LETTER TO THE EDITOR

Rapid aneuploidy screening

To the Editor—We read with interest the study of Lim et al[1] reporting on the diagnoses missed by rapid aneuploidy screening (RAS) compared to karyotyping.

While rejecting the use of a stand-alone RAS as an alternative to karyotyping in the present strategy of two-step prenatal screening, they do see a place for RAS if non-invasive prenatal diagnosis (NIPD) were to become available. If they accept more limited testing in the latter context, why not also in the former? Their argument is that in view of the risk of miscarriage attached to current invasive procedures, it would be "prudent to…reveal the maximum information possible". But who is to determine what the most prudent choice of testing would be, and in the light of what considerations?

Rapid aneuploidy screening is favoured by many because it is cheap, fast, and targeted: RAS fits in better with the preceding risk assessment aimed at chromosomes 21, 13 and 18 only and does not generate findings of unclear clinical significance. Therefore anxiety and difficult decision-making in the post-test situation can be reduced. Professionals and women evaluate the findings missed by RAS differently.[2]

Because of these different views, we think that the choice between RAS and karyotyping should be left to the women themselves. This approach would best serve the generally accepted aim of prenatal screening, namely to offer opportunities for autonomous reproductive choice.[3] Of course, this should then apply equally to the NIPD scenario, should broader tests become available in that context.

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3. Health Council of the Netherlands. Prenatal Screening: Down's syndrome, neural tube defects, routine-ultrasonography. Publication no. 2001/11. The Hague, Health Council of the Netherlands; 2001.

Authors' reply

To the Editor—We thank Ms de Jong, Drs Dondorp and de Wert for their letter and would like to respond to their query.

It has always been our position that RAS should not be used as a stand-alone test. Rapid aneuploidy screening in the context of non-invasive prenatal diagnosis is at best a concession. However, we do agree with the authors that the choice between RAS and karyotyping should be left to the women themselves. This has always been the case in our practice.

Nevertheless, the medical fraternity has a deep moral responsibility to ensure that these women are provided a complete clinical picture so that they can make an informed choice. They must be made aware that RAS as a stand-alone test while not generating "findings of unclear clinical significance" will miss cases of mental and/or physically disabling chromosomal disorders.

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