G23 Postmortem Diagnosis of Marfan's Syndrome In Pregnancy: Cause of Death or Incidental Finding?

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After attending this presentation, attendees will have a better understanding of the importance of an adequate Marfans Syndrome (MS) diagnosis to realize an appropriate monitoring of pregnant patients to prevent and control pregnancy-related risk.

This presentation will impact the forensic science community by highlighting prevention pregnancy-related risk in Marfans syndrome.

Introduction: In the vast area of connective tissue diseases are also counted MS, Loeys Dietz Syndrome (LDS), and the Ehlers-Danlos syndrome. These diseases are all characterized by mutations of genes that encode proteins that are the constitutive elements of elastic fibers and that determine elasticity alterations of the involved tissues. Marfan's syndrome (OMIM #154700) is an inherited, autosomal dominant disorder that affects the skeletal, ocular, and cardiovascular systems. The disease displays high penetrance and wide clinical variability both within and between families, therefore a patient may have mild to severe symptoms that may or may not correlate with other affected family members. Although Marfan's syndrome has a range of characteristic morphological features involving the ocular, cardiovascular, and musculoskeletal systems, the phenotype is variable. In addition, mutations have been identified in the gene encoding for fibrillin-1 and also in the transforming growth factor-b receptor 2 (TGF-bR2) gene. In the postmortem diagnosis of these pathologies, it is necessary to detect external anatomical characteristics and peculiar internal signs. Common phenotypic findings are muscle-skeletal and cutaneous alterations. Among the internal signs are found ocular, cardiovascular abnormalities, and pulmonary manifestations. The most common internal signs of vascular origin are structural abnormalities of the aorta. The pregnancy is, for these patients, a high risk event, especially in consideration of the anatomical and structural anomalies that the high weight involves, as well as the intrinsic risks to the pregnancy state.

Case Report: The case of a young woman, pregnant, at the 37 weeks of pregnancy, with a weight of 129kg, operated with a caesarean section, who died two days later for unknown causes was analyzed. The external inspection of the corpse showed the presence of *facies lunaris*, wide attack of the palate, cutaneous striations, and muscle-skeletal anomalies in correspondence of the right hand. The autopsy stated pituitary hyperplasia, thymic hyperplasia, massive pulmonary embolism, and complete aortic dissection, type A in Stanford classification. Histological examination showed a complete degeneration of the tunica media of vessels affected by the dissection.

Results: In the studied case, the postmortem diagnosis of connective tissue disease, in the absence of aorta rupture, has not been only an accidental finding but a pathological, pre-existing, and concurrent (dissection) cause of the pulmonary thrombo-embolic event, regarded as the cause of death of the young woman.

Conclusions: It is possible to say that pregnancy in obese patients with connective tissue diseases, particularly in patients with aortic ectasia, presents a high risk of mortality and morbidity in relation to the genesis of aortic dissection. For this reason, a correct and adequate MS diagnosis is fundamental, to prevent pregnancy-related risk and realize an adequate monitoring during pregnancy through implementation of a proper diet and timely eco-cardiotographic controls.

Marfan's Syndrome, Pregnancy, Aortic Dissection

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