VIEWER'S CHOICE (LETTER TO EDITOR)

Spectrum Of Chronic Granulomatous Disease in Pediatric Patients: A Review of 20 Patients

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Chronic granulomatous disease (CGD) is a rare primary immunodeficiency caused by a defect of the enzyme nicotinamide adenine dinucleotide phosphate oxidase (NADPH-oxidase) characterized by a reduced ability of neutrophils to destroy phagocytosed microorganisms. The prevalence of CGD varies among the populations investigated, with studies showing variations from one of 1,000,000 to one of 160,000 individuals. (1) The pattern of CGD inheritance can be X-linked (about 70% of cases) or autosomal recessive. (2)

The disease is very often diagnosed in the first years of life but there is some reports of late onset of CGD. (3, 4) While there is great clinical heterogeneity among patients with CGD, most develop bacterial infections particularly produced by catalase-positive microorganisms, mycobacteria and fungal diseases. Such infections are severe and repeated suppurative infections mainly located in the lungs, skin, and lymph nodes, but also affecting other organs. The most common clinical manifestations are lymphadenitis, pneumonia and hepatosplenomegaly.

This retrospective study based on the review of the patient's medical records in order to obtain clinical, laboratory, radiological and epidemiological data including personal and family history during admission and follow-up were obtained by review of 20 patients charts of all patients with chronic granulomatous disease admitted in the Pediatric Pulmonary Ward at NRITLD, a referral center for Tuberculosis and lung disease, from 2002-2007. The diagnosis was based upon WHO criteria for chronic granulomatous disease. In this research the collected data of 20 patients were evaluated and analyzed. There were 13(65%) male and 7(35%) female cases. Mean age was 8.4 years old. These patients belonged to 13 families; Positive family history was detected in 10 (50%) and parents of 19 patients (95%) were relatives. The mean age of onset of symptoms was 2 years (1month-12 years). Common initial manifestation was pneumonia, present in 18 of 20 children (90%), followed by lymphadenopathy in 80%, skin abscesses in 40% and oral ulcers in 30 %. Other relevant data obtained by physical examination were hepatosplenomegaly in nine of 20, another patient had only hepatomegaly. Aspergillosis was detected in pulmonary secretion of 8 (40%) patients suffering from pneumonia as well as in the biopsies material obtained from rib of 1 patient (Figure-1). The most radiological (Chest X-Ray) and CT Scan manifestations were nodular infiltration in all patients and hilar lymphadenopathy in 95% of patients (Figure-2). In regard to laboratory findings, Nitroblue Tetrazolium Test (NBT) was less than 5% in 5 (3 male, 2 females) while in rest of the cases it was 0. Pathological findings demonstrate non caseous granulomatous formation in 13 (65%) patients. In this group of under study, two patients (a single 14-year old boy and a single 11-year old girl) expired as a result of respiratory failure due to drug resistant pneumonia. In the remaining 18 cases, prophylactic regimen (Itraconazole and Cotrimoxazole) was administrated and patients are under follow-up.

Figure 1-Microbial distribution frequency of infected CGD patients

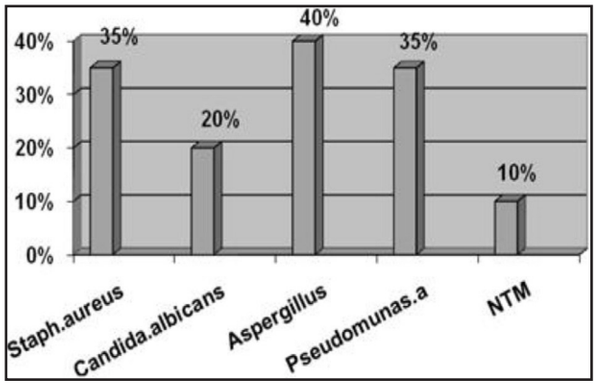
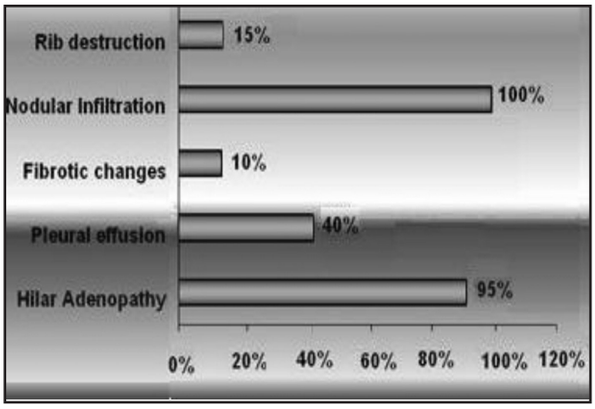


Figure 2-Chest CT-scan findings in CGD patients



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