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Case report- chronic granulomatous disease in newborn

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Abstract:

Background: Chronic granulomatous disease (CGD) is a rare, potentially life-threatening primary immunodeficiency caused by defects in the NADPH oxidase complex, leading to impaired phagocytic function and recurrent bacterial and fungal infections. Neonatal onset is extremely uncommon and may present with atypical, severe manifestations.
Case Presentation: We report a male neonate presenting at 44 days of life with persistent fever, nasal regurgitation of feeds, severe perianal excoriation, and necrotic palatal ulceration. Laboratory evaluation revealed leukocytosis, elevated C-reactive protein, and blood cultures positive for Klebsiella pneumoniae, Acinetobacter baumannii, and Pseudomonas aeruginosa. Galactomannan ELISA suggested concomitant Aspergillus infection despite a negative KOH mount. Dihydrorhodamine (DHR) assay demonstrated a markedly reduced neutrophil oxidative index (3.4%), confirming CGD. The patient was managed with targeted intravenous antibiotics, surgical debridement, and initiation of prophylactic cotrimoxazole.
Conclusion: This case illustrates an unusual neonatal presentation of CGD with extensive necrosis involving the hard palate, nasal septum, and perianal region, highlighting the heterogeneity and diagnostic challenges of early-onset disease. Prompt recognition via DHR assay is essential for timely prophylactic therapy and multidisciplinary management. Awareness of such atypical presentations may facilitate earlier diagnosis, guide intervention, and improve neonatal outcomes. Longitudinal studies are warranted to clarify prognostic implications and optimize management strategies in extremely early-onset CGD.

Keywords:

Chronic granulomatous disease; CGD; neonatal immunodeficiency; NADPH oxidase defect; necrotic palatal ulceration; perianal necrosis; Dihydrorhodamine assay; early-onset CGD; bacterial and fungal infections

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Introduction

Chronic granulomatous disease (CGD) is a rare inherited primary immunodeficiency disorder characterized by defective phagocyte function, leading to recurrent bacterial and fungal infections. The reported incidence is approximately 1 in 200,000 live births in the United States [1] and 1 in 25,000 in India [2], with regional variation likely reflecting genetic and epidemiological differences

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[1,2]. The underlying defect involves the nicotinamide adenine dinucleotide phosphate (NADPH) oxidase complex, most commonly due to mutations in gp91phox, p47phox, p22phox, p67phox, or p40phox genes [3]. Approximately 70% of cases are X-linked, caused by mutations in the CYBB gene encoding NOX2 (gp91phox) on the X chromosome, which explains the higher prevalence in males [3].

Most patients are diagnosed between one and three years of age when recurrent infections become clinically apparent. The lungs, lymph nodes, liver, and skin are the most frequently affected sites, with common pathogens including Staphylococcus aureus, Burkholderia cepacia, Serratia marcescens, Nocardia spp., and Aspergillus spp., particularly A. fumigatus and A. nidulans [4]. Although CGD typically presents in early childhood, neonatal presentation is extremely uncommon. Atypical manifestations such as gastrointestinal mucormycosis, cardiac empyema, or phlebitis have been described, which may complicate recognition and delay diagnosis in this age group [2,5].

Case Presentation

A male neonate weighing 2600 g was delivered at term to a G3P2+1L2 mother with gestational diabetes mellitus (GDM) via lower segment cesarean section (LSCS) at a private hospital. The neonate cried immediately after birth, was stable, and was initially bottle-fed with formula milk.

At 44 days of life, the infant presented with a 15-day history of fever, a 3-day history of regurgitation of feeds through the nose, and severe perianal excoriation. Two days prior to presentation, palatal gangrene had developed. There was no family history of immunodeficiency or similar illnesses.

On examination (Figures 1–3), a 2 × 2.5 cm area of posterior palatal gangrene with thick purulent discharge and perforation was noted. Severe perianal excoriation with loss of subcutaneous tissue and pus discharge was present. Crusting was observed in the anterior nares, and removal revealed a septal perforation.

