THE LAL DEFICIENCY REGISTRY

THE FIRST GLOBAL REGISTRY FOR LYSOSOMAL ACID LIPASE DEFICIENCY (LAL-D)

The LAL Deficiency registry is dedicated to helping physicians improve management of LAL-D

LAL-D registry

LAL-D IS A LIFE-THREATENING GENETIC DISEASE WITH ONGOING, PROGRESSIVE, MULTIORGAN DAMAGE LEADING TO PREMATURE DEATH[1]

Patients with LAL-D are missing a vital enzyme, and experience uncontrolled accumulation of cholesteryl esters and triglycerides that leads to systemic complications[1]

Multiorgan damage—a devastating consequence of LAL-D[1,2]

ORGAN SYSTEM	SYMPTOMS MAY INCLUDE
Liver manifestations: 86%[1,a]	
	• Hepatomegaly[1] • Increased hepatic fat content[1] • Transaminase elevation signaling chronic liver injury[1] • Progression to fibrosis and cirrhosis[1] • Complications of end-stage liver disease[1]
Cardiovascular manifestations: 87%[1,a]	
	• Dyslipidemia[1] • Increased risk of cardiovascular disease[1] • Increased risk of accelerated atherosclerosis[1]
Gastrointestinal (GI) manifestations: 22%[1,a]	
	• Malabsorption[1] • Growth failure[1]
Spleen manifestations: 36%[1,a]	
	• Splenomegaly[1] • Anemia[1] • Thrombocytopenia[2]

LAL-D affects patients of all ages, as clinical complications manifest from infancy through adulthood. In infants, LAL-D is rapidly progressive and fatal, with nearly 90% mortality within 12 months after birth.[1,3]

In an observational study, approximately 50% of pediatric and adult patients with LAL-D with a clinical biopsy assessment progressed to fibrosis, cirrhosis, or liver transplant within 3 years of symptom onset.[4,b]

LEARN MORE—for more information, please visit LALDSOURCE.com.

[a]Based on an analysis of 55 genotyped patients with LAL-D in a cohort of 135 cases.[1]
[b]Based on modeling, using a subset of 31 patients (≥5 years of age) in an observational study who received a liver biopsy, and 1 additional patient with no biopsy who received a liver transplant. Patients selected by their clinician for liver biopsy are expected to have more evidence of disease progression than patients with LAL-D overall.[4]

THE LAL DEFICIENCY REGISTRY: ESTABLISHING A LONG-TERM DATABASE TO ENHANCE UNDERSTANDING OF THIS RARE DISEASE

Your participation is key to increasing knowledge of LAL-D

- All physicians managing patients with LAL-D can participate
- Benefits of registry participation include
  - Global collaboration with other physicians dedicated to this chronic, systemic, progressive, life-threatening disease
  - Access to regular feedback with registry updates and findings
  - Contribution to safety monitoring
  - Increased knowledge to better manage the challenges that patients with LAL-D face
- Registry enrollment is open to any patient with a diagnosis of LAL-D (by enzyme activity or DNA sequencing)
- For the LAL Deficiency registry to be representative of the whole patient population, all eligible patients should participate

Why is it important?

- To assess the long-term consequences of LAL-D due to missing lysosomal acid lipase, a vital enzyme[1]
- To identify other clinical outcomes, including mortality and morbidities, in patients with LAL-D[1]
- As with registries for other rare diseases, the LAL Deficiency registry helps researchers and physicians raise awareness of signs and symptoms of LAL-D and discover ways to advance treatment

While participation in the LAL Deficiency registry is voluntary, obtaining the most robust and representative data sets possible helps physicians better understand and treat this disease.

REGISTRY ENROLLMENT IS OPEN TO ANY PATIENT WITH LAL-D, REGARDLESS OF TREATMENT STATUS

- Participation in the LAL Deficiency registry requires approval from your institutional review board or ethics committee
- Data are submitted to the registry on electronic case report forms via a secure web-based system and are reviewed for quality before inclusion in aggregate analyses available to all registry physicians
- Patient authorization is required; patients are identifiable only to the patient's physician via a unique ID code

HOW TO ENROLL YOUR PATIENTS IN THE REGISTRY

- Contact your local Medical Affairs representative or email **laldregistry@alxn.com** to discuss participation in the registry
- Complete site startup process to become an active registry user
- Visit **LALDEFICIENCYREGISTRY.COM/ENROLL** to access the electronic data capture system and enroll your patients
- You will be responsible for the patient data that are submitted to the registry
  - You may add other team members to your registry site—those who are directly involved in the care of your patients and/or those who will be entering data

FOR MORE INFORMATION, VISIT
LALDEFICIENCYREGISTRY.COM

References: 1. Bernstein DL, et al. Cholesteryl ester storage disease: review of the findings in 135 reported patients with an underdiagnosed disease. *J Hepatol.* 2013;58:1230-43. doi:10.1016/j.jhep.2013.02.014. **2.** Hoffman EP, et al. Lysosomal acid lipase deficiency. In: Pagon RA, et al, eds. *GeneReviews.* Seattle, WA: University of Washington; 2015. http://www.ncbi.nlm.nih.gov/books/NBK305870/. Accessed April 28, 2016. **3.** Jones SA, et al. Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants [published online August 27, 2015]. *Genet Med.* doi:10.1038/gim.2015.108. **4.** Data on file, Alexion Pharmaceuticals.

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