G9 Sudden Unexpected Death in an Achondroplastic Dwarf

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After attending this presentation, attendees will be able to describe the anatomic features of achondroplastic dwarfism and list causes of sudden death in this patient population.

This presentation will impact the forensic science community by discussing a case identified as a pathologic substrate for sudden death in achondroplastic dwarfism, which may be more common than currently recognized. This case also emphasizes susceptibility to sudden death in this patient population and may assist in assigning the proper manner of death.

Achondroplastic dwarfism, the most common form of dwarfism, is a syndrome of characteristic anatomic findings resulting from a genetic mutation. The findings are predominantly skeletal and consist of an enlarged skull with a small skull base, a narrowed foramen magnum and spinal canal, short, flat, vertebral bodies, thickened intervertebral disks, shortened tubular bones and ribs with metaphyseal cupping, and shortened metacarpals and phalanges, among others.

This study presents the case of a 3-year-old boy with a past medical history significant for achondroplastic dwarfism. The decedent was at home with his two siblings in the care of his mother when he suffered a mild fall from a plastic step stool, causing him to fall backward and strike his head. Investigation revealed no history or suspicion of abuse.

Examination at autopsy revealed an enlarged cranium and short stature. A full skeletal survey confirmed the diagnosis of dwarfism and revealed no fractures. External examination revealed two green contusions on the forehead measuring 1.2 x 0.2cm and 2.0 x 0.4cm. There were no other external injuries. Autopsy examination was unremarkable aside from the achondroplasia, external trauma related to the fall, and a syrinx in the caudal medulla. No other traumatic lesions, recent or remote, were identified externally or internally.

Achondroplastic dwarfs are at increased risk of sudden death, although the anatomic basis for this is unclear and has been addressed only obliquely in the literature. One report by Mohindra et al suggests atlanto-occipital instability as a cause of death in these patients.[1] A study by Hecht et al found that there is an increased risk of sudden death in these patients which is attributable to brain stem compression in patients less than 4 years of age, while spinal stenosis contributed to the cause of death more frequently in older patients.[2] The findings in this case suggested that skeletal abnormalities associated with achondroplasia, in particular a narrow foramen magnum, led to a syrinx in the lower medulla near vital cardiac and respiratory centers, increasing the likelihood that minimal trauma could lead to apnea and sudden death. Of note, lesions would go unnoticed unless sampled for histology. This case highlights the possibility that syringobulbia may be more common in achondroplasia than currently recognized and may represent a significant substrate for sudden death in these patients.

References:

1. Mohindra S, Tripathi M, Arora S. Atlanto-Axial Instability in Achondroplastic Dwarfs: A Report of Two Cases and Literature Review. Ped Neurosurg 2011: 47(4): 284-287.
2. Hecht J, Francomano C, Horton W, Annegers J. Mortality in Achondroplasia. Am J Hum Genet 1987: 41:454-464.

Achondroplastic Dwarf, Sudden Death, Syringobulbia

 ** Presenting Author*