GI MANIFESTATIONS AND GROWTH FAILURE PROMPT A BATTERY OF TESTS THAT DELAY DIAGNOSIS[1]

CASE REPORT: Male infant patient with Lysosomal Acid Lipase Deficiency (LAL-D)[1]

Based upon a published case report:
Sadhukhan M, et al. *BMJ Case Rep*. 2014. doi:10.1136/bcr-2013-202652.

YEARS OF AGE

8 DAYS

INITIAL PRESENTATION
- Watery diarrhea

2 WEEKS

PATIENT WAS HOSPITALIZED:
- Poor weight gain
- Vomiting
- Diarrhea

LIVER FUNCTION, RENAL FUNCTION, FULL BLOOD COUNT:
- AST 44 U/L (considered mildly elevated)[a]
- All other results normal
- Hgb 14 g/dL (considered normal)

INITIAL DIAGNOSTIC CONSIDERATION:
- Protein intolerance to cow's milk

INITIAL MANAGEMENT:
- Amino acid–based formula
- Patient was discharged following symptom improvement

5 WEEKS

PATIENT WAS HOSPITALIZED:
- Continued vomiting
- Gluteal wasting
- Poor weight gain
- Hepatosplenomegaly

LIVER FUNCTION, RENAL FUNCTION, FULL BLOOD COUNT:
- AST 75 U/L[a]
- All other results remained normal
- Hgb 9 g/dL

IMAGING RESULTS:
- No evidence of adrenal gland calcification on abdominal X-ray
- Hepatosplenomegaly confirmed on ultrasound

SUPPORTIVE MANAGEMENT/INTERVENTIONS:
- Started on antacid medication for possible reflux
- Feeding through nasogastric tube

DIAGNOSTIC CONSIDERATIONS:
- Metabolic condition due to hepatosplenomegaly—normal preliminary screen
- Cystic fibrosis

PATIENT TRANSFERRED TO TERTIARY CENTER FOR FURTHER METABOLIC WORKUP

10 WEEKS

LABORATORY RESULTS:
- Vacuolated lymphocytes and plasma chitotriosidase was markedly elevated indicating macrophage activation, suggestive of an underlying metabolic storage disorder

ADDITIONAL DIAGNOSTIC CONSIDERATIONS:
- Metabolic storage disorder (Niemann-Pick, Gaucher)
- Familial immune disorder (familial HLH)

LAL-D DIAGNOSIS
- Low LAL activity in enzyme-based blood test
- Sequencing of *LIPA* revealed homozygous mutation

SUPPORTIVE MANAGEMENT:
- Nasogastric tube feeding with amino acid–based formula and energy-rich, carbohydrate-based formula
- Weight gain remained suboptimal; patient received blood transfusion for anemia

12 WEEKS

PATIENT TRANSFERRED TO ANOTHER HOSPITAL FOR ADDITIONAL TREATMENTS

SUPPORTIVE MANAGEMENT:
- Modular low-fat feed via gastrostomy
- Enzyme replacement therapy

[a]Normal values: AST, 32-162 U/L (0-14 days), 20-67 U/L (15 days-<1 year).[1,2]

Abbreviations: AST, aspartate aminotransferase; GI, gastrointestinal; Hgb, hemoglobin; HLH, hemophagocytic lymphohistiocytosis; LAL, lysosomal acid lipase.

KEY TAKEAWAY

- LAL-D presenting in infants is a **medical emergency** and, if left untreated, leads to **imminent death**[3]

LAL-D IS RAPIDLY FATAL IN INFANTS—LEARN TO RECOGNIZE LAL-D AND AVOID MISDIAGNOSIS[3]

Devastating consequences of LAL-D in infants

Infants with LAL-D experience early and severe symptom onset.[3]

- 43% of patients experience symptom onset before 1 month of age[3]

Some of the most notable signs and symptoms of LAL-D in infants include

- Prominent hepatosplenomegaly[3]
- Adrenal calcification (seen in about 50% of patients)[1,3]
- Growth failure[3]

With many devastating multiorgan manifestations, a multidisciplinary team approach can expedite diagnosis of LAL-D.[4,5]

LAL-D IN INFANTS IS FATAL—RAPID DIAGNOSIS IS CRITICAL[3]

- Test for LAL-D with an enzymatic DBS test[4,6]

Abbreviation: DBS, dried blood spot.

References: 1. Sadhukhan M, et al. Infant case of lysosomal acid lipase deficiency: Wolman's disease. *BMJ Case Rep*. 2014;2014:bcr2013202652. doi:10.1136/bcr-2013-202652. **2.** Colantonio DA et al. Closing the gaps in pediatric laboratory reference intervals: a CALIPER database of 40 biochemical markers in a healthy and multiethnic population of children. *Clin Chem*. 2012;58:854-68. doi:10.1373/clinchem.2011.177741. **3.** Jones SA, et al. Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. *Genet Med*. 2016;18:452-8. doi:10.1038/gim.2015.108. **4.** Reiner Ž, et al. Lysosomal acid lipase deficiency—an under-recognized cause of dyslipidaemia and liver dysfunction. *Atherosclerosis*. 2014;235:21-30. **5.** 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. **6.** Hamilton J, et al. A new method for the measurement of lysosomal acid lipase in dried blood spots using the inhibitor Lalistat 2. *Clin Chim Acta*. 2012;413:1207-10. doi:10.1016/j.cca.2012.03.019.

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