Objectives
Noonan syndrome (NS) is the most commonly reported single gene disorder associated with nuchal translucency (NT) above 99th centile in the I Trimester. The aim of this study was to evaluate how the introduction of a systematic screening protocol could increase prenatal diagnoses of NS.

Methods
From April 2017 all the fetuses with with NT ≥ 3.5 mm in the I Trim were tested for standard karyotype and array-CGH. Those with normal karyotype and nuchal fold (NF) thickness ≥ 6 mm at 16 wks were also tested for NS panel (PTPN11, SOS1, BRAF, RAF1, MAP2K1, MAP2K2, KRAS, NRAS, SHOC2, HRAS, RT1, CBL, LZTR1 and SOS2 genes). Pregnancy outcome was recorded for all cases.

Results
Nine fetuses were included in the study. Median NT measurement was 6.4 mm (range 3.5-10.9 mm). Median NF was 7.6 mm (range 6-11.8 mm). Two fetuses were positive for NS (2/9, 22%) and the median NT value in this subgroup was higher compared to the 7 NS-negative fetuses (8.7 vs 5.3 mm, p= 0.04), while no difference was found for NF value. Figures 1, 2 and 3 show US appearance of one case diagnosed with of NS.

Conclusions
A systematic screening protocol allows prenatal identification of Noonan Syndrome with an incidence of 22% in screen positive fetuses (euploids with persistent lymphatic disorder).