# Pompe Disease
## Patient Clinical Journey

EVERYLIFE FOUNDATION FOR RARE DISEASES

***LEGEND:*** ▲ *Severe Pompe (IOPD)* ■ *General Pompe (LOPD)* ● *Both*

## DELAYED DIAGNOSIS – CLINICAL OR SYMPTOM-PRESENTING

*Progression/severity of listed items is significantly reduced with a timely diagnosis.*

### PRE-DIAGNOSIS

**Typical Accompanying Diagnoses**

- Cardiomyopathy – *May lead to heart failure* ▲
- Hearing issues/speech delays ▲
- Severe muscle weakness ▲
- Respiratory distress – *up to and including respiratory failure* ▲
- Failure to thrive ▲
- Hepatomegaly ▲
- Global developmental delay ▲
- Cardiac abnormalities ●
- Cough weakness – *increased risk of choking* ●
- Difficulty sleeping ●
- Dysphagia (feeding/swallowing difficulties) ●
- Elevated biomarker signals (CK, Glc4, AST, ALT, CO2) ●
- Progressive muscle weakness and damage ●
- Respiratory decline – *higher risk for related illness* ●
- Ptosis ●
- *Misdiagnoses* ●
- Fatigue ■
- Lingual weakness ■
- Incontinence ■
- Hypercapnia ■

**Testing & Procedures**

- Frequent blood and urine tests ●
- Biopsy (muscle, liver, skin) ●
- Lumbar puncture (spinal tap) ●
- Imaging – *MRI, X-Ray, CT, ultrasound, and more* ●
- Breathing tests ●
- Electrocardiogram (ECG/EKG) ●
- Electromyography (EMG) ●
- Endoscopy ●
- Hearing tests ●
- Swallowing test ●
- Physical testing ●
- Sleep study ●
- Genetic testing ●

**Specialists**

- Primary care ●
- Pediatrician ●
- Neurologist ●
- Pulmonologist ●
- Nutritionist ●
- Physical Therapist/Occupational Therapist/Speech Pathologist (PT/OT/SP) ●
- Geneticist ●
- Cardiologist ●
- Audiologist ●
- Ophthalmologist ●
- Gastroenterologist ●
- Dentist ●

**Events**

- Cardiac failure – *and associated interventions* ▲
- Misdiagnosis ●
- Emergency department visits ●
- Death ●
- Respiratory failure – *and associated interventions* ●
- Frequent falls ■

*For IOPD patients, symptom progression is rapid. Mortality within the first year if not diagnosed and treated. Effectiveness and durability of treatment reduced as time/progression accumulates – high morbidity and mortality still associated with delayed treatment.* ▲

*For LOPD patients, symptom progression is slower. While symptom progression starts at birth, the rate is much slower. Years to decades without a diagnosis are typical. Effectiveness and durability of treatment reduced as time/progression accumulates.* ■

### TIME OF DIAGNOSIS

**Typical Accompanying Diagnoses**

- *All previously listed diagnoses may remain as an accompanying diagnosis until treatment begins*

**Testing & Procedures**

- *The following are standards of diagnosis:*
- GAA enzyme activity level ●
- Genetic testing ●
- CRIM status – *may be missed, leading to treatment rejection* ▲
- *The following are typical tests to characterize disease progression:*
- Biomarker tests – AST, ALT, CK, Glc4 ●
- Respiratory testing (pulmonary function testing) ●
- Echocardiogram/EKG ●
- Muscle/Respiratory/Brain imaging – *MRI, X-Ray, CT, ultrasound and more* ●
- Sleep study ●

**Specialists**

- *All previously listed specialists may continue to provide care.*

**Treatments & Supportive Therapies**

- Short-term immunomodulation ●
- Enzyme replacement therapy (ERT) ●
- Nutrition adjustment ●
- Physical therapy/exercise/activity adjustments ●
- Speech therapy/occupational therapy/feeding therapy ●
- *Additional treatments/therapies are currently being developed*

### POST DIAGNOSIS

**Typical Accompanying Diagnoses**

- Cardiac abnormalities ●
- Gastrointestinal issues ●
- Difficulty sleeping ●
- Dysphagia (feeding/swallowing difficulties) ●
- Respiratory insufficiency ●
- Progressive muscle weakness and damage triangle ▲
- *Other diagnoses may remain after treatment initiation, specific to each patient.*

