Pediatric Oncall Journal (Oct-Dec 2021)
Volume 18, Issue 4: 138-139
DOI: https://doi.org/10.7199/ped.oncall.2021.42

PEDIATRIC ONCALL
CHILD HEALTH CARE

TEACHING FILES (GRAND ROUNDS)

# MASSIVE HEPATOMEGALY AS A PRESENTATION OF CHRONIC GRANULOMATOUS DISEASE

Rhea Vyas[1], Ira Shah[2].

[1]Seth G S Medical College, Mumbai;
[2]Pediatric Infectious Diseases and Gastroenterology, V Care Polyclinic, Mumbai, India.

**KEYWORDS**
Hepatomegaly, Chronic Granulomatous Disease.

**ARTICLE HISTORY**
Received 7 July 2021
Accepted 5 August 2021

**Clinical Problem**

A 4½-month-old boy, first by birth order, born of a third degree consanguineous marriage, presented at his first immunisation visit with episodes of recurrent diarrhoea for 3 months and hepatomegaly. His birth weight was 3.6 kg and he had been hospitalised in the neonatal intensive care unit for 2 weeks after birth for respiratory distress. He was immunized with one dose each of BCG, oral polio, Hepatitis B and HiB vaccines and referred to us for further management of the hepatomegaly. He was exclusively breast fed. On presentation he was malnourished, his weight was 4 kg (<3$^{rd}$ centile) and length was 57.5 cm (<3$^{rd}$ centile). He had massive hepatomegaly (liver was 10 cm palpable with span of 12 cm) with splenomegaly (spleen was 2 cm palpable). Examination of other systems was normal. On investigations, hemoglobin was 9.3 gm/dl, white blood cell count was 9,400 cells/cumm (44% polymorphs, 56% lymphocytes) and platelet count 216,000/cumm. Liver function tests showed a bilirubin of 0.7 mg/dl, SGOT 90 IU/L, SGPT 46 IU/L, total protein 6.9 gm/dl, albumin 3.2 gm/dl, prothrombin time 12.3 secs and partial thromboplastin time of 22.7 sec. Ultrasound abdomen showed hepatomegaly with normal echotexture of liver. Stool examination was normal and negative for cryptosporidium. In view of recurrent diarrhea with failure to thrive an underlying immunodeficiency was suspected. HIV Elisa was negative and lymphocyte subsets with serum immunoglobulin levels were normal. Nitroblue tetrazolium (NBT) was 7% and dihydrorhodamine test was 6% suggestive of Chronic Granulomatous Disease (CGD). Genetic confirmation could not be done due to unaffordability.

*How commonly does CGD present as massive hepatomegaly?*

**Discussion**

CGD is a rare congenital immunodeficiency disease with an incidence of 1 in 2,00,000 to 1 in 2,50,000 live births that causes repeated bacterial and fungal infections from early childhood and infancy.[1,2] Due to X linked or autosomal recessive inheritance of the gene encoding NADPH oxidase, the neutrophils are unable to produce the enzyme and thus unable to destroy invading organisms.[3,4] Most patients present in the first years of life and the most common presentation is of respiratory tract infections (70-80%), lymphadenitis (60-80%) with hepatomegaly (50-90%).[1,5]

X-Linked CGD is seen in males and patients present at an earlier age and have a more severe clinical course compared to those that inherit the defect via the autosomal recessive pattern.[1] The autosomal recessive pattern may be seen more commonly in children born of a consanguineous marriage. Our patient, being a male and presenting at his first immunisation visit is most likely suffering from the x-linked variant. Those affected suffer from infections of organs that are frequently exposed to the environment like pneumonia, cutaneous infections or infections of the lymph nodes draining them. This can cause lymphadenitis, and subsequent hematogenous spread which can involve other organs and cause hepatic abscesses, osteomyelitis and brain abscesses.[5] The common organisms causing infections are Staphylococcus aureus, Serratia marsescens, Nocardia species, Aspergillus species and Burkholderia cepacia.[6] Due to the increased frequency of infections and increased load on the body and immune system to fight these infections, patients are also seen with involvement of the reticuloendothelial system leading to hepatomegaly, splenomegaly lymphadenopathy, hypergammaglobulinemia, and anemia of chronic disease.[5] Although most cases present first with recurrent infections and are then investigated for CGD, hepatomegaly is often noted at the time of presentation.[5] Finding of hepatomegaly before onset of infections if followed by investigations might reveal the underlying immunodeficiency. Thereafter timely prophylactic therapy and avoidance of live vaccines may be possible which could improve clinical outcomes.[7]

**Address for Correspondance:** Dr Rhea Vyas
Seth G S Medical College, Mumbai 400012.
**Email:** rheavyas97@gmail.com



**Compliance with ethical standards**

Funding: None
Conflict of Interest: None

**References**

1. Winkelstein JA, Marino MC, Johnston RB, Boyle J, Curnutte JT, Gallin JI, Malech HL, et al. Chronic granulomatous disease: report on a national registry of 368 patients.

Medicine (Baltimore) 2000; 79:155-169.

2. Rawat A, Singh S, Suri D, Gupta A, Saikia B, Minz RW, et al. Chronic granulomatous disease: Two decades of experience from a tertiary care centre in North West India. J Clin Immunol. 2014;34:58-67.
3. Roos D, de Boer M. Molecular diagnosis of chronic granulomatous disease. Clin Exp Immunol. 2014;175(2):139-49.
4. Segal BH, Leto TL, Gallin JI, Malech HL, Holland SM. Genetic, biochemical, and clinical features of chronic granulomatous disease. Medicine (Baltimore) 2000; 79:170-200.
5. Dinauer MC, Newburger PE, Borregaard N. The phagocyte system and disorders of granulopoiesis and granulocyte function. In: Nathan DG, Orkin SH, Ginsburg D, Look AT, Fischer DE, Lux SE, editors. Nathan and Oski's Hematology of Infancy and Childhood. 8th edn. WB Saunders. Philadelphia. 2015: 833-841.
6. Marciano BE, Spalding C, Fitzgerald A, Mann D, Brown T, Osgood S, et al. Common severe infections in chronic granulomatous disease. Clin Infect Dis. 2014;60:1176-83.
7. Rubin L, Levin M, Ljungman P, Davies EG, Avery R, Tomblyn M, et al. 2013 IDSA clinical practice guideline for vaccination of the immunocompromised host. Clin Infect Dis. 2014;58:e44-100.