G37 Gorham-Stout Syndrome: A Presentation of Two Cases and a Review of the Literature

Cristoforo Pomara, MD, PhD, University of Foggia, Dept Forensic Path, University of Malta, Dept of Anatomy, Faculty of Med & Surg Biomedical Sci, Foggia, Misida, Malta 71100, ITALY; Ruggero Parente, MD, Viale degli Aviatori 1, Foggia 71100, ITALY; Stefania C. Bello, MD, Ospedale Colonnello D'Avanzo, viale degli Aviatori, Foggia 71100, ITALY; Santina Cantatore, Viale degli Aviatori 1, Foggia 71100, ITALY; and Benedetta Di Battista, MD, Viale degli Aviatori, 1, Ospedale "Col. D'Avanzo," Foggia 71100, ITALY*

The goal of this presentation is to investigate cause of death in Gorham Stout Syndrome (GSS). Prognosis is most dependent on the site of involvement, extent of disease, and complicating factors. It remains a fact that several cases involve the thoracic duct and the death can occur as a sequelae of malnutrition and lymphopenia. Chylothorax is a serious complication and there are no uniform therapeutic guidelines to follow. The very low incidence of this syndrome as well as the chylotorax clinical presentation and the poor understanding about the etiology make diagnosis difficult.

This presentation will impact the forensic science community by describing morphological features of a rare syndrome and by confirming the importance of detailed postmortem forensic investigations as a unsubstitutable way to improve diagnostic criteria and therapy.

GSS is a rare disorder characterized by progressive osteolysis that leads to the disappearance of bone. Lymphvascular proliferation causes the local destruction of bony tissue. Children and teenagers, without differences between males and females, are usually affected. The first clinical symptoms are pain, tumefaction, and spontaneous fractures of bones. The progression of osteolysis may stop at any moment, but it often evolves with a complete waste of the bone, with the replacement of a fibrous band. Less than 200 cases are documented in the literature. The outcome is poor in patients with soft tissue involvement, with lesions of the axial skeleton and chylothorax. Twenty-five (17%) of patients have been reported to have chylothorax as a complication, according to the last review of the literature.

This presentation reports two fatal cases of GSS.

Case 1: A 9-year-old child affected with GSS was referred to the local Emergency Department (ED) because of severe dyspnoea. Respiratory Function (RF) was 36/min with a rough breath sound. Oxygen saturation was 88%. Tachycardia was recorded with Heart Failure (HF) at 140/min. Blood Pressure (BP) was unremarkable. Respiratory acidosis was detected at lab tests (pH 7.28, pO2 30.3, pCO2 57.9, HCO3 22.0). Complete opacification of the whole left lung with right pleural effusion were observed on an X-ray of the chest. Therapy with an oxygen mask was immediately started and the child was moved to the intensive care unit because of acute respiratory failure. He was unconscious, cyanotic, tachypneic with oxygen saturation at 78%, and 500cc of fluid was drained from the left thorax. Dopamine infusion was quickly carried out; however, after an episode of severe bradycardia, the child collapsed. A hospital autopsy was performed the day after the child's death. The child was 139cm in length and weighed 27kg. At external examination, asymmetry of the head due to osteolysis in the parietal, temporal, and mandibular regions was described. Chylothorax and massive pulmonary edema was described at autopsy. Gross examination of the heart was unremarkable. The histological findings of the lungs were suggestive of diffuse pleural effusion characterized by large, thin-walled, and variably sized lymphatic vessels. The immunohistochemical staining method revealed vascular proliferation in the parietal and visceral pleura. Acute respiratory failure in GSS was indicated as the cause of death following a mediastinic syndrome with metabolic and respiratory acidosis and inspiratory dyspnoea.

Case 2: A 5-year-old child with a history of mediastinic syndrome affected by GSS was admitted to the local pediatric surgery department due to high fever and left chest pain. Chilothorax was observed in the chest and 600cc of lymphatic fluid was seen in the left thorax. Fatal bradycardia occurred suddenly. A hospital autopsy was performed and the external examination was unremarkable. Massive cerebral edema was recorded with white foam on the main bronchi. Yellowish liquid in the left thorax was detected. Disarray of the myofibers and multiple foci of contraction band necrosis were observed at the microscopic study of the heart. Interstitial myocardial edema and lymphangectasia were also described. Pleural lymphatic vessel proliferation with focal pulmonary edema was recorded as well as broncho-pneumonia and bronchiolitis. The histological examination of the liver revealed dilatation of linfatic vessels. The immunohistochemical staining method revealed generalized vascular proliferation of all organs. Acute cardiac failure in GSS was indicated as cause of death.

Gorham Stout Syndrome, Osteolysis, Child Disease

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