G69 Sickle Cell Disease and Sudden Death

Michelle A. Jorden, MD, Jennifer A. McReynolds, PhD, and Adrienne E. Segovia, MD, Cook County Medical Examiner, 2121 West Harrsion Street, Chicago, IL 60612*

The goal of this presentation is to educate and alert the forensic community to the common causes of death in sickle cell patients.

This presentation will impact the forensic community by demonstrating the importance of recognizing pulmonary complications at autopsy that are frequently seen in the sickle cell population and which are responsible for causing sudden death. Lastly, the possibility of sickle cell disease should be entertained in any young African-American person who dies suddenly and unexpectedly without a known history of sickle cell disease.

Sickle cell disease afflicts one of every 650 African Americans and an estimated 8% of African Americans are heterozygous for the sickle cell gene. Sickle cell anemia is attributed to profound morbidity as well as mortality to those afflicted with the disease. In the clinical setting, sickle cell anemia can present as recurrent infection especially in the younger population and as sickle cell pain crisis, stroke, and sudden death in the adult population. In 1949, the discovery that sickle hemoglobin exhibited an abnormal electrophoretic mobility has pioneered our current understanding that sickle cell disease is a molecular disease/diagnosis. With the advent of immunizations and vaccinations, antibiotic therapy and the implementation of newborn screening programs, the mortality rate of individuals with sickle cell disease has declined.

Although morbidity and mortality from sickle cell disease has declined in recent years, a subset of patients die from sudden and unexpected deaths. The most common causes of sudden death in individuals succumbing to sickle cell disease are acute and chronic pulmonary complications that encompass pulmonary edema, pulmonary thromboembolism/thrombosis, and pulmonary hypertension. Although the literature comments on the presence of pulmonary thromboembolism as a frequent autopsy finding, the literature is unclear as to whether these patients also exhibited deep venous thrombosis. Therefore, sickle cell patients may not share the same risk factors for development of pulmonary thromboembolism as the rest of the population (i.e., recent surgery, obesity, and immobilization). Instead, sickle cell patients may undergo pulmonary thrombosis as a consequence of in-situ sickling of red blood cells within blood vessels during hypoxic episodes. For forensic pathologists who perform autopsies on all cases of sudden death, the history of sickle cell disease may be absent or may never have been diagnosed, especially in athletic adolescents. Therefore, in those individuals who are of African American race, younger age, and presenting with pulmonary thromboembolism/thrombosis in the absence of known associated risk factors, the importance of a thorough autopsy examination including a detailed gross and microscopic examination of the heart and lungs, and dissection of the lower extremity veins, in conjunction with postmortem hemoglobin solubility/electrophoresis tests are underscored.

During the past year at the Cook County Medical Examiner's Office in Chicago, IL the authors have encountered two (2) cases of young African American individuals (one female at 31 years old and one male at 38 years old) who carried a diagnosis of sickle cell disease and who were found unresponsive with no known antecedent symptoms. After a complete autopsy, the cause of death in both cases was pulmonary thromboembolism/thrombosis in the absence of deep venous thrombosis. In both cases, the microscopic sections displayed acute and organizing pulmonary thrombi. In addition, both cases displayed severe pulmonary hypertensive changes characterized by thickened pulmonary arterioles and plexiform arteriopathy.

Given these findings, the decision was made to pull all cases of young African American individuals who died suddenly from pulmonary thromboembolism/thrombosis without a known history of sickle cell disease over a two year period. Three (3) additional cases were identified and consisted of two females and one male (ages 20-40s). Hemoglobin solubility tests were performed on the postmortem blood of these individuals. The hemoglobin solubility tests were negative in all three cases. Although the tests were negative, we eliminated the possibility of sickle cell disease as a contributory factor and upon further review of these cases, all these cases demonstrated deep venous thrombosis, and an identifiable risk factor was observed in one case.

Sickle cell disease is a common disease that afflicts the African American population. For forensic pathologists, the findings of pulmonary thrombosis in the absence of deep venous thrombosis and associated risk factors in the young African American population should alert the forensic pathologist to the possibility of sickle cell disease and further laboratory testing of postmortem blood for hemoglobin solubility.

Sickle Cell Disease, Sudden Death, Pulmonary Complications

 ** Presenting Author*