Fetal prognosis and neonatal outcome in fetuses with increased nuchal translucency and normal karyotype: a retrospective study

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Objectives:

The objectives of this study are to compare the incidence of fetal anomalies and neonatal outcome in relation to the degree of increased nuchal translucency (NT) in fetuses with a normal karyotype.

Methods:

Singleton pregnancies whose fetuses were detected with a nuchal translucency greater than 3mm at the first trimester ultrasound examination were included. The study was conducted by the Department of Prenatal Diagnosis of San Paolo Hospital in Milan from 2016 to 2022. Based on the degree of nuchal translucency they were divided into two groups:

- group 1: NT between 3 and 3,5mm (42/104 (40,4%))
- group 2: NT above 3,5 mm (62/104 (59,6%)).

Epidemiological data, ultrasound follow-ups and neonatal outcome were analyzed.
The statistical analysis was performed with SPSS, the linear variability with t test, while the nominal variables were analyzed with chi-square test.

Results:

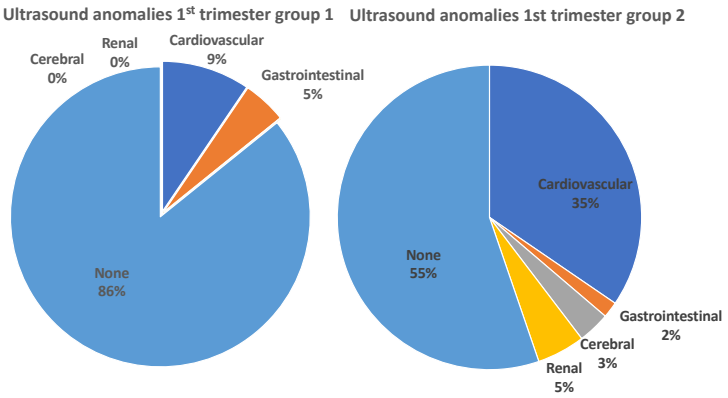
The characteristics of the population in terms of age, BMI, race, habits and parity are reported below and were homogenous among the two groups, without any statistical significance.

	NT 3-3,5 mm	NT>3,5 mm
Age	34,7 (5,5) years	34,8 (5,4) years
BMI	25,4, kg/m2	23,1 (5) kg/m2
Caucasic	37/42 (88,1%)	52/62 (83,9%)
Smokers	1/36 (2,3%)	1/46 (2,2%)
Multiparous	15/36 (42,9%)	16/46 (37%)

The ultrasound differences during first trimester examination among group 1 and 2 are described below. The pulsatility index of the ductus venosus was statistically significant, together with hypoplasia of the nasal bone, regurgitation of the tricuspid valve and anatomical anomalies.

	NT 3-3,5 mm	NT>3,5 mm	p value
CRL	64,7 (9,1) mm	59,1 (10,9) mm	
DV PI	1,13	1,56	0,01
Hypoplasia NB	2/41 (4,9%)	15/55 (27,3%)	0,01
Regurgitation tricuspid valve	2/39 (5,1%)	12/52 (23,1%)	0,01
Anatomical anomalies	6/42 (14,3%)	34/62 (54,8%)	0,01

For what concerns major anomalies on the first trimester ultrasound, the most common finding was cardiac anomaly, which was detected in both groups, but significantly more prevalent in the group with nuchal translucency above 3,5 mm (p<0,01). Gastrointestinal, renal and cerebral anomalies were rare, and not statistically significant among the two groups.



Among the women with fetuses with increased nuchal translucency 20/42 (47,6%) in group 1 and 55/62 (88,7%) in group 2 underwent an invasive procedure. 8/20(40%) in group 1 and 28/55 (50,9%) in group 2 resulted in a pathologic karyotype. 8/8 in group 1 and 27/28 in group 2 decided to terminate pregnancy.

Among the fetuses with a normal karyotype, on the second trimester ultrasound scan, 3/34 (8,8%) in group 1 and 0/29 (0%) in group 2 had gastrointestinal anomalies; 1/34 (2,9%) in group 1 and 3/29 (10,3%) in group 2 had renal anomalies; 1/34 (2,9%) in group 1 and 4/29 (13,8%) in group 2 had cerebral anomalies, 5/34 (14,7%) in group 1 and 9/29 (31%) in group 2 had cardiac anomalies. All of them were not statistically significant. The majority of the anomalies were minor and 5 patients of group 2 were lost in the follow up.

On the third trimester ultrasound, most of the anomalies were not present anymore: 1/34 (2,9%) in group 1 and 2/29 (6,9%) in group 2 had renal anomalies, 1/34 (2,9%) in group 1 and 2/29 (6,9%) in group 2 had cardiac anomalies.

	NT 3-3,5 mm	Nt>3,5 mm	P value
Gestational age	39,15±1,1 wk	39,5±0,9 wk	n.s.
Newborn weight	3385±387 g	3519±275 g	n.s.
Apgar score≥ 7	100%	100%	n.s.
NICU admissions	0%	0%	n.s.
PH umbilical cord>7	100%	100%	n.s.

The mean gestational age at birth among the two groups was 39,15±1,14 weeks and 39,5±0,9 weeks, the mean newborn weight was 3385±387 g and 3519±275 g. All the results were not statistically significant. All newborns were born with Apgar score ≥ 7and EGA >7, none were admitted to NICU units. The patients who had major anomalies on the ultrasound scan were transferred to third level hospitals in order to be managed properly.

Conclusions:

The US markers of aneuploidies were significantly increased in group with NT>3,5 mm, as described in the literature. Cardiac anomalies were more prevalent on first trimester ultrasound scan among the patients with a NT>3,5 mm. Among the patients with a normal karyotype there were not significant differences in terms of ultrasound anomalies and neonatal outcomes among the 2 groups, therefore once an abnormal karyotype is excluded the prognosis is favorable.