**Specialists**

- Counseling Support Services ●
- Neurologist ●
- Geneticist ●
- Pulmonologist ●
- Nutritionist ●
- Physical Therapist (PT)/Speech Pathologist (SP)/Occupational Therapist (OT) ●
- Audiologist ●
- Ophthalmologist ●
- Gastroenterologist ●
- Dentist ●
- Cardiologist ●
- Dedicated nursing ●

**Testing & Procedures**

- Biomarker monitoring ●
- Immune response monitoring ●
- Muscle/Respiratory/Brain imaging – *MRI, X-Ray, CT, ultrasound and more* ●
- Feeding tubes ●
- Port insertion ●
- Heart/spinal surgeries ●
- Other surgeries – *specific to each patient* ●
- *All characterization of prognosis tests will continue. Additional procedures may be necessary, depending on the severity of Pompe and timing of diagnosis.* ●

**Events**

- Emergency department visits ●
- Death ●

**Treatments & Supportive Therapies**

- Medications to manage affected symptoms ▲
- Enzyme replacement therapy (ERT) ●
- Nutrition adjustments – *may include modified diet, texture adjustments, or a J/G/GJ-tube* ●
- Communication support – *may include hearing aids or ACC device* ●
- Developmental support – *such as an IEP or 504 plan* ●
- Ventilatory support – *may be full-time or periodic, depending on progression* ●
- Ambulatory/movement support – *may include orthosis, cane/walker, or powered wheelchair* ●
- Home/workplace accommodations ●

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

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## TIMELY DIAGNOSIS – NEWBORN SCREENING (NBS) OR PRENATAL SCREENING

### TIME OF DIAGNOSIS

#### Testing & Procedures

- *Timely diagnosis for IOPD is generally inclusive of a prenatal or newborn screening diagnosis (within approx. one week of life).* ▲
- *Timely diagnosis for LOPD is generally inclusive of a prenatal or newborn screening diagnosis. Symptom progression is slower in LOPD and can be monitored to identify an appropriate time to start treatment before progression is significant and irreversible.* ■
- Prenatal diagnosis – *CVS or amniocentesis* ●
- Fetal echocardiogram/fetal cardiac MRI ●
- Newborn screening (dried blood spot) ●
- Genetic testing ●
- GAA enzyme activity level ●
- Biomarker tests – AST, ALT, CK, Glc4 ●
- Cardiac ultrasound, ECG/EKG ●
- CRIM status ▲

#### Treatments & Supportive Therapies

- Short-term immunomodulation ●
- Enzyme replacement therapy (ERT) ●
- Physical therapy ●
- Nutrition adjustment ●

#### Typical Accompanying Diagnoses

*The following may be detectable for an IOPD patient prior to and around the time of birth:*

- Cardiac abnormalities ▲
- Elevated biomarker signals ▲
- Hearing/speech issues – *significantly reduced* ▲
- Dysphagia – *significantly reduced* ▲

*Treatment timing in LOPD is case-specific and may potentially be years after birth. The following may still present:*

- Elevated biomarkers ■
- Fatigue ■
- Global developmental delay ■
- Gross motor delay ■

#### Specialists

- Primary care ●
- Pediatrician ●
- Neurologist ●
- Geneticist ●
- Pulmonologist ●
- Nutritionist ●
- Physical Therapist/Speech Pathologist (PT/SP) ●

### POST DIAGNOSIS

#### Treatments & Supportive Therapies

- Enzyme replacement therapy (ERT) ●
- Nutrition adjustments ●
- Ambulatory/movement support – *orthotics* ▲
- Communication support – *may include hearing aids, speech training, or IEP* ▲

#### Specialists

- Counseling Support Services ●
- Primary care ●
- Pediatrician ●
- Neurologist ●
- Geneticist ●
- Pulmonologist ●
- Nutritionist ●
- Physical Therapist/Speech Pathologist (PT/SP) ●

#### Testing & Procedures

- Biomarker monitoring ●
- Immune response monitoring ●
- Muscle/Respiratory/Brain imaging – *MRI, X-Ray, CT, ultrasound and more* ●
- Respiratory testing (pulmonary function testing) ●

*All characterization of prognosis tests will continue. Additional procedures may be necessary.*

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