



## Characteristics and outcome of fetal nuchal translucency above 3.5 mm in the first trimester

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### Objective

To describe the natural history of fetuses with a nuchal translucency (NT) above 3.5 mm when the crown-rump length measures between 45 and 84 mm.

### Methods

We performed a retrospective cohort study of fetuses with first trimester NT above 3.5 mm between January 2013 and March 2017.

### Results

A total of 75 cases with NT > 3.5 mm in the first trimester were identified. 3 cases were excluded (lost to follow-up), 72 cases were included. Additional first trimester markers of aneuploidy in 16 cases and structural abnormalities in 5 cases were visible. 2 women declined invasive testing. The karyotype was abnormal in 43% (30/70), including 17 cases of trisomy 21. There were 36 livebirths (51%), 24 terminations of pregnancy (34.3%) and 12 fetal deaths (16.7%). The 36 live-born infants were followed up. There were 3 cases of trisomy 21, 1 case of unilateral hypoplasia of orbicularis oris, 1 Noonan-like syndrome, 1 case of 8p23.1 duplication syndrome and 1 case of lower limb lymphedema.

### Conclusion

The prognosis of fetal NT above 3.5 mm in the first trimester is poor when associated with karyotype abnormalities or structural abnormalities but is substantially better if neither of these conditions are observed.