THINK OF ASMD (Niemann-Pick Disease A, A/B and B) IN YOUR DIFFERENTIAL DIAGNOSIS

There is an overlap of symptoms with GAUCHER DISEASE[1]

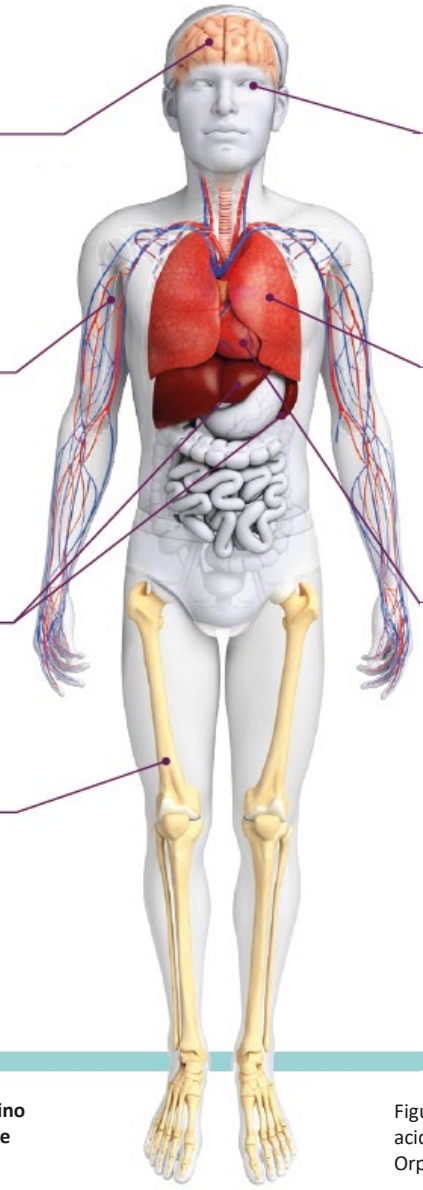
COMMON SIGNS AND SYMPTOMS[2]

NEUROLOGICAL MANIFESTATIONS
- No neurodegeneration
- Range from mild hypotonia/hyporeflexia to loss of motor function, cognitive impairment

HEMATOLOGIC ABNORMALITIES
- Thrombocytopenia
- Anemia and leukemia
- Easy bruising and excessive bleeding

SPLENOMEGALY / HEPATOMEGALY
- Liver fibrosis
- Liver dysfunction (elevated ALT and AST)

SKELETAL DISEASE
- Back, limb or joint pain
- Osteopenia and osteoporosis
- Skeletal fractures
- Growth delay in adolescents
- Decreased BMC and BMD in pediatric patients



EYES
- Macular cherry-red spots

PULMONARY DISEASE
- Interstitial lung disease
- Respiratory infections

CARDIAC DISEASE
- Cardiac valve disease
- Mixed dyslipidemia early in disease course
- Coronary artery disease

OTHER
- Headache
- Recurrent ear infections
- Delayed growth and puberty
- Fatigue

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ASMD – Acid Sphingomyelinase Deficiency; ALT – Alanine Amino Transferases; AST – Aspartate Amino Transferase; BMC – Bone Mineral Content; BMD – Bone Mineral Density

Figure adapted from: Disease manifestations and burden of illness in patients with acid sphingomyelinase deficiency (ASMD) by McGovern, Margaret M., et al., 2017, Orphanet Journal of Rare Diseases

ASMD
ACID SPHINGOMYELINASE DEFICIENCY

DIAGNOSIS OF **ASMD** IN PATIENTS WITH **SPLENOMEGALY** AND/OR HEPATOMEGALY

EARLY DIAGNOSIS IS CRITICAL

to successful management of the disease[1]

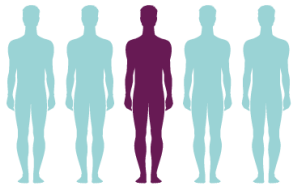
SUSPECT ASMD IN CASE OF (any combination)[2]

>90% of patients have splenomegaly

>80% of patients have interstitial lung disease

>70% of patients have hepatomegaly

>50% of patients have thrombocytopenia



OF 5 PEOPLE YOU SUSPECT OF HAVING GAUCHER DISEASE, ~1 HAS ASMD[3]

Because common symptoms of Gaucher disease and ASMD overlap.[1]

PARALLEL TESTING

for Gaucher disease and ASMD[1]

ASMD

- Lysosomal storage disease with a singular cause: enzyme deficiency (acid sphingomyelinase) leading to accumulation of sphingomyelin[2]
- Serious and progressive disease with autosomal recessive inheritance[2]
- Affects both pediatric and adult patients[2]
- Potentially life threatening and often leads to premature death[2]

DIAGNOSTIC TESTING FOR ASMD IS SIMPLE AND STRAIGHTFORWARD[1]

1. **ENZYME ACTIVITY TESTING** ASM — DBS OR Whole blood sample
2. **GENOTYPING** SMPD1-gene — DBS OR Whole blood sample
3. **CONFIRMATION OF ENZYME ACTIVITY ON LEUCOCYTES** — Whole blood sample

McGovern, Margaret M., et al. "Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency." Genetics in Medicine 19.9 (2017): 967. 2. McGovern, Margaret M., et al. "Disease manifestations and burden of illness in patients with acid sphingomyelinase deficiency (ASMD)." Orphanet journal of rare diseases 12.1 (2017): 41.3
3. Oliva P, Mechtler TP, Schwarz M, et al. Differential diagnosis of Niemann-Pick A/B disease (ASMD) in cases of suspected Gaucher disease. March 2021. https://www.archimedlife.com/incidence-of-asmd-in-suspected-gaucher-patients/