CASE REPORT

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A Case of Panhypogammaglobulinemia Masquerading as Child Abuse

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ABSTRACT: A case of panhypogammaglobulinemia in a 15-month old boy is presented. The child was followed at a local university teaching hospital for pneumonia, failure to thrive, and possible child abuse/neglect. Following minor trauma to the face, massive sepsis developed in the child. The mother was afraid to seek medical care because she was fearful of legal action against her. Upon the child's demise at home, police and medical examiner involvement ensued. The correct diagnosis was established at autopsy.

KEYWORDS: pathology and biology, agammaglobulinemia, child abuse, immune deficiency state, childhood development, child nutrition

Child abuse is an ever present concern to child protection teams, prosecutors, and forensic pathologists. Much has been advocated in standard texts and treatises so that the diagnosis is not overlooked. However, somewhat axiomatically, not everything may be what it first appears to be. This report concerns an unusual immunologic disorder originally thought to be child abuse.

Case Report

L., a 15-month old white male child, was born at a local University teaching hospital. He was a full-term child who was twin "B" in a caesarian section delivery. His 1- and 5-min Apgar scores were 8 and 9, respectively. At birth he weighed 3.14 kg and measured 50 cm in length. He went home and did well until 9 months of age. At that time he was admitted to the same university teaching hospital for pneumonia. He was treated with penicillin and amoxicillin for a 2-week period. Later that month, he was seen in the emergency department for a vague history of a facial hematoma after falling out of a crib. He next was seen at 11 1/2 months of age for vomiting and diarrhea. He weighed 6.5 kg and measured 66.5 cm in length at that time. Failure to thrive, possible neglect, iron deficiency anemia, pyuria, adenopathy, and an ulcerated penis were diagnosed. He was discharged from the university teaching hos-

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pital two weeks later and the mother was instructed to take the child to a "Women, Infant and Children" program. Over the next several months there is a vague history of a Health and Rehabilitative Service visit admonishing the mother concerning child abuse. The child fell from the crib during this time and an "ulcer"-like area developed on the face. The mother, afraid to seek medical attention, treated this increasingly foul smelling area with Listerine® mouthwash, Desiten® skin cream, and rubbing alcohol. The child was found dead in bed several days later and accordingly police were summoned.

At autopsy the child weighed 5.45 kg and measured 72.5 cm. Exteriorly, there was a generalized exfoliative dermatitis on the scalp, trunk, and buttocks. A foul smelling noma of approximately 3 in. (7.6 cm) in diameter was noted in the naso-oropharyngeal region (Fig. 1). Internally, multiple foul smelling abscesses, some 1-in. (2.5 cm) in diameter, eroded both lungs.

The thymus weighed 7.8 g. Multiple ulcers in the upper esophagus were observed. In the cecum, a 1/2-in. (1.3-cm) ulcer was noted. This area was appliqued to the tail of the pancreas. No fractures were seen. No subdural hematoma was noted.

Bacteriologic cultures of the blood, mouth, and lung abscesses grew *P. Aeroginosa, C. Freundii, K. Pneumoniae,* and *S. Aureus.* Immunologic studies revealed an Immunoglobulin A (IgA) of 64 mg/dL (normal 70 to 312 mg/dL), an IgG of 703 mg/dL (normal 639 to 1349 mg/dL), and an IgM of 97 mg/dL (normal 56 to 352 mg/dL).

Complement studies revealed a C3 of 44 mg/dL (normal 83 to 177 mg/dL) and a C4 of 7 mg/dL (normal 15 to 45 mg/dL).

The blood had clotted such that T and B cell subsets could not be performed.

Histologically, the areas of the mouth and lung revealed abscesses with hemorrhagic necrosis of arteries (Fig. 2).

The thymus was markedly involuted (Fig. 3).

Discussion

Antibody deficiency states are a heterogeneous group of disorders [*1*] having in common hypogammaglobinemia, a decreased ability to produce antibodies following antigenic challenge, and an increased incidence of infections [*2*]. Several subtypes have been identified including: (1) X linked infantile agammaglobulinemia, (2) X linked immunodeficiency with hyper IgM, (3) immunodeficiency with normal or hyperimmunoglobulinemia, (4) adult anti-

