Genetic Defect in Phagocytic Function Case Study

An infant boy, 6 days of age, developed severe diaper rash. Nine days later, in spite of topical treatment and oral antibiotics, he was hospitalized for evaluation of sepsis because of fever and pustules. His history was remarkable only in that there was a papule at the base of the fifth finger, which was present at birth.

The infant’s chest and abdominal CT scans revealed lesions in the liver and a lung nodule. Upon biopsy, neutrophils were noted in the liver abscesses and *Serratia marcescens,* a gram-negative

bacillus, grew after culture of the liver specimens. Biopsy of the lung nodule revealed evidence of acute pneumonitis, and *Aspergillus,* a fungus, was identified in the lung specimen. Fortunately, after appropriate antibiotic therapy, the lesions healed and the child recovered.

Further testing revealed that the infant’s neutrophils had a negative reaction in two tests designed to assess phagocyte activity (the nitroblue tetrazolium test (NBT) and in the eutrophil

cytochrome *b* 558 assay). These tests monitor the ability of phagocytes to produce toxic oxygen

products during a process called the *respiratory burst.* Based on these data, as well as the boy’s

history, he was diagnosed with X-linked, cytochrome-negative, chronic granulomatous disease

(CGD) of childhood.

* *What are some of the signs and symptoms of CGD that this infant exhibited?*
* *What is typically the problem with phagocytic function in someone diagnosed with CGD?*
* *Individuals with this condition are more susceptible to what type of microbial agents?*