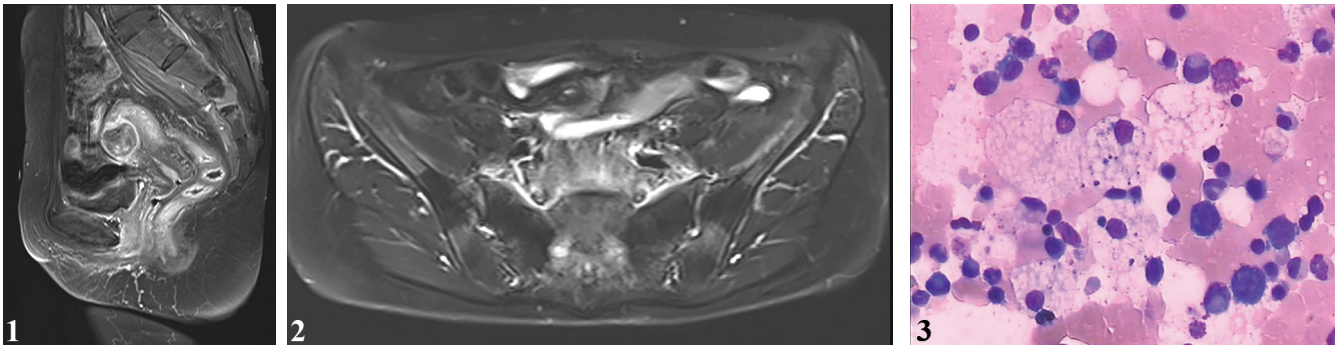


# Unusual Cause of Abdominal Pain: Niemann-Pick Disease

Dehui Yu, Hu Lin, Qingyuan Huang

Department of Traditional Chinese Medicine, Meizhou People's Hospital, Meizhou Academy of Medical Sciences, Meizhou, Guangdong Province, China



A 54-year-old woman was referred to our hospital for left upper abdominal pain that had been recurring with sudden onset and termination for more than 2 months and accompanied by nausea and vomiting. Gastroscopy at another hospital revealed no peptic ulcer, malignant tumor, or pyloric obstruction. However, head magnetic resonance imaging (MRI) revealed brain atrophy. The patient also had a history of anxiety disorder and depression. Computed tomography showed no enlargement of the liver or spleen. However, abnormal changes in the third sacral vertebral body were observed, which was confirmed as a pathological fracture on MRI (Fig. 1). MRI further revealed abnormal bone signal changes in the thoracic and lumbar vertebrae and pelvis (Fig. 2). Bone marrow aspiration biopsy revealed numerous Niemann-Pick cells (Fig. 3). After further genetic testing, the patient was diagnosed with Niemann-Pick disease type C (NPC). Ambulatory electroencephalography monitoring showed diffuse epileptic discharges. The cause of abdominal pain was abdominal epilepsy. The patient was ultimately diagnosed with Niemann-Pick disease secondary to abdominal epilepsy. Antiepileptic treatment with oral phenytoin sodium resolved her abdominal pain. No recurrence was observed throughout more than 3 years of follow-up.

NPC is a rare autosomal recessive genetic lysosomal storage disorder mainly affecting the nervous system and internal organs [1]. Because it affects the brain, it can also cause mental

health disorders. The clinical manifestations of NPC are diverse [2, 3]. Some patients may exhibit focal or generalized epileptic seizures, while others may have anxiety disorder, depression, schizophrenia, bipolar disorder, or other psychiatric symptoms alone or in combination. In this report, we describe a case of Niemann-Pick disease in which the latest presentation was abdominal pain initially of unknown cause.

**Corresponding author:** Qingyuan Huang, [hqy33005@163.com](mailto:hqy33005@163.com)

**Conflicts of interest:** None to declare.

## REFERENCES

1. Hosseini K, Fallahi J, Razban V, et al. Overview of clinical, molecular, and therapeutic features of Niemann-Pick disease (types A, B, and C): Focus on therapeutic approaches. *Cell Biochem Funct.* 2024;42:e4028. doi:[10.1002/cbf.4028](https://doi.org/10.1002/cbf.4028)
2. Tirelli C, Rondinone O, Italia M, et al. The genetic basis, lung involvement, and therapeutic options in Niemann-Pick disease: A comprehensive review. *Biomolecules.* 2024;14:211. doi:[10.3390/biom14020211](https://doi.org/10.3390/biom14020211)
3. Fu CL, Diacovo MJ. Niemann-Pick disease with isolated leukemic nonnodal mantle cell lymphoma of the spleen. *Blood.* 2020;135:778. doi:[10.1182/blood.2019003479](https://doi.org/10.1182/blood.2019003479)