Case Report

Utility of fluorine-18-fluorodeoxyglucose positron emission tomography/computed tomography in a child with chronic granulomatous disease

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ABSTRACT

We report the fluorodeoxyglucose positron emission tomography/ computed tomography (FDG - PET/CT) findings in an 11-month-old boy with suspected milk protein allergy, presented to the hospital with 2-month history of fever of unknown origin and failure to thrive. It showed FDG avid lymphadenopathy above and below the diaphragm and splenic focus, which could represent diffuse inflammatory process or lymphoma. Subsequent jejunal biopsy showed non-necrotizing granulomas.

Keywords: Chronic granulomatous disease, fluorodeoxyglucose positron emission tomography/computed tomography, recurrent infection

INTRODUCTION

Chronic granulomatous disease (CGD) is a rare pediatric disease, presenting with a multitude of systemic signs and symptoms. A multi-modality approach is usually required to reach the diagnosis of CGD. We discuss the role of FDG PET/CT in arriving at the diagnosis of CGD in an 11-month old boy with failure to thrive and recurrent infections.

CASE REPORT

An 11-month-old boy with suspected milk protein allergy presented to the hospital with a 2-month history of fever of unknown origin and failure to thrive. After extensive negative work-up for inflammatory, rheumatologic and malignant pathology, FDG PET/CT was performed to aid in determining the etiology. It demonstrated FDG avid lymphadenopathy above and below the diaphragm [Figures 1 and 2]. A focus of increased FDG uptake was also noted in the enlarged spleen [Figure 3], which showed hypoechoic lesions on ultrasound [Figure 4]. These imaging findings could represent a diffuse inflammatory process or lymphoma. Bone marrow biopsy was normal. Subsequent jejunal biopsy performed showed nonnecrotizing granulomas, which raised the suspicion for CGD. A positive oxidative burst test demonstrated compromised neutrophil function and established the diagnosis. The child showed improvement in symptoms and inflammatory markers with steroids and with voriconazole to treat a fungal infection in the spleen. He is now awaiting hematopoietic stem cell transplant.

DISCUSSION

CGD is one of the conditions seen as a part of the spectrum of disorders of phagocytic function. There is an intrinsic defect in the phagocyte nicotinamide adenine dinucleotide phosphate (NADPH) oxidase, eventually resulting in an inability to destroy the microbe once phagocytosed. Epidemiologically, it is genetically heterogeneous, more common in males,

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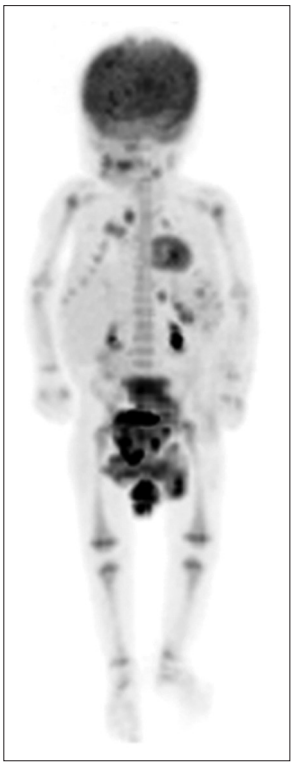

Figure 1: Maximum intensity projection image of FDG PET/CT in an 11-month old boy done after injection of 37MBq of F18-FDG intravenously, showing FDG avid foci in neck, mediastinum, upper abdomen and spleen

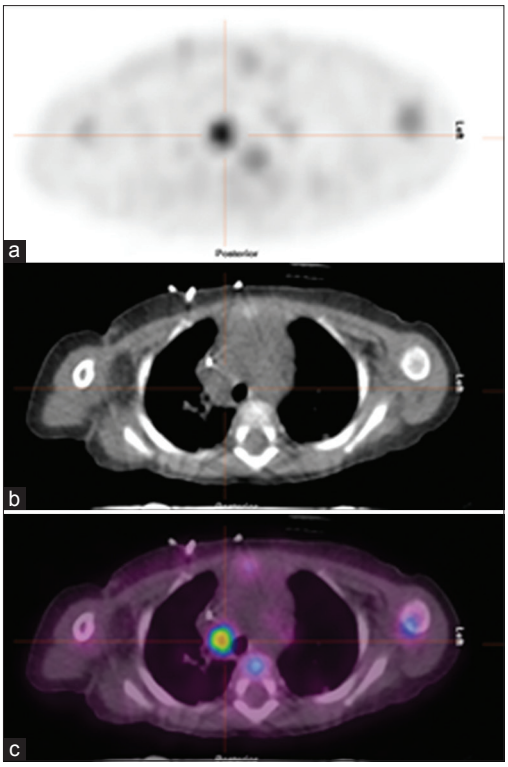

Figure 2: Axial PET(a), low dose CT(b) and fused PET/CT(c) images showing enlarged hypermetabolic mediastinal lymph nodes (e.g., right paratracheal, maximum standardized uptake value 3.9). Similar enlarged FDG avid lymph nodes were seen involving multiple lymph node groups including right intraparotid, cervical, lung hilum, splenic hilum, mesenteric, and inguinal nodal stations

