Sir,

A 6-year-old female child presented with gradually increasing firm lump in the left upper abdomen with abdominal distension since 3 years. The child also had history of recurrent episodes of cough and cold for last 4 years. There was history of breathlessness and poor weight gain since 3 years. The child used to clench teeth while feeding. Birth history was insignificant; however, history of mild developmental delay was present more so in motor domain. The child was immunized till date. There was no history of fever, altered sensorium or abnormal body movements. No history of jaundice, hearing or visual deficit was present. The child was born to a non-consanguineous marriage. There were the deaths of two previous siblings with history suggestive of splenomegaly; however, no specific diagnosis was ascertained in these children.

General examination revealed a malnourished child with presence of pallor and grade III clubbing. No evidence of icterus, cyanosis or lymphadenopathy was present. Per abdominal examination revealed enlarged firm liver with sharp border. Spleen was also enlarged and firm-to-hard in consistency. Respiratory rate was increased (60 per minute) with evidence of nasal flaring. Diffuse crepts were seen on both the sides. Bronchial breath sounds were present in right infrascapular level. Routine hematological examination revealed macrocytic anemia. The ultrasound abdomen revealed hepatosplenomegaly with normal echotexture of liver and spleen. Echocardiography was normal. Chest X-ray PA view revealed bilateral diffuse reticular markings [Figure 1]. Subsequently HRCT Chest was done which revealed geographic areas of ground glass opacities with interlobular as well as intralobular septal thickening giving crazy paving appearance [Figure 2]. No evidence of any consolidation, atelectasis or bronchiectasis was seen. Based on clinical, biochemical and radiological findings, a provisional diagnosis of storage disease was considered. Bone marrow aspiration was done which showed normoblastic erythroid hyperplasia and normal myeloid and megakaryocytic series along with few histiocytes displaying ‘crumpled tissue paper’ like cytoplasm resembling Gaucher’s cells [Figure 3]. Enzyme assay revealed low level of β-glucocerebrosidase enzyme measuring 5.50 nmol/hr/mg (Normal value ≥6 nmol/hr/mg). There was 1000 times fold increase in chitotriosidase enzyme which measured 30305 nmol/hr/ml (normal value <400 nmol/hr/ml).

Gaucher disease is an autosomal recessive disorder which results from the enzyme Glucocerebrosidase deficiency. As a result there is accretion of lipid laden macrophages known as Gaucher’s cells in the organs like liver, spleen, bone marrow as well as lung.\[1\] The disease is more prevalent in Ashkenazi Jews.\[2\] These patients present with hepatosplenomegaly, bony and neurological involvement which varies in severity from nystagmus to severe mental retardation.\[3\]

Gaucher disease is divided into three subgroups depending on age of presentation, severity, clinical features and neurological involvement:

Type 1 - Chronic non-neuropathic form which is seen in early adulthood or adolescence and patients present with anemia, massive hepatosplenomegaly and bone marrow involvement.

Lung lysed: A case of Gaucher disease with pulmonary involvement

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Type II - Acute neuropathic or infantile form and is characterized by severe progressive neurological deterioration with aggressive course. It presents with severe hepatosplenomegaly, progressive neurological involvement and rarely bony involvement.

Type III - Subacute neuropathic or juvenile form, is the least common and the age group affected is between 2 and 6 years. Predominantly presents with hepatosplenomegaly and mild neurological symptoms with late-onset skeletal changes as was seen in our case. These patients succumb to complications due to systemic changes.

Lung involvement varies and is seen in the severe form of the disease.[1] There is wide range of pulmonary involvement from clinically asymptomatic with normal or mild radiographic changes to severe respiratory symptoms with marked radiographic findings. Lung involvement is the result of infiltration of either alveoli, interstitium, bronchi or pulmonary vasculature by the Gaucher’s cells. AV shunting is also seen due to hepatopulmonary syndrome.[2] Combined interstitial and alveolar involvement with geographic pattern of involvement is rarer type of lung involvement and is only described in one case report by Aydin et al. Lung involvement is seen in Type II and III (severe forms). There is higher risk of pulmonary involvement in patients of Gaucher’s disease who are homozygous for the 1448G (L444P) mutation.[3]

Chest X-ray and HRCT chest is done in patients presenting with respiratory complaints. Chest X-ray can be either normal or shows diffuse reticular markings as seen in our case too. Three different patterns of lung involvement are seen on HRCT i.e., either only interstitial involvement or only ground glass opacities or a combination of two. Mixed interstitial and alveolar involvement of lung with geographic pattern seen in our case is a rare presentation. Atelactasis and bronchiectasis can also be seen in few cases.

Complications include chronic liver disease and hepatopulmonary syndrome which has unfavorable outcome.[4] Clinical and biochemical correlation is required for the diagnosis of Gaucher’s disease. In addition to reduced levels of glucocerebrosidase, there is 100-fold increase in chitotriosidase enzyme activity which is a sensitive marker. Definitive diagnosis includes bone marrow aspiration, enzyme assay, bronchoalveolar lavage or lung biopsy. Type 1 patients respond well to enzyme replacement therapy. Substrate reduction therapy and bone marrow transplantation are other modes of treatment. Schiffmann et al. reported the improvement in lung function test in type III by combined treatment with substrate reduction therapy (miglustat) along with enzyme replacement therapy.[5]

Pulmonary involvement in Gaucher’s disease in pediatric population is rare and has worse outcome. Lung damage in Gaucher disease has unfavorable prognosis even...
after enzyme replacement therapy as some damage is irreversible and some patients respond slowly to the treatment.\[7\]

This case highlights the geographic pattern of lung involvement in a child with interstitial as well as alveolar involvement which is very rare type of pulmonary involvement in Gaucher disease.

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Conflicts of interest
There are no conflicts of interest.

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