PRENATAL CF-DNA TESTING FOR DOWN’S SYNDROME AND OTHER CHROMOSOMAL ABNORMALITIES – 5 YEAR’S EXPERIENCE IN CLINICAL PRACTICE

Darija Strah¹, Nina Ružić-Gorenjec ²

Objectives: The use of prenatal cf-DNA testing as a highly accurate screening method for aneuploidies is bringing radical changes in the field of fetal medicine. It results in decline of the number of invasive procedures in women with high risk (HR), based on maternal age and/or NT screening due to substitute for cf-DNA testing. The average age of women deciding to have cf-DNA testing has been similar in the last 4 years; however, the number of tests is increasing also in the low risk group due to high reassurance.

Methods: We present the results of prenatal cf-DNA testing in the period from 1. 1. 2013 until 30. 6. 2017. In our study, 314 pregnant women were included.

Results: The number of women having NT screening and adding cf-DNA testing has increased over the last 4 years (2013: 39, 2014: 70, 2015: 48, 2016: 157). The percentages raised from 1.45 % in 2013 to 9.13 % in 2016. 13.38 % of women, HR only due to their age (37 years or more) had tests in 2013, however 44.03 % decided to have tests in 2016. 5.65 % of women, HR for both age and T21 had tests in 2013, in 2016 46.30 % decided to undergo cf-DNA testing instead of invasive procedure. 4.24% women younger than 37 years decided to undergo cf-DNA testing regardless of low-risk result for T21.

Cf-DNA test after NT screening changed women’s decision for adding biochemistry test as a part of traditional screening tool. The percentage of combined test among all pregnant women remained around 30%. The main difference in the uptake appeared among women from 35 - 37 years. The percentage decreased from 82.86% in 2013 to 68.10 % in
2016. 48% of women of 37 years or more had the tests in 2013, yet 23.94% decided for it in 2016.

The average age of women with cf-DNA testing has been similar in the last 4 years (2013: 36.1 years, 2014: 36.2 years, 2015: 36.4 years and 2016: 36.3 years). In 2016, we have noticed significant increase in test numbers. Half of all our cf-DNA tests were performed in 2016. Out of total 157 pregnant women, 59 (37.58%) were of advanced maternal age (37 years or more); 36 (22.93%) were high risk for T21 based on prior screening. 84 (53.5%) women were high risk regarding age and prior screening. 62 (39.49%) had low risk for T21 regarding age and prior screening.

The number of invasive procedures in Slovenia is still high due to well-established NT screening performed from year 1998 on (5.71% in 2013, 5.3% in 2014 and 4.99% in 2015). The number of invasive procedures decreased in two age groups. 9.66% of women aged 35-37 years had a procedure in 2013 compared to 5.81% in year 2015. 37.24% of women aged 37 years or more had the procedure in 2013 compared to 30.58% in year 2015. We are still waiting for the data from year 2016, when the highest number of tests was performed. We expect further decrease in number of invasive procedures.

In general, cf-DNA testing yielded 100% sensitivity (95% CI: 63.06%-100%) and 99.66% specificity (95% CI: 98.14%-99.9%) with the PPV of 88.9% for all aneuploidies (95% CI: 51.75%-99.72%). Sensitivity and specificity values only for T21 amounted to 100%.

**Conclusions:** Our results confirmed that prenatal cf-DNA testing represents highly accurate approach in advanced screening of most common aneuploidies. The number of tests has been increasing; however, the average age of women, opting to have cf-DNA screening, remains the same. The expected decline in number of invasive procedures among HR pregnant women due to their age and/or prior NT
screening results is to be continued. However, the ultrasound evaluation of the fetus determines which further test or procedure will follow.

Institution 1 name: Strah Diagnostic Centre
Institution 1 country: Slovenia
Institution 1 town: Domzale

Institution 2 name: Institute for Biostatistics and Medical Informatics, Faculty of Medicine, University of Ljubljana
Institution 2 country: Slovenia
Institution 2 town: Ljubljana

Submitting name and surname: Darija Strah
Submitting email: darija@strah.si
Submitting phone: 00386 41 330 501

Presenting name and surname: Darija Strah
Presenting email: darija@strah.si
Presenting phone: 00386 41 330 501