G53 Corpora Amylacea and Sudden Death: A Case of Adult Polyglucosan Body Disease Diagnosed at Forensic Autopsy

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After attending this presentation, attendees will learn about a case of adult polyglucosan body disease diagnosed at forensic autopsy.

This presentation will impact the forensic community by educating them about the first reported case of adult polyglucosan body disease presenting as sudden death and diagnosed at forensic autopsy.

Adult polyglucosan body disease (APBD) is a rare neurodegenerative condition characterized by typical onset in middle age, progressive neurological impairment that is heterogeneous between those affected, and death within 1-14 years of diagnosis. The histopathological hallmark of the disease is massive deposition of corpora amylacea (designated polyglucosan bodies in this context) in the central nervous system, and variable deposition of similar material in other sites. While the cause of the disease is as yet unknown, recent research has identified mutations in proteins involved in glycogen metabolism in a subset of cases. Some of these mutations are similar to mutations identified in cases of glycogen storage disease type IV (GSD IV), a disease that classically is present in the first year of life, is of very heterogeneous manifestation, and is also characterized by massive deposition of corpora amylacea. The genetic and histopathological similarities between these two conditions have lead to speculations that APBD may represent an adult form of GSD IV.

In this presentation, a case of sudden death is presented wherein APBD was diagnosed at forensic autopsy. Scene details, relevant medical and social history, and autopsy and histopathological findings are presented and richly illustrated with supporting images.

The case provides an excellent example of a prolonged and enigmatic presentation involving a complex interplay of medical, social, and forensic issues. The histopathology is particularly illustrative of this rare disease, showing massive deposition of corpora amylacea in the central nervous system, and marked accumulation of similar material in the heart. The latter was determined to be the mechanism of death (cardiac arrhythmia) with APBD the underlying cause.

This presentation represents the first case of APBD reported in a forensic context. APBD is reviewed and its relationship with other diseases characterized by massive deposition of corpora amylacea is outlined. The role of forensic autopsies in the diagnosis of rare conditions is discussed.

Neuropathology, Corpora Amylacea, Sudden Death

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