Duchenne Muscular Dystrophy (DMD)
Patient Clinical Journey

Parent Project Muscular Dystrophy — JOIN THE FIGHT. END DUCHENNE.

EveryLife Foundation for Rare Diseases

DELAYED DIAGNOSIS

Items in red may be prevented with a Timely Diagnosis.

PRE-DIAGNOSIS (ages 0-5)

Typical Accompanying Diagnoses
- Abnormal transaminase
- Cognitive impairments
- Gait issues
- Gross Motor Delays
- Muscle Weakness
- Neurodevelopmental Delays
- Speech and language disorder

Events
- Incidental hospitalizations

Specialists
- Developmental Pediatrician
- Doctor's visits
- Gastroenterologist
- Geneticist
- Hepatologist
- Neurologist
- Neuropsychologist
- Orthopedist
- Orthotist
- Physical Therapist (PT)/Occupational Therapist (OT)/Speech Pathologist (SP)

Testing & Procedures
- Developmental screen
- *The following are often ordered by specialists suspecting other conditions:*
  - Genetic testing
  - Liver enzyme
  - Liver biopsy

Treatments & Supportive Therapies
- Ankle-Foot Orthotics (AFO) – *Can get prescribed at PT after children miss milestones*
- Supramalleolar Orthosis (SMO) – *Seen particularly in younger kids*

TIME OF DIAGNOSIS (ages 4-7)

Typical Accompanying Diagnoses
- *All previously listed diagnoses may remain as accompanying diagnoses as treatment begins.*

Specialists
- Cardiologist
- Geneticist
- Neurologist
- Neuropsychologist
- Nutritionist - *Care guidelines suggest seeing nutritionist at introduction of steroids. Constant need but treatment changes during diagnostic journey.*
- Pulmonologist
- PT/OT/SP

Testing & Procedures
- Behavior Evaluation
- Developmental Screen
- Echocardiogram
- Genetic Testing
- Muscle Biopsy

Treatments & Supportive Therapies
- AFO's / SMO's
- Exon Skipping Therapy
- Steroids

POST DIAGNOSIS

AMBULATORY

Testing & Procedures
- Bone Density (DEXA) Scan, Bone Health Labs
- Cardiac MRI, Electrocardiogram (ECG)

Treatments & Supportive Therapies
- Gene therapy
- Bisphonphonate – *At the time of first fracture*
- Cardiac medications – *By age 10*
- Cough assist device – *Sometimes proactively prescribed by clinics.*
- Scooters/Wheelchairs – *Some families get a wheelchair prior to loss of ambulation to help conserve energy. Some families wait until non-ambulation. This is a personal decision to be made with each family's specialists.*
- Splints – *Especially for nighttime stretching.*

Events
- Hospitalizations – *Could be related to Duchenne or incidental.*

Specialists
- Cardiologist; Geneticist; Neurologist; Neuropsychologist; Pulmonologist; Counseling Support Services
- Nutritionist; PT/OT/SP
- Rehabilitation – *At some clinics can be introduced at time of diagnosis or before*

NON-AMBULATORY

Typical Accompanying Diagnoses
- Loss of ambulation - *A variety of Durable Medical Equipment (DME) can be needed to care for a non-ambulatory patient, including but not limited to lifts, hospital beds, and home modifications.*

Testing & Procedures
- Scoliosis surgery

Treatments & Supportive Therapies
- Feeding tube
- Implantable heart device
- Ventilation support (non-invasive or invasive)

Without treatment, life expectancy is approximately 26 years

26 YEARS

TIMELY DIAGNOSIS

Care guidelines will only be established as more babies are diagnosed at birth. It is still unclear if many of the following treatments/therapies are delayed or unnecessary by early diagnosis. Some of these services may remain but can be more appropriately administered because of the diagnosis.

While a timely diagnosis extends the mean age of life expectancy and delays the loss of ambulation, the exact age ranges are currently unknown. The time of diagnosis age ranges displayed here are based on typical ages of diagnosis and input from patients and experts.

TIME OF DIAGNOSIS (ages 0-2)

Testing & Procedures
- Newborn Screening – *dried blood spot for elevated CK levels*
- Prenatal testing
- Genetic testing
- Echocardiogram

Specialists
- Cardiologist
- Geneticist
- Neurologist
- Neuropsychologist
- PT/OT/SP

Treatments & Supportive Therapies
- Exon Skipping Therapy
- Steroids – *There is still debate among specialists about the appropriate time to begin steroid treatment*

Typical Accompanying Diagnoses
- Cognitive impairments
- Gross motor delays
- Muscle weakness
- Neurodevelopmental Delays
- Speech Language Delays

POST DIAGNOSIS

AMBULATORY

Testing & Procedures
- Developmental Screen; Echo

Treatments & Supportive Therapies
- Gene therapy
- AFO's / SMO's
- Steroids – *By age 4 if not begun as an infant*
- Bisphonphonate – *Time of first fracture*
- Cardiac medications – *By age 10*
- Cough assist device – *Sometimes proactively prescribed by clinics*
- Scooters/Wheelchairs – *Some families get a wheelchair prior to loss of ambulation to help conserve energy. Some families wait until non-ambulation. This is a personal decision to be made with each family's specialists*
- Supplements (Calcium & Vitamin D)

Events
- Hospitalizations – *Could be related to Duchenne or incidental*

Specialists
- Cardiologist; Geneticist; Neurologist; Neuropsychologist; Pulmonologist; Counseling Support Services
- Nutritionist – Care guidelines suggest seeing a nutritionist at introduction of steroids Constant need but treatment changes during diagnostic journey.
- PT/OT/SP; Orthotist
- Rehabilitation

NON-AMBULATORY

Typical Accompanying Diagnoses
- Loss of ambulation

Testing & Procedures
- Scoliosis surgery

Treatments & Supportive Therapies
- Feeding tube; Implantable heart device
- Ventilation support (non-invasive or invasive)

**This guide is not intended as a clinical decision-making tool. Please consult your specialists for any medical decisions.*