

Pediatric Patient Case Study

Initial Presentation: Respiratory virus and enlarged abdomen

ALEX

"There were no alarms going off to suspect something was wrong."

-ALEX'S MOM

Patient's name, photo, and other identifying details have been changed to protect anonymity.



ALEX'S JOURNEY

13
MONTHS

6+
HCPs

4+
SUSPECTED
CONDITIONS

- Asthma
- Acid reflux
- Lymphoma
- Leukemia

7
TESTS

SEE ANY COMBINATION OF THESE SYMPTOMS?

Hepatomegaly?

Pulmonary involvement?

Splenomegaly?

Thrombocytopenia?

Gastrointestinal issues?

IT'S NOT WHAT YOU THINK...

Would you recognize this progressive, genetic disease?

Know the signs. Enable early diagnosis and symptom management.

To learn more, visit ASMDfacts.com

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IT STARTED EARLY

At a young age, Alex exhibited unexplained gastrointestinal (GI) and abdominal symptoms and his mother had concerns about him meeting developmental milestones. Alone, these signs might appear isolated, but considered together, they are among the hallmark signs of a progressive, genetic disease.

Nondescript signs and inaction

From the very beginning of his life, Alex experienced respiratory complications and GI-related issues including labored breathing, constant congestion, and abdominal bloating.

3 WKS EMERGENCY ROOM



SYMPTOM

Aspiration during diaper change prompted Alex's mother to bring him to the ER



DIAGNOSIS

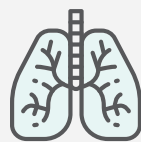
Acid reflux



SYMPTOM MANAGEMENT

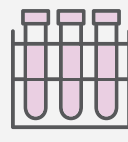
The ER doctor suggested that Alex be kept upright 20 min after eating

4 MOS PEDIATRICIAN



SYMPTOM

A barking cough drove Alex's mother to take him to his pediatrician



DIAGNOSIS

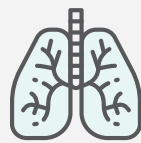
Alex was tested for respiratory syncytial virus (RSV), and the test returned a positive result



SYMPTOM MANAGEMENT

Alex's pediatrician prescribed medication to help manage his RSV

8-9 MOS URGENT CARE



SYMPTOM

Consistent congestion and labored breathing prompted Alex's mother to visit urgent care



REFERRAL

Concerned with the sound of Alex's lungs, the urgent care doctor referred Alex to the ER. The ER doctor ordered a chest x-ray

EMERGENCY ROOM (ER)



DIAGNOSIS

ER doctor suspected, but couldn't conclusively diagnose asthma due to Alex's age. Based on radiologic findings and symptomology, Alex was instead diagnosed with an upper respiratory tract infection, bronchospasm, and reactive airway disease



SYMPTOM MANAGEMENT

ER doctor prescribed medication to help manage his respiratory symptoms

Unexplained symptoms

Alex maintained a daily routine and a healthy diet but his mother was concerned he that he could be missing other key milestones in his development.

9-12 MOS PEDIATRICIAN



SYMPTOMS

Due to lack of weight gain and delayed walking, Alex's mom had questions about her child's mobility and took him to see his pediatrician



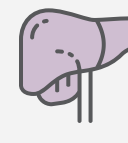
NO DIAGNOSIS

The doctor raised no concerns, explaining that metabolism may differ from child to child, and boys tend to take longer to walk than girls

One step closer

Alex was scheduled for a minor urological procedure, but during the pre-operative examination, the anesthesiologist noticed Alex's abdomen was protruding.

13 MOS SURGERY CENTER



SYMPTOM

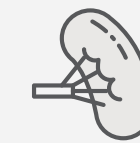
During preparation for a hydrocele surgery, the anesthesiologist noticed Alex's belly looked full, so he pressed on it and discovered his liver was enlarged



REFERRAL

The anesthesiologist informed the urologist, who canceled the surgery and referred the family to the local hospital

HOSPITAL



TEST RESULTS

At the hospital, a CT scan, MRI, X-ray, and ultrasound of his abdomen were performed. In addition to his liver, findings revealed his spleen was significantly enlarged



DIAGNOSIS

A hematologist-oncologist was consulted, who suspected the cause of the enlarged liver and spleen might be leukemia or lymphoma

Confirmed Diagnosis: ASMD

The hematologist-oncologist referred Alex to a gastroenterologist at the hospital who performed a liver biopsy. They initially believed it was a glycogen storage disorder, but a blood test confirmed that it was a lysosomal storage disorder historically known as Niemann-Pick disease types A, A/B, and B, also called ASMD.

4 YRS



How Alex is doing now...

Alex is currently 4 years old and learning to live with ASMD. He loves going to school, swimming, and he regularly sees his pediatrician and a multidisciplinary team of specialists who monitor and help manage his symptoms.

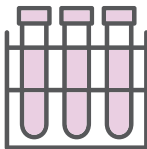
ASMD

DIAGNOSTIC TESTING IS SIMPLE

ASMD can be diagnosed with one blood test.

While Alex's story may sound atypical, there is no one story for patients with **acid sphingomyelinase deficiency (ASMD)**. Clinical manifestations, severity of signs and symptoms, rate of progression, and patient age at symptom onset can vary. As illustrated by Alex's journey, signs and symptoms of ASMD are similar to those of other, regularly considered conditions—which is why misdiagnosis is common.

One test can make a difference.



An accurate ASMD diagnosis is essential for an appropriate symptom management plan. The recommended method for diagnosing ASMD is a blood test to measure the amount of ASM enzyme activity. A diagnosis of ASMD can be confirmed if the test shows decreased ASM enzyme activity.

ASMD is an autosomal recessive condition, so there is potential risk of inheritance. If a family member has been diagnosed with ASMD, relatives should consider talking with their doctor about family screening.

Early diagnosis of ASMD is a priority for symptom management.
For more information on ASMD and testing, visit [ASMDfacts.com](https://www.asmdfacts.com).