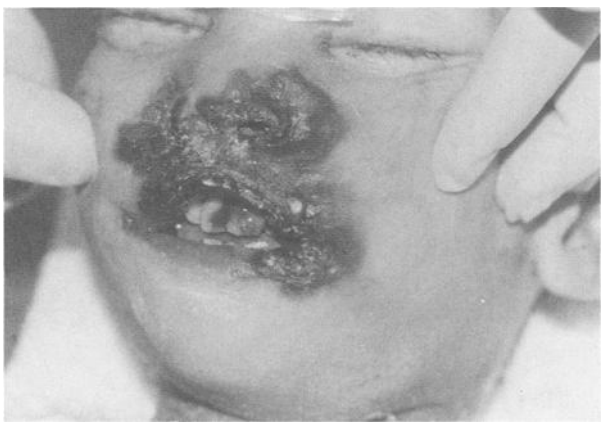

FIG. 1—*Noma of face.*

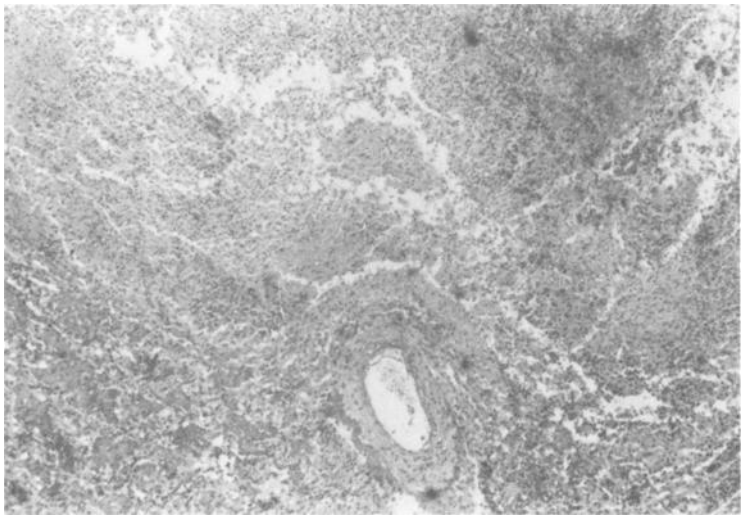

FIG. 2—*Lung with abscesses and necrotizing arteritis, ×40.*

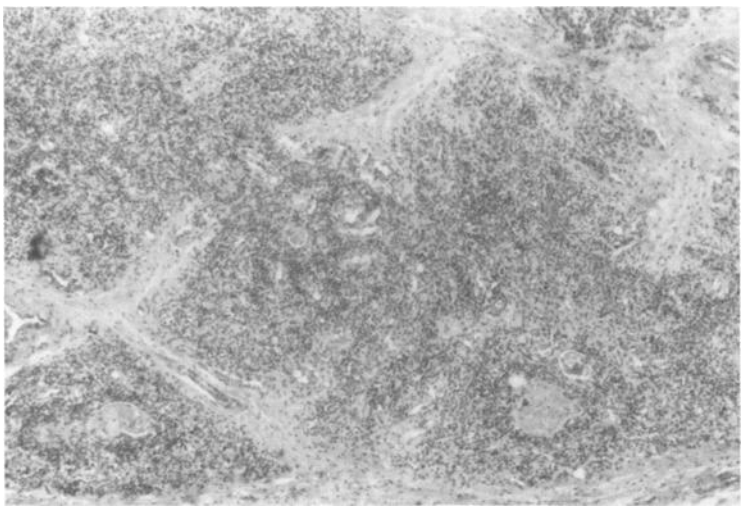

FIG. 3—*Thymus, markedly involuted, ×40.*

body deficiency, and (5) selective IgM deficiency [*3*]. Additional immunologic alterations in children include IgA deficiency [*4*] and alterations involving cellular mediated responses as well as humoral antibodies [*5*]. Such cases are usually diagnosed and treated by a pediatric specialist. In this instance, the forensic pathologist was the "consultant." In some respects this "consultation" was limited. One limitation was the absence of T and B cell subsets. Accordingly, further differentiation as to whether this child's hypogammaglobulinema was solely a humoral defect or combined with a cell mediated defect cannot be ascertained. Certainly, genetic counseling and examination of the child's siblings will also be important in

further delineation of this diagnosis. However, despite such limitations, this report is useful for its implications.

The implication of this case is that while child abuse may be suspected, a careful autopsy is necessary to confirm the diagnosis. Given the absence of fractures and subdural hematomas, such a diagnosis in this instance is untenable. In the postmortem examination of this case, several factors led to the correct diagnosis. First, massive infection with lung abscesses led the author to question how the child could have become infected. Appropriate bacteriologic cultures of the blood, the lung abscesses, and the noma point to bacterial organisms as an etiology. Given such a small thymus, immunoglobulin and complement studies were ordered during the gross autopsy. Finally, thorough police investigation failed to disclose a significant adult male parent figure who could have mistreated the child. The mother seemed sincere in stating she had not harmed the child. Furthermore, no instrument (for example, curling iron, hair dryer, and so forth) could be found at the scene to inflict the noma or the excoriative dermatitis. Finally, search of the literature [*1-5*] disclosed case reports with similar clinical course and pathology similar to that noted in this patient. Accordingly, the correct diagnosis was established.

Future work in this case will include appropriate genetic counseling of the victim's siblings, who exhibited no signs of maltreatment. Others on the child protection team were made aware of the diagnosis and further prosecution of the mother was stopped. Clinicians, social workers, police, and forensic pathologists should recognize this and similar entities in a child with failure to thrive, repeated infections, and abnormal immunoglobulins.

References

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