Author's response to reviews

Title: Implementing non-invasive prenatal testing into publicly funded antenatal screening services for Down syndrome and other conditions

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Author’s response to reviews:

Dear Ms Boodhun

We thank the reviewers for their positive comments, and we addressed their points as below. In the manuscript we have made our changes in and highlighted them in yellow for ease of reviewing. We would also like to request amending our title so that it is more aligned with New Zealand screening service terminology.

With thanks
Sara Filoche

Reviewer reports:

Jim Bonham (Reviewer 1):

The Authors describe arguments for the considered and culturally sensitive introduction of publically funded NIPT in Aotearoa New Zealand to detect Downs syndrome. The paper is interesting and topical as it describes some of the contrast between a medical view of screening, largely based from a Western philosophical mind-set versus the culturally distinct views of Maori and Pacific women. The issues that this paper raises extend beyond the particulars of Maori culture and emphasise the disconnect often witnessed between the health care professionals who organise screening programmes and the public who are the recipients of
screening. Understanding and expectations differ between these groups and this is often heightened significantly within particular cultural groups. The introduction of genetic testing into this mix adds to the potential for confusion. As screening programmes spread around the world into culturally and economically diverse groups models exploring and proposing culturally informed decision making with user input are particularly valuable.

I think that this paper should be published and the authors encouraged to consider how the model that they propose may inform the introduction of screening, both antenatal and newborn, into populations and groups with differing world views and cultural heritage.

We thank this reviewer for their comments and are encouraged that our model is of international significance. We have added a paragraph as:

“While our paper focusses on the implementation of publicly funded NIPT in New Zealand, we propose that the principles and recommendations for engaging groups that traditionally have been disenfranchised, or where barriers in access to healthcare services exist, would be applicable internationally. Our model may also have relevance around the introduction of other genetic and genomic screening initiatives including expanded newborn screening.”

John Williams (Reviewer 2):

The authors present a discussion of the issues surrounding the implementation of noninvasive prenatal testing (NIPT) as a publicly funded service in Aotearoa New Zealand. Specifically, they address the precautions that should be taken at various levels including: healthcare system, healthcare organizations and personnel to ensure that access to services is equitable and responsive to the needs and concerns of diverse cultural groups, and to safeguard women's rights to make informed decisions. Although the discussion specifically relates to implementation of publicly funded NIPT in New Zealand, the principles and recommendations for engaging groups that traditionally have been disenfranchised or have had limited access to healthcare services is applicable to other countries with ethnically diverse populations.

We thank this reviewer for their comments and are encouraged that our model is of international significance. We have added a paragraph as above.

Some comments for the authors are listed below.

On page 1, line 62, the authors state: "...and the mother will likely undergo unnecessary invasive diagnostic testing, which in itself carries around a 1 in 100 chance of a miscarriage." To support this statement, they cite guidelines for antenatal screening published by the Ministry of Health in 2012. However, these guidelines do not cite a reference for this statistic. Statements such as these are inaccurate and misleading because they do not take into consideration the natural baseline risk for miscarriage that is gestational age dependent and that exists for all pregnancies. Recent studies that control for background risk of miscarriage show that the "procedure-related" risks for pregnancy loss with amniocentesis and CVS are 0.1% and 0.2% respectively.

We have included this reference, added a new reference and reworded the sentence as “for which there are is an associated risk of miscarriage – reported as 0.1% [7] to 1% [8].”