G100 Case Presentation: Infant Death Due to Epidermolysis Bullosa

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The goal of this presentation is to acquaint the forensic community with the manifestations of epidermolysis bullosa, so that they might include this disease process in their differential when confronted with similar infant deaths.

When confronted with an infant death, this presentation will impact the forensic community and/or humanity by demonstrating the importance for the forensic staff to consider the possibility of a natural disease process with manifestations that could mimic traumatic injuries. The skin lesions of epidermolysis could be mistaken for thermal injuries if a thorough history is not available, and if the possibility of a disease is not considered.

This presentation reviews a case of death caused by complications of epidermolysis bullosa in a 17-month-old Asian infant.This child began to form skin bullae in the diaper area days after birth; these soon spread to include the extremities and face. A biopsy-proven diagnosis of epidermolysis bullosa simplex (Dowling-Meara subtype) was given at age 2 months. Mucosal involvement became apparent when white plaques were noticed in the oral cavity. Recurrent reflux hindered feeding, and resulted in a lack of adequate weight gain. Persistent respiratory difficulties necessitated tracheostomy tube placement at the age of 4 months. The tracheostomy site soon became infected; although treated, a mucoid discharge, on occasion culture-positive for *Staphylococcus aureus* and *Pseudomonas aeruginosa*, continued to drain from the tracheostomy site. A gastrostomy tube was placed at the age of 5 months, due to the child's difficulty swallowing. By his seventeenth month of life, the child's condition had seemed to stabilize and even slightly improve, when, one afternoon, he was found gasping for air; when the father attempted to suction the tracheostomy site, the child stopped breathing. An ambulance transported the child to a nearby hospital, where he was pronounced dead.

An autopsy was performed by the medical examiner's office. The small-for-age body had numerous scattered skin lesions, ranging from unruptured, thin-roofed bullae containing clear fluid, to superficial, red, weeping erosions, to red-brown, dried erosions with a crusted base. The teeth were dysplastic. The fingernails and toenails were absent; the nailbeds had erosions. The oropharyngeal and laryngeal mucosae were markedly edematous, with multifocal ulceration, scarring, and stenosis. The trachea and mainstem bronchi had superficial mucosal erosions as well. Hematoxylin-eosin stained sections of these mucosal erosions had extensive chronic inflammation and submucosal fibrosis. Sections of lung parenchyma had mucus plugs in scattered bronchioles. There was no evidence of trauma. The cause of death was listed as: "Complications of epidermolysis bullosa."

Epidermolysis bullosa is a group of rare genetic disorders that result in fragile epithelium that splits and blisters when subjected to minor trauma. Subtypes are classified by the level of the disrupted epithelium or by genetic basis in newer classifications. Epidermolysis bullosa simplex, due to mutations in genes forming keratins, results in splitting within the superficial layers of the epidermis. Junctional epidermolysis bullosa is due to mutations in genes forming hemidesmosomes or anchoring filaments, and results in separation of the basal cell layer from the basement membrane. Dystrophic epidermolysis bullosa is a result of mutation of the gene forming type VII collagen, and causes separation of the epidermis from the underlying dermis. These disorders range from mild to lethal, and can present at birth or later in life.

When presented with an infant death, it is important for the forensic staff to consider the possibility of a natural disease process with manifestations that could mimic traumatic injuries. The skin lesions of epidermolysis bullosa could be mistaken for thermal injuries if a thorough history is not available, and if the possibility of a disease process is not considered.

Epidermolysis Bullosa, EB, Epidermolysis Bullosa Simplex

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