Prenatal diagnosis of double-aneuploidy: Down- Klinefelter Syndrome.
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Introduction
Down - Klinefelter Syndrome is very rare, incidence is calculated in 0.4 - 0.9/10000 male newborn and 11.7/10000 cases of Down Syndrome. 30% of all the cases reported until now have been suspected by prenatal testing, with abnormal findings in first trimester ecography associated with Down Syndrome. Most of cases have been associated with advanced maternal age.

Case report
OES 38 years old. GIII CII. GI 2010, GII 2012.
GIII LMP: November 2013. First visit in 30-Jan-2014. Single pregnancy 8.5 weeks by CCL. First trimester screening on 01-March-2014 increased nuchal translucency 3.7mm, Nasal bone present (Fig1) Ductus venosus normal.
FETALTEST software reports increased risk 1/2 for Down Syndrome and 1/6 por 18 Trisomy.
The patient decided to perform NIPT (Fetal DNA in maternal blood)

Results
The result of the analysis of fetal DNA in maternal blood reports HIGH RISK for Down Syndrome and Klinefelter Syndrome. The patient decided interruption of pregnancy and fetal tissue have been send for definitive study and diagnosis. Cariotype reports 48 XXY +21 (Fig2)

Conclusion
Increased Nuchal Translucency is the most important finding in the first trimester screening. It is necessary to perform complementary studies to make the definitive diagnosis. The detection rate of combined first trimester screening is 97% with a false positive rate of 5%. The analysis of Fetal DNA in maternal fetal blood has a detection rate of 99.9% for Down Syndrome with 0.01% of false positive rate. In this case report, the cariotype confirms the report the Fetal DNA test.