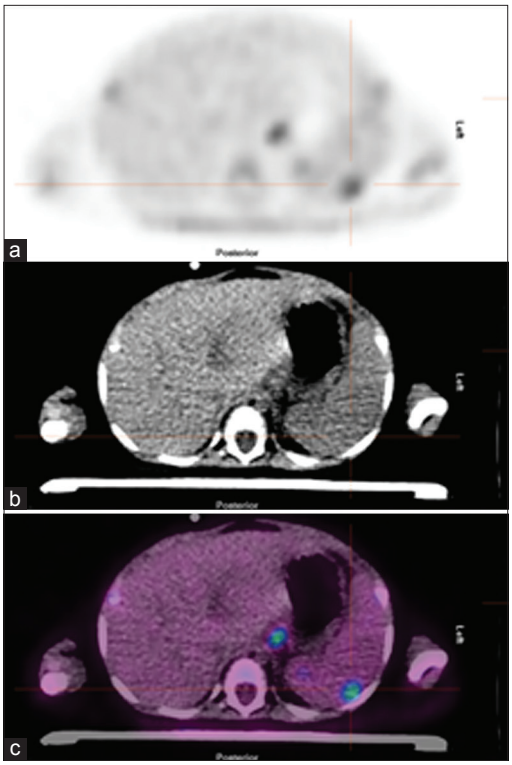

Figure 3: Axial PET(a), low dose CT(b) and fused PET/CT(c) images images showing a hypermetabolic lesion in the spleen (maximum standardized uptake value 2.4) with no definite low dose computed tomography correlate

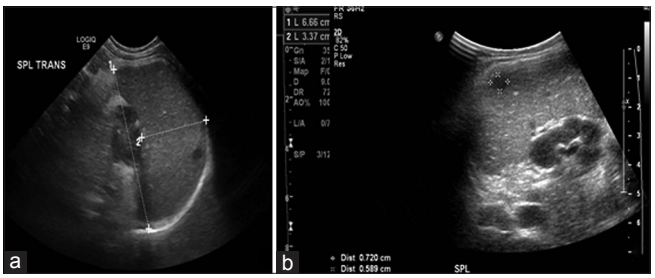

Figure 4: Sonogram of the abdomen showing splenomegaly (a), diffusely heterogeneous spleen and with hypoechoic subcentimeter splenic nodules (b), which could be secondary to fungal infection or multiple abscesses

occurring with a frequency of 1:200,000 live births in the United States.[1] Although it can present any time from infancy to late adulthood, the median age is 2.5-3 years.[2] It most commonly presents with severe, recurrent infections of the respiratory tract and skin by microorganisms that are catalase positive (most bacterial and all fungal pathogens are catalase positive). Other presentations include defective response to mycobacteria, growth failure, dermatitis/eczema granuloma formation in the gastrointestinal and genitourinary tract, hepatosplenomegaly and lymphadenopathy. Since laboratory workup may show nonspecific signs (e.g., hypergammaglobulinemia, anemia, elevated erythrocyte sedimentation rate and C-reactive protein), specialized testing of neutrophil function, immunotyping and

genoblot are recommended in suspected patients. The nitroblue tetrazolium dye reduction test is one of the oldest screening tests used to establish the adequacy of neutrophil function while dihydrorhodamine flow cytometry is the method of choice for diagnosis.[3,4] It is also used for prenatal diagnosis for families with a known history of such disorders. FDG PET has also demonstrated some utility in detecting active lesions in patients with CGD. The mechanism of FDG uptake has been postulated to be incorporation into phagocytosis during energy dependent adenosine triphosphate generation or non-NADPH oxidase related killing.[5] In an article by Güngör *et al*. FDG PET has been shown to be helpful in finding active infective foci, locating potential biopsy sites, prebone marrow transplant (BMT) evaluation and response to therapy.[6] Allogenic hematopoietic stem cell transplantation (BMT) is currently the only the curative treatment for CGD, FDG PET has also been used to document complete resolution of infective foci post BMT.[7]

CGD is a rare diagnosis, and only a few cases have been reported in the remote literature. Advanced diagnostic techniques are now available to help solve the clinical dilemma, as in our case. It demonstrates the integrated use of various modalities to diagnose and determine the distribution of disease in a timely manner.

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Conflicts of interest

There are no conflicts of interest.

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