Adult Patient Case Study
Initial Presentation: Gastrointestinal issues and bone pain

NITA

"I lived with uncertain symptoms for years without a diagnosis."

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

NITA'S JOURNEY

25+
YEARS

15+
HCPs

10+
TESTS

9
SUSPECTED CONDITIONS

- Mononucleosis
- Splenomegaly
- Cytomegalovirus
- Cancer
- Depression
- Iron Deficiency
- Peripheral Neuropathy
- Arthritis
- Gaucher Disease

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

Ever since she was a child, Nita encountered multisystemic symptoms such as gastrointestinal issues and bone pain with little-to-no explanation. It wasn't until after she gave birth to her second son, however, that she started to really question the spectrum of symptoms she was experiencing. Considered alone, the symptoms Nita was experiencing may appear isolated, but considered together they are among the hallmark signs of a progressive, genetic disease.

Progressive weakness and an unbreakable fever

Symptoms during childhood such as rectal bleeding and constant bone pain plagued Nita. Her symptoms limited her physical activity, but it wasn't until a tumultuous pregnancy with her second son at age 25 that her diagnostic journey truly began.

25 YRS — **HOSPITAL**

SYMPTOMS

Nita developed a fever five days after giving birth. Her symptoms increased to the point where she felt so weak she could not lift her newborn son or stand up to walk. She was readmitted to the hospital for 10 additional days.

DIAGNOSIS & REFERRAL

During an ultrasound, her obstetrician-gynecologist discovered an enlarged spleen and suspected mononucleosis. The test for mononucleosis was positive, but she was subsequently also shown to have cytomegalovirus, and an infectious disease specialist was brought in to manage the virus.

An isolated path

Still uncertain about the source of her symptoms and why her pain kept persisting, Nita searched the internet for possible causes of her enlarged spleen. She developed a list and started crossing off items as her doctors tested for them.

27-31 YRS — **DAILY ROUTINE**

SYMPTOMS

Continued pain, fever, nausea, shortness of breath, and fatigue left Nita feeling exhausted and drove her to stop both working and volunteering in her community.

IMPACT

Ultimately, this uncertain disease path increased stress on her social life and relationships.

New symptoms lead to new questions

At 31, Nita began developing new symptoms, such as hand tremors. After a referral to a neurologist, she was diagnosed with an iron deficiency and prescribed iron infusions, but still her hand tremors continued.

31-32 YRS — **HEMATOLOGIST-ONCOLOGIST AND PATHOLOGIST** — **HOSPITAL**

SYMPTOMS

Still uncertain about Nita's symptoms and why she continued to be in such pain, her hematologist-oncologist conducted an ultrasound of Nita's spleen. Concerned that her spleen might rupture if left alone, the hematologist-oncologist concluded that a splenectomy was needed, and referred her to a surgeon.

PATHOLOGY & SUSPICION

After Nita's spleen was removed, the pathologist performing tests on the spleen suspected that a lysosomal storage disorder might be the cause of her splenomegaly and other multisystemic symptoms.

REFERRAL

After consulting with the pathologists, the hematologist-oncologist referred her to a larger hospital system with more comprehensive care management resources so Nita could have subsequent tests and procedures as necessary.

REFERRAL & TESTING

The hematologist-oncologist at the new hospital referred Nita to a geneticist. Based on her symptomology and medical history, the geneticist suspected Gaucher disease type 1, however the **β**-glucosidase enzyme assay routinely used to test for Gaucher disease came back negative.

Confirmed Diagnosis: ASMD

While Nita's **β**-glucosidase enzyme assay for Gaucher disease returned a negative test result, the geneticist was aware of another condition that shares significant phenotypic overlap with Gaucher disease type 1: a lysosomal storage disorder called ASMD (historically known as Niemann-Pick disease types A, A/B, and B). The geneticist tested Nita for ASMD using an ASM enzyme assay, and the result came back positive.

42 YRS

How Nita is doing now...

Since her diagnosis, Nita has become an advocate for other ASMD patients, speaking to raise awareness of ASMD; she is occasionally joined by her sons who offer their perspective of their mother's journey and the importance of testing early. Nita is passionate about telling her story in an effort to bring hope to other ASMD patients navigating uncertain diagnostic journeys.

ASMD

DIAGNOSTIC TESTING IS SIMPLE

ASMD can be diagnosed with one blood test.

While Nita'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 Nita'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.

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