Pediatr Radiol (2002) 32: 674–676
DOI 10.1007/s00247-002-0761-0

CASE REPORT

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# Mesenteric mass in a young girl – an unusual site for Gaucher's disease

Received: 1 January 2002
Accepted: 8 April 2002
Published online: 10 July 2002


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**Abstract** We report the first case of a child with Gaucher's disease and a large mesenteric mass, confirmed histologically to be Gaucher's cell infiltrates. We describe the radiological findings and discuss further management. The advent of enzyme replacement therapy has prolonged survival and the emergence of previously undocumented manifestations of the disease is being observed. The radiologist and clinician should be alert to the possible development of these new problems and the fact that in Gaucher's disease a palpable right upper-quadrant mass need not necessarily represent hepatomegaly.

**Keywords** Gaucher's disease · Mesenteric mass · Child

## Introduction

Gaucher's disease is an autosomal recessive disease characterised by deficiency of glucocerebrosidase, a lysosomal enzyme responsible for the degradation of glucocerebroside. This results in the accumulation of glucocerebroside-laden macrophages (Gaucher's cells), which are deposited in the reticuloendothelial system throughout the body. Deposits of Gaucher's cells typically accumulate in the liver, spleen, bone marrow, lymph nodes, brain and lung [1]. We report a young girl with a collection of Gaucher's cells within the small-bowel mesentery. To the best of our knowledge, this is the first such case.

## Case report

A 13-month-old girl presented with vomiting, poor appetite and progressive abdominal distension for 1 month. She had stopped walking and on examination was pale with a distended abdomen and massive hepatosplenomegaly. She also had an oculomotor apraxia, but no other neurological symptoms of note. Routine blood tests revealed a pancytopaenia. Bone-marrow aspiration demonstrated infiltration with Gaucher's cells. The diagnosis of Gaucher's disease was confirmed by the demonstration of decreased leucocyte $\beta$-glucosidase activity.

In view of the oculomotor apraxia, a diagnosis of type III Gaucher's disease (see below) was made. Baseline investigations revealed significant pulmonary as well as skeletal involvement. She subsequently had a partial (95%) splenectomy and was started on fortnightly enzyme replacement therapy (ERT), initially alglucerase (Ceredase) and later, imiglucerase (Cerezyme). Her respiratory symptoms, fatigue and poor appetite improved. The liver decreased in size but still remained enlarged.

There was, however, progression of her skeletal manifestations of the disease. Her chitotriosidase levels, which were very high prior to commencing ERT, also decreased, but not to the levels expected. This suggested that there was a source of chitotriosidase, i.e. Gaucher's cells, that were not responding to ERT. Approximately 2 years after commencing treatment, a palpable right upper-quadrant mass was noted. Abdominal US revealed hepatosplenomegaly, but also a solid, heterogeneous 5-cm mass, anterior to the

