Introduction

Chronic Granulomatous Disease (CGD) is an inherited disorder characterized by recurrent infections and granulomatous inflammation. It affects individuals who lack the capacity to generate the respiratory burst necessary for the generation of superoxide and hydrogen peroxide. This defect is primarily due to mutations in the genes encoding for NADPH oxidase. This defect leaves patients susceptible to infections caused by catalase-positive bacteria and fungi. Patients with CGD often present with recurrent infections, particularly in the skin, respiratory, and gastrointestinal tracts.

Ear, nose, and throat infections have been reported to account for 12% of all infections in patients with CGD, with fungal infections being the most common. Other common infections include respiratory infections caused by Pseudomonas aeruginosa, Staphylococcus aureus, and Aspergillus fumigatus.

Ataxia and decreased mental status shows abnormal increased signal within the pons (arrow) and extending to the left cerebellum (Fig. 3).

Brain/ENT

For patients with CGD has improved. The typical patient with CGD will now survive into adulthood. Overall, the mortality of patients with CGD in the United States is lower than that of patients with similar conditions.

Initially, CGD was known as fatal granulomatous disease of childhood due to its high mortality rate. As modern treatment strategies have been implemented, the prognosis for patients with CGD has improved. However, mortality still occurs in patients with CGD, particularly in the first few years of life. Mortality is more common in patients with X-linked CGD, which is more severe than the autosomal recessive form.

X-linked CGD is typically diagnosed in their second decade or later [1]. Patients with X-linked inheritance typically are diagnosed at a younger age and have more severe manifestations.

The purpose of this presentation is to review the imaging findings of CGD that can manifest throughout the body.

Atrophied right kidney (arrow).

Pulmonary infections may have a prolonged course, complicated by mediastinal or hilar adenopathy (Figs. 6 and 7), pulmonary fibrosis (Fig. 13), honeycomb lung, pulmonary empyema in up to 20% of cases (Fig. 10) [10].

Pulmonary infection. Coronal F18-FDG PET shows multiple areas of abnormal FDG uptake in the lungs and perihilar regions. In the chest (arrows). Abnormal areas are present in both lungs and perihilar regions. Abnormal areas are present in both lungs and perihilar regions.

Liver calcification from prior abscess. Axial T1-W MRI of the liver shows a rounded area of decreased signal intensity near the dome of the liver (arrows). A thrombosed portal vein extends inferiorly from the lesion in the dome (dashed arrow).


References


