G93 Maternal Congenital Antithrombin-III Deficiency in an Intrauterine Fetal Death

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The goal of this presentation is to show how fetal autopsy and a careful placental examination as well as a complete genetic study, on both mother and fetus in this case, play a substantial role not only in identifying the cause of death and in serving justice, but also as a means of helping clinical and forensic practice.

This presentation will impact the forensic science community by showing how fetal autopsies should be performed following recommended protocol and should include a careful placental examination and genetic analysis.

Thrombophilia is a multigenetic disorder caused by an inherited and acquired defect and has been described as a predisposition to thrombosis. There has been growing interest in thrombophilia and its connection to the pathogeneses of certain pregnancy complications (such as gestational losses) because of the prothrombotic state it creates. In normal pregnancy, thrombotic risk increases and has been characterized as an evolutionary advantage against severe blood loss after delivery and placenta separation. A recent hypothesis has suggested, however, that maternal thrombotic predisposition could interfere with the initial development of an adequate uteroplacental circulation and may result in the production of microthrombosis in the placental vessels. Massive perivillous fibrin deposition (MPFD) and the related lesion maternal floor infarction (MFI) are rare but serious causes of placental insufficiency, occurring in 0.03 e 0.5% of deliveries.

The presented case concerns about an intrauterine fetal death at 30-weeks gestation in a 24-year-old woman who was affected by congenital antithrombin-III deficiency, with previous family history of venous thrombosis and venous thromboembolism, and who was receiving anticoagulant prophylaxis during pregnancy. During her pregnancy, a reduction of physiological protein S and antithrombin were reported and the woman was subsequently treated pharmacologically by her gynecologist.

At 30-weeks gestation, the woman was admitted to the department of gynecology after noting a few days with no movement from her fetus. After a rapid clinical and echographic study showed a typical image of "Spalding" (overlap of the fetus's cranial bones), the woman was induced and then vaginally delivered the lifeless fetus.

A complete autopsy of the fetus was performed 48-hours after death. The external examination revealed the presence of maceration over up to 40% of the fetal surface. The autopsy excluded the occurrence of acute and significant abnormalities in all fetal organs.

A complete examination of the placenta (size: 16x15x4cm, weight: 560 grams) was performed after fixation in buffered formalin. Upon external examination, a series of voluminous, fluid-filled serous cysts were noted to be occupying a total area of 12x10x5cm. On coronal sections, the peripheral portion of the placenta showed multiple whitish-bluish, hard areas, 16x4cm wide. At the point of insertion of the umbilical cord, two cysts were observed, each 1.5x1.0cm in size.

The etiopathogenetic definition was outlined by histological examinations, which were preformed on placenta tissue samples using haematoxylin-eosin (H&E), Trichrome stain and Perl's, and revealed an intraluminal thrombosis of placental vessels with signs of villa ischemia and massive perivillous fibrin deposition (MPVFD).

The intrauterine fetal death was consequently attributed to acute respiratory failure (hypoxia fetal) by placental thrombosis secondary to maternal thrombophilia. A careful placental examination as well as a complete genetic study, on both mother and fetus in this case, therefore played an important role in identifying the possible cause of intrauterine fetal death. These practices remain important so that such data may help to advise parents who are considering whether or not to consent to a postmortem examination.

Intrauterine Death, Antithrombin-III Deficiency, Placental Examination

