Case Study

A Case Report on X-Linked Chronic Granulomatous Disease (CGD)

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ABSTRACT

The Chronic Granulomatous Disease is a rare primary immunodeficiency disease, principally attacking white blood cells, characterized by an increased susceptibility to the recurrent bacterial and fungal infections along with the genesis of granulomas in tissue. Staphylococcus and aspergillosis are the main pathogens responsible for majority of infections in CGD. Here we report a case of 5yrs old boy who came with the complaints of fever, cough, and multiple abscess over the right leg, right ankle and left hand with pus discharge. DHR test was performed and have shown the positive results for chronic granulomatous disease.

1. Introduction

Chronic granulomatous disease (CGD) is a genetic disorder of immune system that fails to destroy certain invading bacteria and fungi due to the defect in phagocytic cells to produce reactive oxygen species. The pathology under phagocytic cell defect is mutations in any one of the four genes encoding for the phagocyte NADPH oxidase, the enzyme that generates oxygen radicals which is essential for microcidal activity[1]. Mutation in the CYBB gene which encodes for gp 91-phox component is responsible for x-linked CGD[2]. Mutations in the CYBA, NCF1 and NCF2 which encodes for p22-phox, p47-phox, p67-phox respectively is responsible for autosomal recessive CGD[3]. There is a male to female ratio of seven to one reflecting the predominance of X-linked CGD over autosomal recessive CGD[4]. The hallmarks of CGD are recurrent infections at epithelial surfaces such as skin, lungs and gut as well as formation of granulomas in tissues[5].

2. Case Presentation

A 5 years boy was brought to Clinic, with the complaints of fever, cough, and multiple abscesses over the right leg, right ankle, and left hand with pus discharge. Similar complaints were seen at 3 years of his age. The child was
Erythrocyte sedimentation rate is high in the 1st hour-48 mm and 2nd hour-90 mm. Lymphocyte subset assay was performed and found that all values were within normal range of CD4+ T Helper cells-37.1 % (28-47 %), CD3+T cells-58.3 % (56-75%), CD19+B cells-33.2 % (14-33 %), CD16&CD56+(NK cells)-6.6 % (4-18 %) except CD8+ Tsuppressor-10.7 % (16-30 %), CD4+CD8 ratio-3.47 (1.26-2.90). DHR assay was performed and the results was abnormal indicating low stimulation index (SI)-2.35 (180). But further genetic studies are needed to confirm the type of CGD. Genetic testing was done and found mutations in CYBB gene and confirmed the diagnosis of X-linked CGD. Treatment given was Inj Metrogyl 25 ml IV TID, Inj Meropenem 250 mg IV TID, Inj Vancomycin 210 mg in 100 ml NS TID for 21 days and Inj Augmentin 700 mg IV BD for 4 days.

3. Results and Discussion

Chronic granulomatous disease was first described in 1957 as recurrent infections occurring in setting of hypogamma-globulinemia[13]. Chronic granulomatous disease is characterized by severe, recurrent and often fatal infections which results from defects in NADPH oxidase system to produce superoxide in patient phagocytes[9]. The common infections in CGD are pneumonia, lymphadenitis, liver abscesses, osteomyelitis, skin abscesses, granulomas typically involving genitourinary system and gastrointestinal system[9]. In CGD as most mutations takes place in X-linked gene, affected individuals are mostly male. Pneumonia is most common type of infection encountered in CGD[9]. In our case child has reported pneumonia at 3 months of age. Cytochrome reduction assay & Nitro blue tetrazolium test measures superoxide activity whereas DHR123 assay and amplex red assay test measures hydrogen peroxide[11]. In our case diagnosis was confirmed by DHR123. Prophylactic use of antibiotics, antifungals, interferon gamma has improved prognosis but still CGD is associated with increased morbidity and mortality from life threatening infections. Haematopoietic stem cell transplantation remains prominent cure of disease, but difficulty in finding suitable donors and associated risks with the procedure have limited its use[12]. In recent years gene therapy has been proposed as alternative to stem cell transplantation for CGD patients. Use of regulated SIN-lentiviral vectors targeting gp91 (phox) in myeloid cells to increase safety and efficacy of GT protocols has proposed but still research need to be done[13].

4. Conclusion

Sound knowledge about primary immune-deficiencies should be present among health care practitioners for differential diagnosis of chronic granulomatous disease and application of test at proper time would help in timely diagnosis of disease.

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Conflict of Interest

The author(s) confirm that this article content has no conflict of interest.

References