

# ARE COUPLES MAKING INFORMED CHOICES WHEN OPTING FOR PRECONCEPTION REPRODUCTIVE GENETIC CARRIER SCREENING?

## Background

Reproductive genetic carrier screening (RGCS) allows to identify couples who have an increased risk of conceiving a child with a genetic condition. Professional organizations have emphasized that the success of RGCS should not solely be measured by the uptake of screening. An assessment of whether or not individuals are making informed choices with regard to RGCS is considered to be as important [1].

## Aim(s)

The aim of this study was to assess informed choice among couples who were offered RGCS for a large test panel including more than 1000 genes associated with autosomal recessive and X-linked conditions.

## Methods

Women visiting a gynaecologist practice were asked to consider participation in a research study where preconception RGCS was offered for free to them and their male partner. Participants were asked to complete an individual self-administered questionnaire at the end of a pre-test counseling session. A modified Multidimensional Measure of Informed Choice was used to determine whether couples who opted for RGCS made an informed choice [2, 3].

## Results

In total, 82% of participants (n=77) made an informed choice with regard to RGCS according to the modified MMIC. Thirteen participants (17%) made an uninformed choice due to insufficient knowledge and one participant (1%) due to insufficient knowledge and value-inconsistency [4].

## Discussion

In practice, limited resources may restrict the availability of face-to-face pre-test counselling or a follow-up visit which could impact knowledge and therefore informed choice [2].

## Implications and future perspectives

Our study results show high rates of informed choice among couples who were offered RGCS in a highly controlled research context where participants received up to 30 minutes of pre-test counseling. Future research should assess if high levels of informed choice could also be achieved outside a controlled research context.

## References

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2. Lewis, C., et al., Development and validation of a measure of informed choice for women undergoing non-invasive prenatal testing for aneuploidy. *European Journal of Human Genetics*, 2016. 24(6): p. 809-816.
3. van den Berg, M., et al., Informed decision making in the context of prenatal screening. *Patient Educ Couns*, 2006. 63(1-2): p. 110-7.
4. Van Steijvoort, E., et al., Exploring informed choice in preconception reproductive genetic carrier screening by using a modified Multidimensional Measure of Informed Choice. *Patient Educ Couns*, 2022. 105(11): p. 3313-3318.