G56 Postmortem Recognition of Sickle Cell Trait

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After attending this presentation, attendees will gain a better understanding of the importance of recognizing the incidental autopsy findings related to sickle cell trait and the implications this diagnosis may have on surviving family members.

This presentation will impact the forensic community through knowledge gained about sickle cell trait and from insight regarding the importance of notification of surviving family members.

Sickle cell trait is defined as the heterozygous condition of having one gene for sickle cell hemoglobin and one for normal hemoglobin. Patients with sickle cell trait tend to lead normal lives, without serious complications; therefore it is not regarded as a disease state. In certain conditions; however, complications not only occur, but they may be fatal. Deaths due to exertional sickling involving young athletes have made headline news on multiple occasions. When an individual with sickle cell trait becomes hypoxic, acidotic, dehydrated, or hypothermic, the typically silent sickle cell trait transforms into a syndrome that resembles sickle cell disease with widespread sickling and subsequent vaso-occlusion.

The presence or absence of intravascular sickled red blood cells in tissue specimens depends on the degree of oxygenation of the sample prior to fixation. Intravascular sickling may occur due to terminal hypoxemia in the setting of sickle cell trait. It is almost impossible to determine the role sickled cells may have played by the presence or absence of intravascular sickling in autopsy specimens.

The events surrounding a death due to exertional sickling will assist the pathologist in this diagnosis. Individuals with sickle cell trait who die without a history of intense exercise prior to death may pose a challenge to physicians in determining if the death is or is not related to their genetic condition. A third possibility exists in which an individual may not be known to carry the sickle cell trait until sickled cells are seen in biopsy or autopsy specimens.

Three decedents autopsied at the Harris County Medical Examiner's Office, ranging in age from 28 to 49 were found to have sickle cell trait. None of the individuals were known to have the trait and when family members were contacted, only one had any knowledge that this condition existed in their family. The sickle cell trait was found to be purely incidental in two of the three decedents and may or may not be related to the cause of death in the third individual.

Two of the decedents were black males who were found unresponsive at work, one outside and one at a desk. One was 39-years- old and the other was 49-years-old. Both had enlarged hearts with coronary artery atherosclerosis. Microscopic examination revealed sickled cells in the heart, liver, lungs, kidney, and brain of both men. Hemoglobin electrophoresis performed on postmortem blood revealed the presence of hemoglobins A, S, F, and A2 in levels suggestive of sickle cell trait with an underlying beta+ thalassemia.

The third case involved a 28-year-old morbidly obese black female (body mass index of 54.1) who became unresponsive shortly after complaining of shortness of breath and abdominal pain. Autopsy findings included bilateral pulmonary thromboemboli, deep venous thromboses, gallstones, and clear bile. Microscopic examination revealed sickled cells in the kidney liver and brain. Hemoglobin electrophoresis results are pending at this time. Family members were contacted and reported knowledge of sickle cell trait in a sibling, but not in the decedent.

All family members contacted were grateful for the information and most were planning a follow up visit with their physician to obtain testing for sickle cell trait. With the exception of possibly the pulmonary emboli, the finding of sickle cell trait was incidental to the determination of the cause and manner of death; however, the information was extremely important for the family members of the decedents. This finding underscores the responsibility of forensic pathologists to perform autopsies with the intent of complete and thorough documentation of all findings, not just determination of cause and manner of death.

Autopsy, Sickle Cell Trait, Incidental

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