Laboratory investigations demonstrated leukocytosis and elevated C-reactive protein levels. Cerebrospinal fluid (CSF) analysis was suggestive of bacterial meningitis; however, CSF cultures were sterile. Blood cultures were positive for Pseudomonas aeruginosa, Acinetobacter baumannii, and Klebsiella pneumoniae. Serological testing including VDRL was non-reactive, and KOH mount was negative. Enzyme-linked immunosorbent assay (ELISA) for galactomannan was positive, suggesting Aspergillus infection.

Given the presence of necrotic palatal perforation and recurrent severe infections, a primary immunodeficiency was suspected. A dihydrorhodamine (DHR) assay revealed a neutrophil oxidation index of 3.4 (normal >70), confirming the diagnosis of CGD. Exome sequencing of CYBB and other NADPH oxidase complex genes was initiated for genetic confirmation. Biopsy samples from necrotic tissue were also obtained for histopathological evaluation.

The infant was managed with broad-spectrum intravenous antibiotics tailored to culture sensitivity. Surgical consultation was obtained for debridement of necrotic tissue. After three weeks of therapy, the lesions healed, although the palatal perforation persisted. Plastic surgery follow-up was planned for reconstructive management. The patient was commenced on cotrimoxazole (septran) prophylaxis and was fed expressed breast milk using a katori spoon. At discharge, the infant was clinically stable and enrolled for long-term multidisciplinary follow-up.

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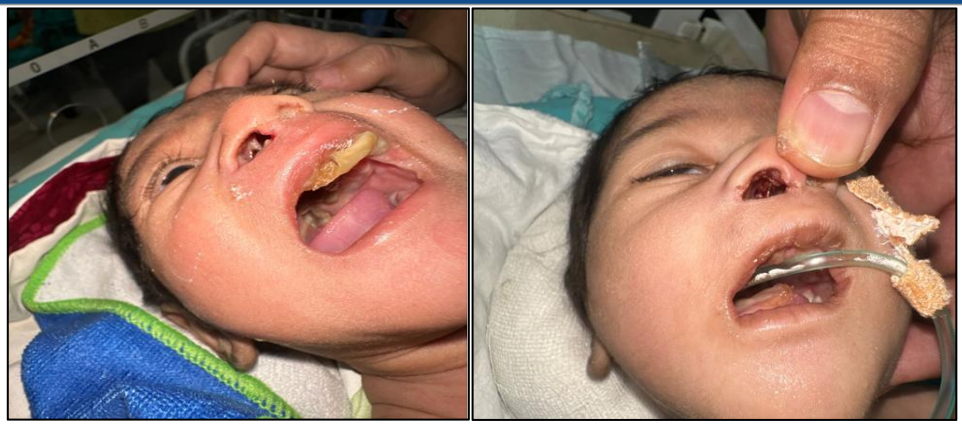

Figure 1: Clinical presentation of neonatal absent nasal septum and associated palatal necrosis

