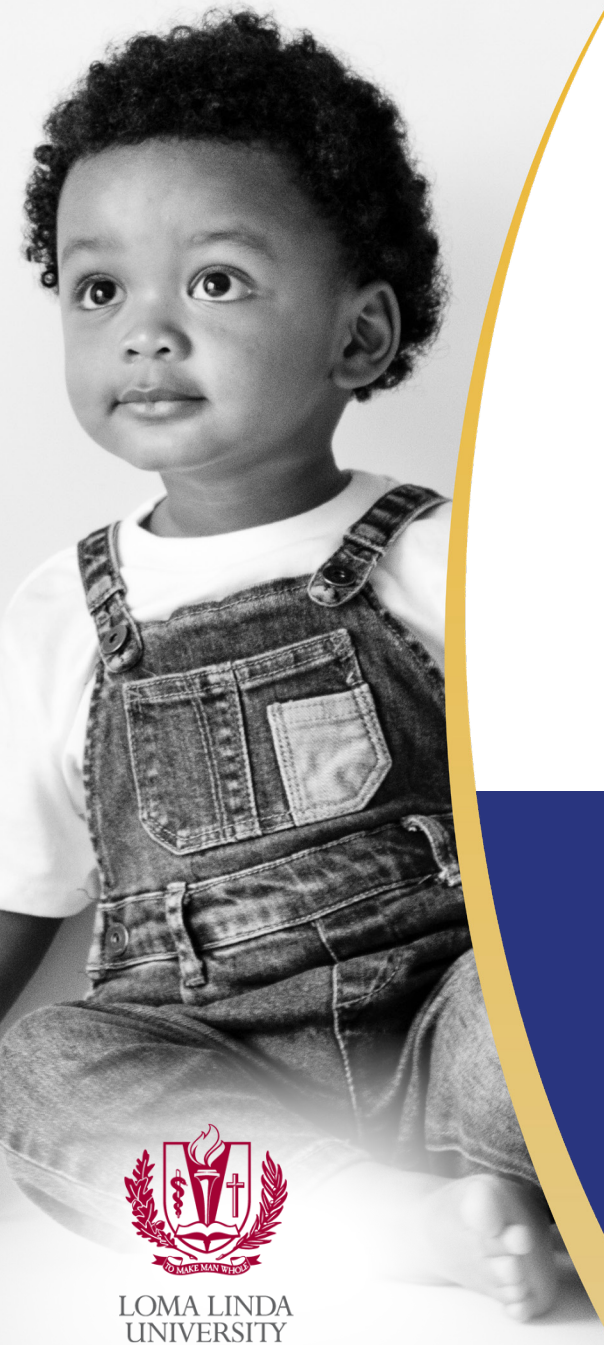
Case Study



LOMA LINDA UNIVERSITY

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

Diagnosis of Duchenne Muscular Dystrophy (DMD) via Telemedicine: *Identification From the ED*

Patient Characteristics and History

- 2-year-old male patient
- CK ordered from pediatrician was 15,000 U/L
- Patient had difficulty running and climbing stairs
- Maternal uncle was diagnosed with DMD and passed in 1989

Background

- The patient went to the ED for abnormal lab results
- ED physician ordered additional labs and noted elevated transaminase levels (10x normal) and CK levels (16,691 U/L)
- ED physician also indicated that the patient had motor difficulties and proximal weakness and used the Gowers' maneuver to get up
- ED physician reached out to pediatric neurology department and instructed the family to make an appointment for the next week, but they were not seen for another month due to insurance issues

Pediatric neurology clinic setup

- Telemedicine platforms used: EPIC and Zoom
- In-office appointments utilized for select assessments only (eg, cardiac echocardiogram)

Outcome

- An initial telemedicine appointment with the pediatric neurologist was scheduled
- The appointment included the occupational therapist to assess patient functionality

  The patient was uncooperative with the functional assessments
- A genetic testing kit was ordered to be sent to the patient's home
- The patient was started on steroid treatment
- The genetic test results are still pending

CK, creatine kinase; ED, emergency department.

SAREPTA THERAPEUTICS