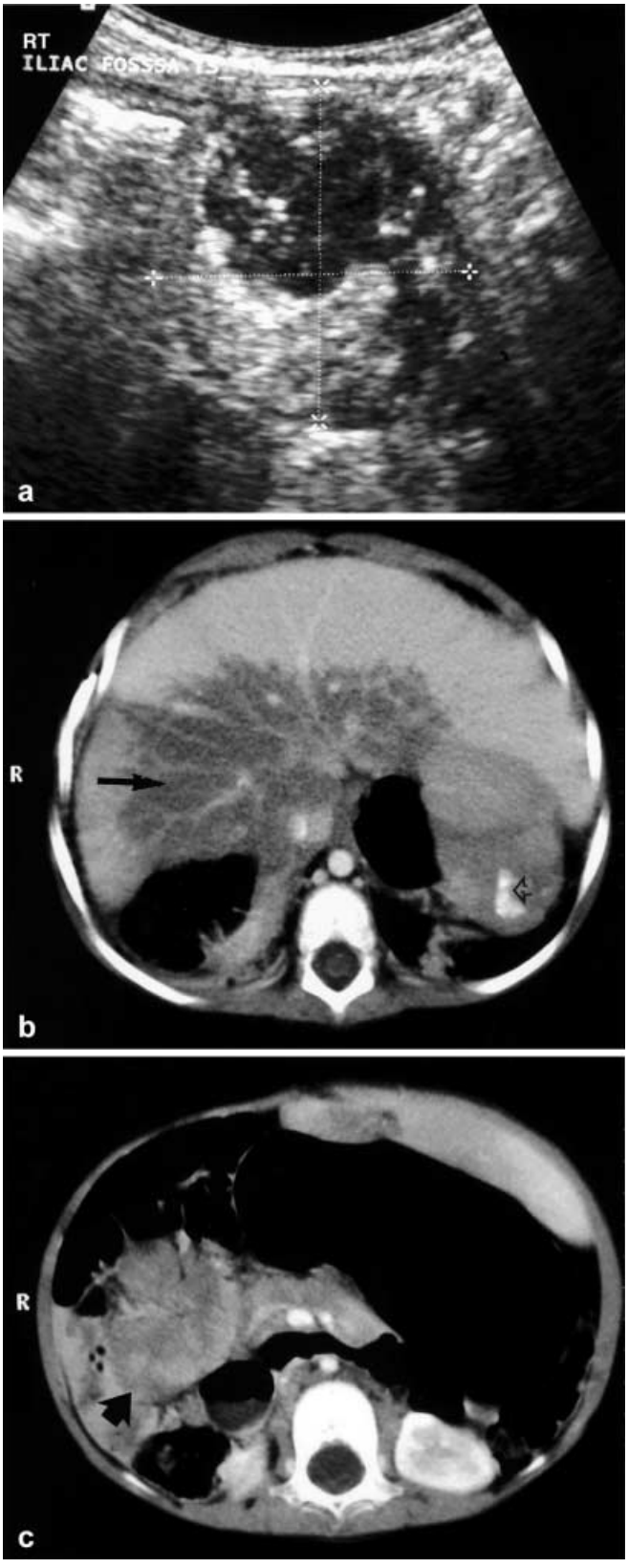


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**Fig. 1.** **a** Transverse US reveals a solid, heterogeneous 5-cm mass, which was separate from bowel and contained cystic locules inferiorly. **b** Contrast-enhanced CT demonstrates infiltration of the liver (*arrow*) and incidental splenic calcification (*arrowhead*). **c** CT also shows that the right abdominal mass is of soft-tissue density (*arrow*) and is surrounded by mesenteric vessels with matted loops of bowel inferiorly. No significant enhancement was seen

right kidney, extending from the right upper quadrant to the right iliac fossa and separate from bowel (Fig. 1a). A subsequent CT scan of the abdomen showed liver infiltration (Fig. 1b) and that the right abdominal soft-tissue density mass was surrounded by mesenteric vessels and matted loops of bowel inferiorly (Fig. 1c). No significant enhancement was seen after contrast medium administration.

Despite the patient being afebrile, an inflammatory mass or abscess was suspected, but a surgical biopsy of this mass revealed that it was a collection of lymph nodes infiltrated with Gaucher's cells at the base of the mesentery. It was felt that removal would interrupt lymphatic drainage and carry a high risk of chylous ascites. The mass was therefore left alone and remains unchanged 3 years on. Unfortunately, the skeletal involvement has progressed, with lytic lesions and medullary expansion in both humeri and proximal femora. Destruction of the petrous bones bilaterally (presumably by Gaucher's cell infiltrates, though this has not been histologically confirmed) has also been documented on subsequent CT and MRI scans.

## Discussion

Chronic Gaucher's disease is the most common lysosomal storage disorder caused by a functional deficiency of glucocerebrosidase. The glucocerebrosidase gene is located on chromosome 1 in the region of q21. Mutations of this gene, of which more than 100 have been detected, provide a spectrum of disease severity [2]. Gaucher's disease can be classified into three major types: type I spares the central nervous system (CNS), whereas the latter is involved to a variable extent in patients with types II and III disease. Type I disease is the most common form and is most frequent in Ashkenazi Jews. Types II and III, which are more severe forms of the illness, are both rare with an incidence of less than 1 in 100,000 live births. Type III is characterised by a combination of neurological and visceral disease; the preponderance of each varies considerably between patients. Most type-III patients tend to have aggressive visceral disease, and in these patients abnormal horizontal eye movements may be the sole CNS abnormality. Prior to the availability of enzyme replacement therapy, death usually occurred in adolescence owing to systemic disease. However, with the advent of enzyme replacement therapy, life expectancy has improved.

The radiographic findings of type III Gaucher's disease in children have been described by Hill et al. [3], the largest series to date, numbering 17 patients. They documented widespread systemic disease with pulmonary infiltrates, thoracic lymph node enlargement, ver-

tebral compression fractures and osteonecrosis of the long bones. The abdominal findings consisted of hepatosplenomegaly with deposits of Gaucher's cells in the liver and spleen only. No lymphadenopathy was seen radiographically, but at autopsy, three patients had generalised mesenteric lymphadenopathy. Extra-osseous extension of Gaucher's cells into soft tissues has also been described [4]. Poll et al. [5] described a middle-aged adult with a palpable epigastric mass that was inseparable from the left lobe of the liver on CT and US. Biopsy of this lesion revealed a collection of Gaucher's cells.

We report, to our knowledge, the first case of a mesenteric collection of Gaucher's cells presenting as a palpable mass and describe its imaging characteristics. The reasons for this mass developing remain unclear. It is possible that lymph nodes represent a 'sanctuary site' that is relatively impervious to intravenously administered enzyme. The natural history of such a lesion is unclear, but it has remained stable over 3 years. We postulate, however, that at this size it could lead to small-bowel intussusception and cause bowel obstruction. Resection is difficult and thus close monitoring is essential.

In the era of ERT it is possible that, concomitant with increased life expectancy, we are beginning to see the emergence of manifestations that were not observed prior to the availability of ERT. This is most likely in patients who have severe visceral disease, which include the majority of patients with type III disease. The radiologist and clinician should be alert to the possible development of these new problems and that in Gaucher's disease a palpable right upper-quadrant mass need not necessarily represent hepatomegaly.

## References


1. Lee RE, Peters SP, Glew RH (1977) Gaucher's disease: clinical, morphologic, and pathogenetic considerations. Pathol Annu 12:309–339
2. Zimran A, Gross E, West C, et al (1989) Prediction of severity of Gaucher's disease by identification of mutations at DNA level. Lancet 2:349–352
3. Hill SC, Damaska BM, Tsokos, M, et al (1996) Radiographic findings in type 3b Gaucher disease. Pediatr Radiol 26:852–860
4. Poll LW, Koch JA, vom Dahl S, et al (2000) Type 1 Gaucher disease: extra-osseous extension of skeletal disease. Skeletal Radiol 29:15–21
5. Poll LW, Koch JA, vom Dahl S, et al (1998) "Epigastric Gaucher cell pseudotumor": image morphological and histological characteristics (in German). Rofo Fortschr Geb Rontgenstr Neuen Bildgeb Verfahr 169:669–672