Clinodactyly associated with Trisomy 21: A case report

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Objective
Trisomy 21 (Down syndrome) is the most common trisomy in live-born infants. The incidence of Down syndrome changes depending on the population, in live births it is between 1: 319 and 1: 1000. The risk of trisomy 21 increases with increasing maternal age and decreases with advancing gestation. In most cases trisomy 21 is due to maternal meiotic nondisjunction (95%). Of the remaining 5%, unbalanced translocation (3-4%) and mosaicism (1%) is held accountable. Here we present a case of Trisomy 21 detected antenatally with sonographic markers.

Methods
This is a case report.

Results
A-19 year-old gravida 2, miscarriage 1, para 0 woman was referred to our clinic at 21 weeks of gestation because of fetal structural abnormalities on prenatal ultrasound. She had no remarkable medical, obstetric or family history. First trimester screening was low risk. In our detailed ultrasound examination at 21 weeks, we detected increased nuchal fold, clinodactyly and nasal bone hypoplasia. The family received genetic counseling and offered invasive diagnostic testing for karyotype. She underwent amniocentesis, which revealed a 47, XY, +21 (Down’s Syndrome) karyotype. After consultation, the family chose to terminate the pregnancy at 23 weeks.

Conclusion
Many major structural abnormalities and minor markers of aneuploidy may be associated with Down’s Syndrome. Findings may include increased nuchal fold, ventriculomegaly, echogenic intracardiac foci, pyelectasis, echogenic bowel, enlarged cisterna magna, liver calcification, and digit anomalies (polydactyly, clinodactyly, sandal gap, and clubfoot). Patients with a major or minor marker for Down syndrome should be offered invasive diagnostic testing with amniocentesis or chorionic villus sampling. Termination of pregnancy is an option.