A case of Pallister-Killian Syndrome in the first trimester

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Objective
We demonstrate this rare case report of tetrasomy 12p (Pallister-Killian Syndrome) presenting with increased nuchal translucency and other markers for chromosomal abnormalities in the routine first trimester scan.

Methods
We present this case report in the light of current literature.

Results
Clinical Presentation: A 32-year-old woman, G4P0, with 3 previous miscarriages was referred to our Fetal Medicine Unit at 12 weeks and 5 days because of increased nuchal translucency noted on the routine first trimester scan. The patient was screened positive for Down Syndrome with a risk of 1 in 5. The risk for Patau's/Edward's Syndromes was 1 in 7691. PAPP-A was 1.08 MoMs and β-hcg 1.1 MoMs. Ultrasonographic examination confirmed increased nuchal translucency of 9 mm, and revealed further findings such as skin edema, frontal bossing, short long bones, abnormal four chambers view and absent nasal bone. On the basis of these results, after counselling about the poor prognosis, invasive testing and termination of pregnancy were offered and the couple opted for termination of pregnancy at 13 weeks and 5 days of gestation. The post mortem examination, using DNA extracted from uncultured cord, showed that there were two additional copies of the subtelomeric region of the short arm of chromosome 12. The QF-PCR analysis using the Devyser Extend kit V2 indicated that there are not additional detectable abnormalities. This interpretation is compatible with the Pallister-Killian Syndrome and the presenting clinical details. Background: Pallister-Killian Syndrome, a rare chromosomal abnormality (tetrasomy of 12p) was first described by Pallister in 1977. It has been shown that there is link to a wide range of abnormalities, like congenital heart defects, short bones, congenital diaphragmatic hernia, urogenital abnormalities, facial defects, mental retardation, abnormal skin pigmentation and short neck. The mechanism which causes this chromosomal abnormality is not clear. Maybe it is related to parental age and most probably to maternal age concerning the formation of isochromosome 12p in PKS. The antenatal diagnosis is usually an incidental finding after karyotyping for maternal age and the presence of fetal defects during the ultrasound examination.

Conclusion
This case reports the Pallister-Killian syndrome presenting with high risk screening for Down’s syndrome and the co-existence of markers for chromosomal abnormalities. The diagnosis was made on the detection of increased nuchal translucency, whereas this syndrome is usually diagnosed later in the pregnancy with the presence of fetal abnormalities as described above. Thus, if the patient didn't have the first trimester scan, the syndrome could not have been identified until 20 weeks of gestation. This case raises the overall number of chromosomal abnormalities that have been found in fetuses with increased nuchal translucency.