Alpha-glucosidase deficiency (Pompe disease)

Decreased Acid Alpha-glucosidase

Assess clinically (see ACT Sheet)
Routine labs (CK, LDH, AST, ALT)

GAA enzyme activity, Leukocytes, or dried bloodspot
Urine Hexose tetrasaccharide (Hex4)

Low GAA Activity
Abnormal Routine Labs
Abnormal Hex4

Low GAA Activity
Normal Routine Labs
Normal Hex4

Low GAA Activity
Abnormal Routine Labs
Normal Hex4

Normal GAA Activity
Abnormal Routine Labs
Normal Hex4

Normal GAA Activity
Normal Routine Labs
Normal Hex4

Molecular GAA Genetic Testing

Positive

Inconclusive

Pompe Disease

Further evaluation as guided by specialist

No further action required.

Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key
GAA = acid alpha-glucosidase
CK = creatine kinase
LDH = lactate dehydrogenase
AST = aspartate transaminase
ALT = alanine transaminase
Hex4 = hexose tetrasaccharide, also called glucose tetrasaccharide

Disclaimer. These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient record the rationale for any significant deviation from these standards and guidelines.