



H53 Madelung's Disease: A Rare and Benign Life-Threatening Condition

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Learning Overview: After attending this presentation, attendees will understand about a rare and benign condition called Madelung's Disease, which can mimic head and neck malignancy and add an asphyxial component to the cause of death.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by describing this rare disorder of unknown etiology, also called Launois-Bensaude Syndrome and Multiple Symmetric Lipomatosis, and provide tools for its diagnosis. The forensic implications to the cause of death are also relevant.

Madelung's disease is characterized by a prominent and symmetric deposition of non-encapsulated adipose tissue, preferentially on the upper region of the body. Symptoms depend on the affected body area. With only 200-300 cases described worldwide until 2015 and an incidence of 1/250,000, it is more frequently observed in middle-aged men with Mediterranean ancestry, strongly associated to chronic alcoholism. Forensically, the excess of fat accumulation around the neck is the most striking feature, because it may compress the aero-digestive tract, and/or the lipomatosis aggregates may undergo malignant transformation. In such cases, surgical excision or liposuction with removal of the entire lesion could be attempted.

Reported here is a case of a 48-year-old man with a history of heavy alcoholism, oligophrenia, and Madelung's Disease admitted to a mental health department for alcohol detoxification and posterior surgical treatment, who died suddenly when sleeping. The patient exhibited a good general condition, BMI of 28.7, and large adipose neoformations on the submandibular, cervical, supraclavicular regions, and upper arms. The patient had previously been submitted to surgery for this disorder.

The autopsy showed, apart from the above-mentioned external lesions, an enlarged amount of anomalous deposition of adipose tissue surrounding the aero-digestive tract with compression of laryngeal structures, although still permeable. Anatomico-pathological examination revealed benign subcutaneous and soft tissue lipomatosis that compressed and deformed the local structures, affecting their natural functions (as shown by marked distention of alveolar spaces and pulmonary infection). Toxicology was negative for alcohol, revealed anti-epileptic, neuroleptic, and benzodiazepines in therapeutic concentrations. The cause of death was acute bronchopneumonia with a probable contribution of the cervical lipomatosis to respiratory compromise.

Forensic pathologists should be aware of the characteristic appearance of this unique and rare disorder, scarcely described in forensic literature, as an important clue to recognize associated risk factors, such as alcoholism, hyperglycemia, hyperlipidemia, diabetes, and polyneuropathy. It is also important to evaluate, for each case, its role in an eventual asphyxial death.

Madelung's Disease, Multiple Symmetric Lipomatosis, Asphyxial Deaths