



# Lysosomal Acid Lipase Deficiency (LAL-D) and your health

This guide will help you

- Track the signs and symptoms of LAL-D
- Talk about your lab tests with your doctor

## WHAT IS LAL-D?

LAL-D is a rare, genetic, progressive condition in which infants, children, and adults have an uncontrolled buildup of fatty material (cholesterol esters and triglycerides) in their liver, intestines, blood vessel walls, and other tissues. This buildup can cause continuous damage that may affect the function of many organs throughout your body.

## WHAT ARE THE RISKS OF HAVING LAL-D?

You're probably already aware that LAL-D is a serious, chronic, life-threatening disease that can damage vital organs such as your liver, spleen, stomach and intestines, and cardiovascular system.

When seen in infants, LAL-D is rapidly progressive, resulting in severe complications within the first year of life. When seen in children and adults, LAL-D can lead to serious health problems that can occur at any time without warning.

Most people with LAL-D experience complications in more than one organ system. However, no single sign, symptom, or lab test can give a complete picture of your LAL-D.

## TALK TO YOUR DOCTOR ABOUT

- Your symptoms of LAL-D (old, new, or different from your last visit)
- The frequency and importance of regular doctor's visits
- Different ways to help manage your LAL-D

Keeping track of how LAL-D is affecting your body and writing down what happens over time may help you and your doctor manage your LAL-D.

## TRACKING THE SIGNS AND SYMPTOMS OF LAL-D FOR YOUR NEXT DOCTOR'S VISIT

Here you will find assessment forms for recording your signs and symptoms of LAL-D. In addition to tracking your signs and symptoms, record your lab test results on the following pages to get a better picture of your LAL-D. By sharing this information, you and your doctor can better understand how well your LAL-D is being managed.

**Contact your doctor or get medical help right away if you are experiencing a medical emergency.**

SYMPTOM	DO YOU HAVE THE SYMPTOM NOW?		NOTES
	Yes	No	When did symptom start?/How long?/How often?
Distended abdomen (large belly size)	Yes	No	
Stomach pain and/or cramps	Yes	No	
Nausea	Yes	No	
Frequent vomiting	Yes	No	
Frequent/watery diarrhea	Yes	No	
Constipation	Yes	No	
Easy bruising	Yes	No	
Fever	Yes	No	
Tiredness	Yes	No	
Yellow discoloration of skin	Yes	No	
Frequent nosebleeds	Yes	No	
Other	Yes	No	

## TRACKING YOUR LABS WITH YOUR DOCTOR

Please note that not everyone with LAL-D will undergo all of these tests. Check with your doctor about what tests are right for you and any other important lab test results that may not be listed here.

LABS AND WHAT THEY EVALUATE	TEST	RESULTS (LOW/NORMAL/HIGH)	DATE
<b>Lipid panel</b>			
Fat in blood (including “bad cholesterol” and “good cholesterol,” which assess risk of heart disease)	Low-density lipoprotein cholesterol (LDL-c)		
	High-density lipoprotein cholesterol (HDL-c)		
	Triglycerides (TGs)		
	Total cholesterol (TC)		
<b>Complete blood count</b>			
Infections, inflammation	White blood cells (WBCs)		
Bleeding, bruising, anemia, tiredness	Red blood cells (RBCs)		
	Hematocrit		
	Hemoglobin		
Blood clotting	Platelets/mean platelet volume (MPV)		
<b>Comprehensive metabolic panel</b>			
Proteins	Total protein		
	Albumin		
Liver function	Alanine aminotransferase (ALT)		
	Aspartate aminotransferase (AST)		
	Bilirubin		

OTHER LABS AND EVALUATIONS	TEST	RESULTS (LOW/ NORMAL/HIGH)	DATE
Growth	Height-for-age percentile		
	Weight-for-age percentile		
Bleeding problems	Prothrombin time/international normalized ratio (PT/INR), partial thromboplastin time (PTT)		
Biomarker (a marker found in your blood that indicates activation of certain immune cells)	Chitotriosidase (ChT, a macrophage inflammatory marker)		
Fat in liver (on ultrasound, magnetic resonance imaging [MRI], imaging technology for fibrosis, or biopsy)	Fatty liver (hepatic steatosis)		
Fat in liver under the microscope (found on biopsy only)	Microvesicular hepatic steatosis		
Scarring/damage in liver (on imaging technology for fibrosis or biopsy)	Fibrosis		
	Cirrhosis		
Enlarged liver	Hepatomegaly		
Enlarged spleen	Splenomegaly		
Hardening of the adrenal glands above the kidneys (on X-rays)	Adrenal calcifications		
Cardiac function	Echocardiogram		
	Electrocardiogram (EKG or ECG)		

Discuss any important test results with your healthcare team.





To learn more about LAL-D, visit [LALDSOURCE.COM](http://LALDSOURCE.COM)  
or follow LAL-D Source on Facebook.

---

Join the LAL Deficiency registry to help physicians learn more about people with LAL-D.  
To learn more, visit [LALDEFICIENCYREGISTRY.COM](http://LALDEFICIENCYREGISTRY.COM).

**ALEXION**<sup>®</sup>



Alexion is a registered trademark of Alexion Pharmaceuticals, Inc.  
Copyright © 2016, Alexion Pharmaceuticals, Inc. All rights reserved. GL/UNB-LAL/16/0025