Objectives: To investigate the association between a low PAPP-A (≤ 0.2 MoM) and the risk of structural abnormalities in the first trimester of pregnancy.

Methods: In this study first trimester screening for chromosomal abnormalities was performed according to FMF guidelines and biochemical analysis is performed using Kryptor Brahms, as provided from the hospital protocol. An invasive procedure was offered in cases with an increased risk for aneuploidies. In cases of PAPP-A ≤ 0.2 MoM and normal karyotype, an ultrasonographic (US) follow-up until 32 weeks gestation was performed to investigate the risk of placental insufficiency.

Results: Since June 2009 to January 2019, 147 patients with low PAPP-A (≤ 0.2 MoM) were found. One hundred forty-two were singleton pregnancies and 5 patients had a twin pregnancy. Twin pregnancies were complicated by a TRAP sequence in 1 case, by a spontaneous abortion of 1 twin in 1 case, and by a structural anomaly of one twin in 1 case. Fetal karyotype was performed in 107 cases, and 48 cases had an abnormal karyotype (trisomy 21 in 18 cases, trisomy 18 in 13, trisomy 13 in 3, triploidy in 9, placental or fetal mosaicism in 5 cases and in 1 case an abnormality at aCGH). In 40 cases an invasive procedure was declined and most of the patients were lost to follow-up. In 59 cases fetal karyotype was normal; however US follow-up found a structural anomaly in 7 cases (12%). In particular a congenital heart disease in 4 cases, a ventriculomegaly in 2 cases and an omphalocele in 1 case were diagnosed.

Conclusions: A low PAPP-A (≤ 0.2 MoM) is a rare condition, well known to be associated with an increased risk for chromosomal abnormalities and placental disfunction like IUGR, stillbirth and abortion. However a careful anatomical evaluation is recommended as there is an increased risk of structural abnormalities.