AN ANTENATAL DIAGNOSIS OF KLINEFELTER SYNDROME- A CASE REPORT

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ABSTRACT:
Klinefelter Syndrome (47, XXY) is the most common sex chromosome disorder, characterized by at least one extra X chromosome to a normal male karyotype, and affects one in 600 males.

The incidence of an antenatal diagnosis is very low, described approximately 0.15%, once there are no ultrasound markers for the diagnosis. In the majority of cases the identification of the fetus with KS is incidental, during Down’s Syndrome screening at the first trimester of pregnancy (increased nuchal translucency as an ultrasound marker, lower PAPP-A than usual as a biochemical marker, advanced maternal age and maternal request).

RESULTS:
The patient delivered at term (38 weeks of gestation) by cesarean section (for option), a newborn with 2910g of birthweight, Apgar 9-10, with no abnormal characteristics. A literature review shows the dilemma of the antenatal Klinefelter Syndrome diagnosis. The major problem is the genetic counseling, the main issue to the parent’s decision to continue the pregnancy. Studies have been showed optimism in prenatal diagnosis, resulting in individuals with phenotype less severe than in those diagnosed postnatally during the childhood, who faced an adequate treatment.

CONCLUSION:
This case of Klinefelter Syndrome was presented here to emphasize that the antenatal diagnosis can be done, even with normal first trimester screening for aneuploidies.

A multidisciplinary center for this difficult prenatal diagnosis is necessary to support the parents, since the counseling until an early referral to a specialist as a form to minimize the effects of probable comorbidities.

CASE REPORT:
A 38-year-old woman, primigravida, conceived spontaneously, presented at 12 weeks and 2 days of gestation for the first trimester screening for Down’s Syndrome. The risk assessment showed normal biophysical markers (nuchal translucency, nasal bone, tricuspid regurgitation and ductus venosus) and biochemical markers (free ßHCG and PAPP-A) with low risk result for aneuploidies. At the moment of the anomaly scan, with 20 weeks and 5 days of pregnancy, all markers were normal as well, a part of a nasal bone at the 5th centile. The patient presented at 29 weeks and 5 days of gestation for a growth scan and the fetal profile was not usual and the nasal bone was below the 5th centile. At this moment we recommend a fetal karyotype and the parents opted for non-invasive prenatal test (NIPT). A fetal echocardiograph was performed with normal result. The NIPT result showed an abnormal sex chromosome, 47XXY (Klinefelter Syndrome).