G44 An Unusual Case of Whipple's Disease With Fatal Outcome in a Young Woman: A Rare Disease and Diagnosis Failure

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After attending this presentation, attendees will gain knowledge on macro and microscopic aspects of the rare disease, Whipple's disease, in order to make the difficult diagnostic process of this illness easier.

This presentation will impact the forensic science community by showing how a "diagnosis failure" which does not always imply a professional negligence; it stresses the importance of not relying only on the epidemiological evidence for the diagnostic prediction of the disease.

Introduction: Whipple's disease is a rare, systemic infectious disease caused by the bacterium *Tropheryma whipplei*. First described by G.H. Whipple in 1907 and commonly considered a gastrointestinal disorder, Whipple's disease primarily causes malabsorption but may affect any part of the body including the heart, lungs, brain, joints, skin, and eyes. Weight loss, diarrhea, joint pain, and arthritis are commonly presented symptoms, but their presentation can be highly variable and approximately 15% of patients do not have these classic signs and symptoms. Whipple's disease is significantly more common in men (87% male) and the incidence has been estimated in around 1/1,000,000, even if no valid estimate of the incidence is available. The disorder has been described most frequently in white people and in Western Europe. For these reasons, in Italy the disease is naturally placed on the National Register of Rare Diseases (RNMR) of ISS.

When recognized and treated, Whipple's disease can usually be cured with long-term antibiotic therapy; untreated the disease is ultimately fatal.

The case: The case of a 27-year-old woman who died six days after admission in a Sicilian hospital for persistent fever, anaemia, and weight loss is presented. For the symptoms presented, doctors evaluated a broad spectrum of diagnoses, not identifying this rare disease. For this reason, the court ordered an autopsy, to clarify the causes of death and any professional liability. The autopsy, together with the histological and microbiological investigation, allowed the diagnosis of Whipple's disease, with systemic manifestation and predominant intestinal involvement (lipodystrophy), heart (fibroadhesive pericardial disease), brain (lipid thesaurismosis), pulmonary and renal fat embolism and erythrophagocytosis.

Discussion and Conclusions: After explaining all aspects of forensic pathology, accompanied by macro and microscopic investigations, the exclusion of other causes of death, attention was focused on the concept of "diagnosis failure" of a rare condition such as Whipple's disease, which does not always imply a professional negligence, as in this case.

What is stated above, doesn't want to mean that a "rare disease" is the same as a "failed diagnosis", rather it wants to point out that a rare illness must be carefully investigated and therefore needs a manifold elaboration "differential diagnostic path" (with other more common pathological conditions) which require more time and more specific examination.

Moreover, it stresses the importance of not relying only on the epidemiological evidence for the diagnostic prediction of the disease.

Whipple's Disease, Diagnosis Failure, Autopsy

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