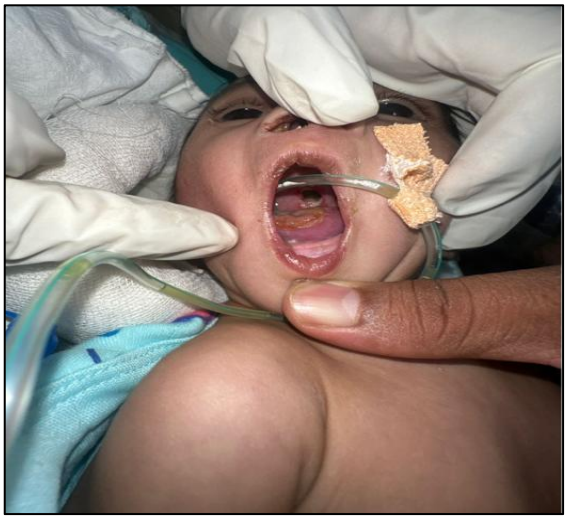

Figure. 2: Extensive palatal ulceration and perforation in a newborn

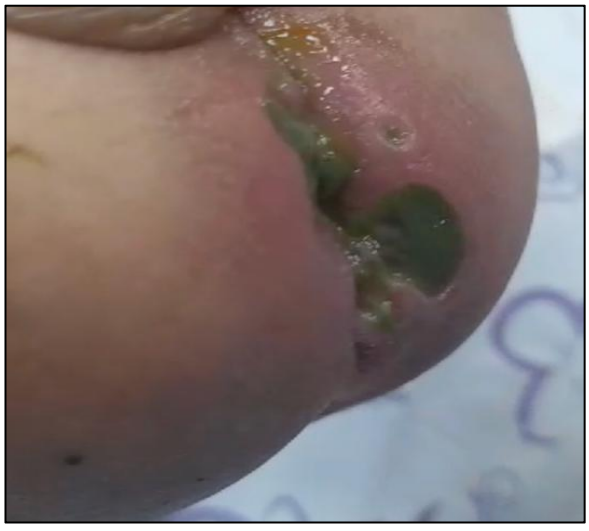

Figure. 3: Perianal ulcerations with surrounding excoriation and inflammatory changes

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Discussion

CGD is a rare primary immunodeficiency caused by defects in the NADPH oxidase complex. This defect impairs phagocytic generation of reactive oxygen species, thereby compromising microbial killing and predisposing patients to recurrent, often life-threatening bacterial and fungal infections [5]. While most patients present in early childhood, neonatal onset is rare [1,5]. A review of the literature has documented 24 neonates who developed CGD-related symptoms within the first six weeks of life, with a slight male predominance and a higher frequency of the X-linked form, consistent with patterns seen in older cohorts [1,2].

The most frequently affected sites in CGD include the skin, lungs, and perianal region. Abscesses may also occur in the liver, spleen, and bones, and hepatomegaly is often reported. Pulmonary involvement may manifest as hilar lymphadenopathy, bronchopneumonia, empyema, or lung abscesses. In neonates, cutaneous manifestations are variable, ranging from erythematous or vesiculopustular lesions to skin abscesses, while papulopustular eruptions are less common [1,2]. In contrast, our patient presented with extensive necrosis of the hard palate, nasal septum, and perianal tissues in the absence of hepatomegaly or intra-abdominal abscesses, underscoring the atypical and severe spectrum of neonatal presentations.

Pathogens most commonly implicated in CGD are catalase-positive organisms, including Staphylococcus aureus, Burkholderia cepacia, and Aspergillus species [3]. Invasive fungal infections, particularly due to Aspergillus spp., affect nearly half of patients. In our case, blood cultures yielded Klebsiella pneumoniae, Acinetobacter baumannii, and Pseudomonas aeruginosa. Additionally, a positive galactomannan ELISA suggested probable Aspergillus infection despite negative KOH microscopy, reflecting the complexity of microbial involvement in CGD.

Diagnosis in neonates is particularly challenging because early symptoms are nonspecific, often resulting in delayed recognition. The median age at diagnosis is approximately eight months [3]. Management centers on lifelong prophylaxis with antibacterial (e.g., cotrimoxazole) and antifungal agents. Hematopoietic stem cell transplantation (HSCT) remains the only curative therapy, with survival rates exceeding 80% in recent series [3,5]. For refractory infections, adjunctive measures such as granulocyte transfusions or recombinant interferon-γ may be employed. Advances in gene therapy, evolving from in vitro correction of B cells to early in vivo applications, hold promise as future definitive treatment options [1,4].

This case highlights the importance of considering CGD in the differential diagnosis of neonates presenting with severe, recurrent, or atypical infections. Early recognition, achievable with simple and cost-effective assays such as the DHR test, allows timely initiation of prophylaxis and multidisciplinary care, which have markedly improved long-term outcomes and quality of life in affected patients

Conclusion

CGD is a rare, life-threatening primary immunodeficiency that typically manifests in early childhood. This case is notable for its onset at birth, presenting as necrotic palatal ulceration with involvement of perianal tissue and the nasal septum—an unusual neonatal presentation. The marked clinical heterogeneity underscores the diagnostic challenges in neonates with atypical severe infections. Early recognition, facilitated by assays such as the DHR test, is essential for timely antimicrobial prophylaxis and multidisciplinary management. Further longitudinal studies are required to clarify the prognostic implications of extremely early-onset CGD and to guide strategies for early diagnosis and optimized care.

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