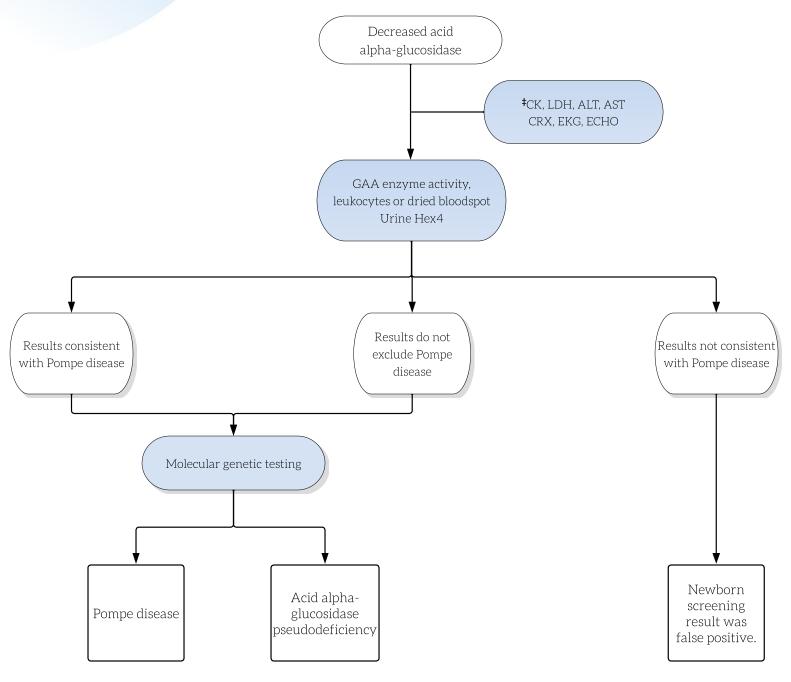


## Pompe disease: acid alpha-glucosidase deficiency



## Kev

- Actions are shown in shaded ovals; results are in the unshaded ovals. Diagnostic outcomes are shown in boxes.
- ‡May be performed locally as a rapid screen to assess severity.

## Abbreviations

ALT = alanine transaminase

AST = aspartate transaminase

CK = creatine kinase

CXR= chest x-ray

ECHO = echocardiagram

EKG = electrocardiogram

GAA = acid alpha-glucosidase

Hex4 = hexose tetrasaccharide, also called glucotetrasaccharides

LDH = lactate dehydrogenase

Disclaimer: This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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 clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.