

Pompe Disease

Parent Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

Disorder Indicated: Pompe Disease is a Lysosomal Storage Disorder (LSD) where the body lacks sufficient activity of the lysosomal enzyme acid alpha-glucosidase (GAA), caused by mutations in the GAA gene, and results in accumulation of glycogen in the lysosomes resulting in swelling, cell damage, and progressive organ dysfunction. There are several forms of Pompe Disease that vary in severity, onset of symptoms, and outcomes.

Incidence		1 in every 40,000 newborns.	
Signs and Symptoms may include: <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	Infantile Onset Pompe Disease (IOPD) <i>Generally symptomatic prior to one year</i>		Late-Onset Pompe Disease (LOPD) <ul style="list-style-type: none"> • Onset after first year through late adulthood • Progressive weakness • May not be clinically significant until juvenile or adult • Respiratory complications • Associated cardiac issues • Significant decrease in quality of life
	Classic	Non-Classic	
	<ul style="list-style-type: none"> • Low muscle tone • Enlarged heart • Failure to thrive • Respiratory infections • Hearing Loss • Most severe 	<ul style="list-style-type: none"> • Low muscle tone • Muscle weakness • Failure to thrive • Respiratory infections • Hearing Loss 	
Next Steps <i>may</i> include:	Follow up with your child’s pediatric provider Clinical assessment Laboratory Testing		
Treatment <i>(if indicated)</i>	Follow up with your child’s pediatric provider and Genetic Specialist		
Additional Resources	VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org Genetics Home Reference https://ghr.nlm.nih.gov/ International Pompe Association http://worldpompe.org/		

Educational content adapted from www.babysfirsttest.org

