

Changes to prenatal screening for Down syndrome suggested

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Western Australian researchers say the inclusion of non-invasive prenatal testing (NIPT) into the current Down syndrome screening program would increase detection rates, reduce invasive diagnostic testing and as a result fewer unaffected foetuses would be lost.

Down syndrome is the most common and recognisable chromosomal abnormality, and its prevalence has increased over the years due to more women giving birth over the age of 35 years.

Curtin University Centre for Population Health Research's Peter O'Leary says current first trimester screening (FTS) is performed between 10-13 weeks gestation, with several measurements taken to identify the risk of Down syndrome.

"That involves a sample of blood from the mother, measuring two hormones in mum's blood and also an ultra sound measurement of the foetus looking at the amount of fluid that's collected at the back of the baby's neck, and that's called the nuchal translucency measurement," Professor O'Leary says.

"There is a risk algorithm which combines mother's age, the maternal hormone concentrations and the nuchal thickness into a risk of the baby having Down syndrome."

On the other hand the NIPT test involves the detection and measurement of cell-free foetal DNA in the maternal blood to detect Down syndrome.

Prof O'Leary says expectant mothers who have a high risk pregnancy would benefit from NIPT because it would provide a more accurate risk assessment, and would help to reduce the number of women who go on to have invasive diagnostic testing like FTS.

The researchers reviewed all the data on singleton pregnancy screenings in Western Australia from 2005-2006.

A total of 32,478 women were screened during this period, ranging from 16-44 years-old.

They compared the number of invasive procedures (and foetal losses) and the total cost of the current FTS program with an alternative model that includes NIPT as a second tier test, prior to invasive [diagnostic testing](#).

Inclusion of NIPT was found to reduce the amount of invasive diagnostic procedures and foetal losses by 88 per cent but cost more to run over the two years.

FTS is the most common screening test used to identify Down syndrome during pregnancy with a detection rate of 85 per cent and a screen positive rate of five per cent.

Most pregnancies have a risk of Down syndrome however that risk increases with maternal age.

"As age increases to 30 years the risk is about one in 400. By the time a woman is 40 years the risk is about one in 20," Prof O'Leary says.

Provided by Science Network WA

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