G49 An Unusual Motorized Vehicle Fatality

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After attending this presentation, attendees will understand the clinical features, classification, and pathogenesis of Osteogenesis Imperfecta. The high risk that these patients have for fatal intracranial injury after relatively minor trauma. The common causes of death of these patients. The possible diagnostic confusion of this disease with child abuse

This presentation will impact the forensic community and/or humanity by making the forensic community become aware of the disease Osteogenesis Imperfecta, its clinical features, and the likelyhood of these patients to die after what would normally be considered minor trauma. The forensic community will also become aware of the typical causes of death of these patients and the possible confusion of this disease with child abuse in young patients. The typical external features and radigraphic appearance of the disease will be shown so that the forensic community will be able to reocognize patients with Osteogenesis Imperfecta in the future.

The goals of this presentation are to present a case that summarizes the typical clinical features of osteogenesis imperfecta (OI) and that highlight the high risk that these patients have for fatal cranial injury following relatively minor trauma.

The decedent was a 20-year-old female with a past medical history of OI who was living independently at a local college. She was riding in her motorized wheelchair on a level concrete path at a unknown speed when it came to a sudden unexpected stop. She was unrestrained and was thrown forward out of the wheelchair. The left side of her head impacted the ground. There was no loss of consciousness or altered mental status at the time of the accident. She was transported to a local hospital where physical examination revealed a small scalp laceration and blood in her left external ear canal. Extreme body dysmorphism consistent with the history of OI was noted, but no new trauma to the extremities was identified. A computerized tomography (CT) scan of the brain revealed a large epidural hematoma and multiple fractures through the left side of the calvarium. Approximately 5 hours after the accident, her level of consciousness decreased. A repeat head CT confirmed a left sided epidural hematoma now with significant midline shift and subfalcine herniation. An emergent craniotomy was performed for evacuation of the epidural hematoma. She died soon after surgery.

Postmortem examination revealed the head to be disproportionately large for the body. Reflection of the scalp revealed a complex comminuted left temporal bone fracture status post repair. The bones of the skull were “egg shell” thin. A 10 x 6 cm temporal-parietal epidural blood clot compressed the underlying brain. A thin layer of subarachnoid hemorrhage overlay the cerebral convexities. Marked cerebral edema was associated with bilateral herniation of the parahippocampal gyri and cerebellar tonsils. Sectioning of the brain disclosed left to right midline shift and subfalcine herniation. Additionally, the sclera were blue and soft. The teeth were opalescent, brownish, and chipped. Both upper extremities were dysmorphic and tortuous. The lower extremities exhibited marked bowing and deformity. Radiographs revealed severe scoliosis of the spine with placement of a fixation rod. The legs contained internal fixation rods in both femurs and tibias. Representative samples of bone from the ribs and spine were soft and friable. Microscopic examination showed decreased amounts of bone that were disorganized and only focally calcified.

OI (“brittle bone disease”) is a heterogeneous genetic disorder of type I collagen. Affected individuals have fragile bones and abnormalities in other tissues rich in type I collagen including teeth, sclera, and ligaments. The disease is due to mutations in the COL1A1 or COL1A2 genes that encode type I procollagen. The disease is divided into types 1 – 4 based on skeletal abnormalities, the extra-skeletal tissue affected, and the genetic defect present. Type I OI is a mild form of the disease characterized by less severe bone involvement, blue sclera, deafness, and variable involvement of teeth (dentinogenesis imperfecta). The most severe form of OI, type II, presents with extreme bone fragility, intrauterine fractures, crumpled long bones and ribs, severe deformity, blue sclera, and is usually fatal in the perinatal period. Type III OI is characterized by progressive bone deformities, frequent fractures, short stature, scoliosis, deafness, dentinogenesis imperfecta, variably hued sclera, and survival often into adolescence and young adulthood. Type IV OI is intermediate in severity between types I and III, and it is characterized by moderate bone fragility and deformity, deafness, variable dentinogenesis imperfecta, and normal sclera.

McAllion and Colin (1996) have reviewed the causes of death for non-type II OI patients. Patients with types I and IV OI had increased numbers of deaths due to respiratory complications of their disease, as well as compression of the brain due to basilar invagination of the skull. However, many type I and IV OI patients had a normal lifespan and died due to diseases that affect the community at large. The type III OI patients often died of respiratory complications of their disease. Five percent of the type III OI patients died due to cranial injury after falling out of a wheelchair.

A concern for the forensic community is the possible confusion of child abuse with OI since both can present with unexplained fractures. It has been estimated that up to 1% of infants presenting with fractures

in the first year of life have OI (Byers, 2000). This highlights the need for accurate recognition of OI. The usual other signs of abuse including lacerations, burns, retinal and intracranial hemorrhages, and signs of sexual trauma should be sought. Laboratory evaluation of collagen, imaging studies of bone, and genetic analysis may provide support for a diagnosis of OI.

This case presentation highlights the increased risk that OI patients have for fatal intracranial trauma after relatively mild traumatic injury. Because of this risk, OI patients should always wear a seat belt when riding in a wheelchair.

Osteogenesis Imperfecta, Epidural Hematoma, Trauma

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