

Dear Doctor:

Your patient is exploring the option of ordering a genetic test for Pompe disease.

The Invitae Pompe Disease Test analyzes the GAA gene, which is the only known gene to cause Pompe disease (also called glycogen storage disease, type II [GSD II]).

This test is intended for any individual, child or adult, who has an abnormal newborn screen for Pompe; a suspected clinical diagnosis of Pompe that is based on the clinical findings of significant hypotonia, cardiomegaly, and hepatomegaly in early infancy; or progressive muscle weakness (especially in a limb-girdle pattern) and respiratory difficulties. Further, any individual with prior low acid alpha-glucosidase (GAA) enzyme activity must undergo molecular analysis for Pompe disease.

Test specifications:

Turnaround time	10–21 days
Sample requirements	Saliva, buccal, or blood. Saliva and buccal collection kits can be sent directly to your patient to assist with remote consultations.
Family variant testing	Genetic test results can impact the risk of disease in family members. This may allow for prevention or early detection of genetic conditions. Invitae offers family variant testing for blood relatives of a patient who was tested at Invitae for which a pathogenic or likely pathogenic variant was found.
Genetic support	Have questions about testing? Our experts are available to help with testing strategy, result interpretation, and case-specific clinical information. Learn more at www.invitae.com/clinical-support-services .
Laboratory accreditation	CLIA/CAP clinical accreditation

By visiting Invitae's website, you and your patient can learn valuable information about our tests and services including:

- What are appropriate clinical indications for genetic testing?
- How can genetic testing be used to guide medical management?
- What type of family history information is most important to collect?

For more details about the Invitae Pompe Disease Test, visit:
www.invitae.com/pompe.

Questions about the testing, ordering, or billing process? Contact apac@invitae.com.

We are committed to ensuring that you have the right tools to provide your patients with the answers they need.