



## Service Priorities and Programmes Electronic Presentations

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### **Changing pattern of prenatal testing for Down syndrome: women's choice**

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#### **Introduction**

Universal prenatal screening for Down syndrome (DS) was started in July 2010. After the introduction of non-invasive prenatal testing (NIPT) (cell –free DNA) in private in August 2011, some women underwent NIPT in private.

#### **Objectives**

The aims of this abstract are to report (a) the women's uptake of invasive prenatal diagnosis (IPD) and (b) prenatal detection of Down syndrome (DS) in a four-year period.

#### **Methodology**

A retrospective study was conducted among pregnant women who were screened positive (with a risk  $\geq 1$  in 250) after a conventional DS screening in our department 1 year before (pre- NIPT) and 1, 2 and 3 years after the introduction of NIPT. We compared differences in their (a) uptake rates of IPD or no further testing, and (b) prenatal detection rate of DS using descriptive analysis and a Chi-square test. Conventional screening and IPD were funded publicly, while NIPT was not. We provided counseling and mid-trimester anomaly scan for those women who underwent NIPT. Outcomes of all pregnancies were traced.

#### **Result**

Results:

Around 88% of the DS screening was performed in the first trimester. Of 4,288, 5,726, 5,618, and 6,544 women screened in pre-NIPT and in years 1, 2 and 3 after the introduction of NIPT, 306 (7.0%), 362 (6.3%), 401 (7.1%), 458 (7.0%) were screen positive respectively. In year 1, year 2 and year 3, 49 (13.5%), 107 (26.7%) and 169 (36.9%) of them underwent NIPT while IPD (including both chorionic villus sampling and amniocentesis) was decreased by 16.1%, 25.6% and 32.6%, respectively ( $p < 0.001$ ). The rate of no further testing was similar in year 1, but decreased in year 3 ( $p < 0.001$ ).

There was no increase in IPD rate for fetal structural abnormalities before and after NIPT ( $p=0.7771$ ).

The prenatal detection rate of DS remained similar in pre-NIPT and in years 1, 2 and 3 (100%, 86.4%, 95.7%, 94.4% respectively;  $p=0.4219$ ). Six (2%) of 325 women who underwent NIPT had a high risk result for DS, all of which were confirmed on subsequent IPD. There were no false negative cases.

Conclusions:

As women chose NIPT to IPD after a positive conventional DS screening test, there was a significant decrease in IPD rate for 3 consecutive years without reducing the prenatal detection rate of DS or increasing the IPD for scan abnormalities. More counseling and anomaly scan are required to support those women choosing NIPT.