G1 Exertional Collapse in a Youth With Hemoglobin SE

Elise F. Arbefeville, MD, and Vernard I. Adams, MD, Hillsborough County Medical Examiner Department, 401 South Morgan Street, Tampa, FL 33602*

After attending this presentation, attendees will understand one of the potentially lethal sequelae of hemoglobinopathy SE.

This presentation will impact the forensic community by helping forensic pathologists recognize occurrences of exertional death in compound heterozygous SE hemoglobinopathy.

Hemoglobin S is the most common abnormal hemoglobin. It occurs in 10% of North Americans of West African descent. Hemoglobin E is the second most frequent abnormal hemoglobin and is found in persons of Southeast Asian ancestry. Because of the wide geographic separation of the original epicenters for these genotypes, the double heterozygous and manifesting as hemoglobinopathy SE is uncommon with less than 30 cases reported. Hemoglobinopathy SE is typically asymptomatic. A twelve-year-old American boy was participating in football practice in the sun in summer when he collapsed ill on the ground. When the paramedics arrived, he was alert, hypotensive and in sinus tachycardia. His temperature was 97.3 degrees Fahrenheit. During transportation to the hospital his heart was unstable and several sternal rubs were performed. He initially responded to the treatment but his heart rate dropped suddenly. He became unresponsive with pulseless electrical activity followed by asystole. Resuscitation efforts started by the paramedics and continued in the Emergency Room were unsuccessful. On gross examination the spleen, lungs, brain, liver, and heart were unremarkable. Microscopically the cerebellum, heart, and kidney had sickled erythrocytes. Hemoglobin electrophoresis revealed 0.9% hemoglobin F, 57.4% hemoglobin S, 34.2% hemoglobin E, and 7.5% other. Follow-up investigation determined that the decedent's father was of West African ancestry and his mother had Thai ancestry. This is an example of exertional collapse in a person with hemoglobinopathy SE with a clinical presentation similar to that sometimes observed in persons with sickle cell trait. The pathophysiology of sickling in persons heterozygous for hemoglobin S is discussed with particular reference to the mitigating and aggravating effects of other abnormal hemoglobin haplotypes.

Hemoglobinopathy SE, Exertional Collapse, Sudden Death

 ** Presenting Author*