

Couple-based expanded preconception carrier screening (ECS) offered by the general practitioner (GP): What is the uptake and

who participates?

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1. Introduction and aims

The University Medical Center Groningen (UMCG) developed a couple-based preconception expanded carrier (ECS) test for 50 severe autosomal recessive conditions.

This test was offered to couples from the general population by their (GP) as part of our **implementation study** to investigate: 1) uptake, 2) feasibility, 3) informed choice and 4) psychological impact.

Here we describe uptake, differences between test-uptakers and test-decliners and level of knowledge before and after pre-test counselling.

See poster P19.49A for the results of the feasibility part of this study

2. Methods

Participating GPs invited 4295 female patients aged 18-40 and their partners to participate in an **implementation study**. All couples were offered a (free) ECS test after GP pre-test counselling. Inclusion criteria: 1) having a male partner 2) planning to have (more) children 3) not being pregnant. For an overview of the implementation study see figure 1.

- Uptake is defined as the no. of women (and their partner) attending pre-test counselling and the no. of women (and their partner) taking the UMCG ECS test.

- Sufficient knowledge is defined as $\geq 3/5$ items answered correctly.

Data analysis: Knowledge, attitude and socio-demographic characteristics between female test-acceptors and test-decliners were compared using Fisher's exact tests for ordinal and T-tests for continuous variables.

Figure 1: Flow diagram implementation study

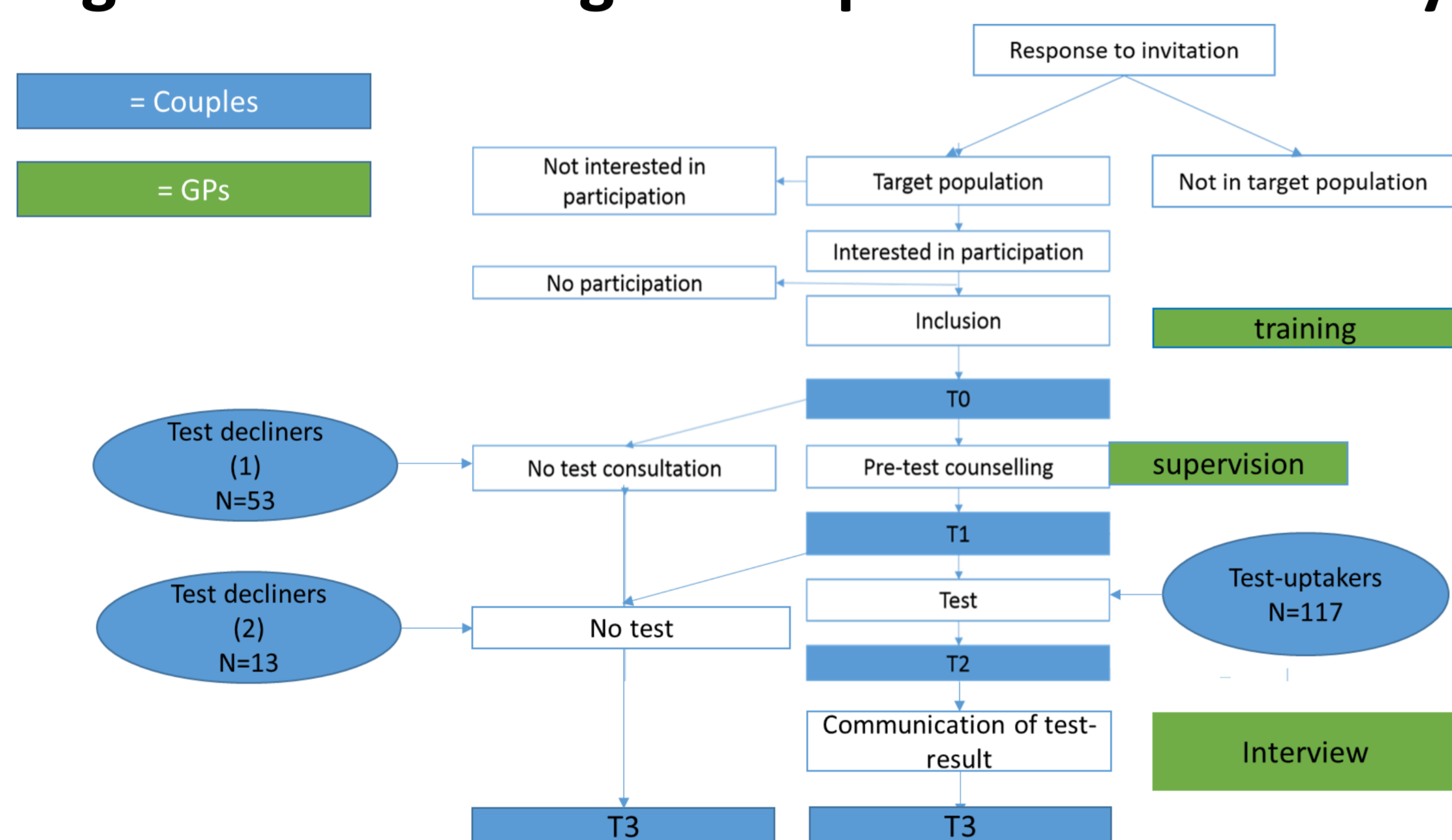
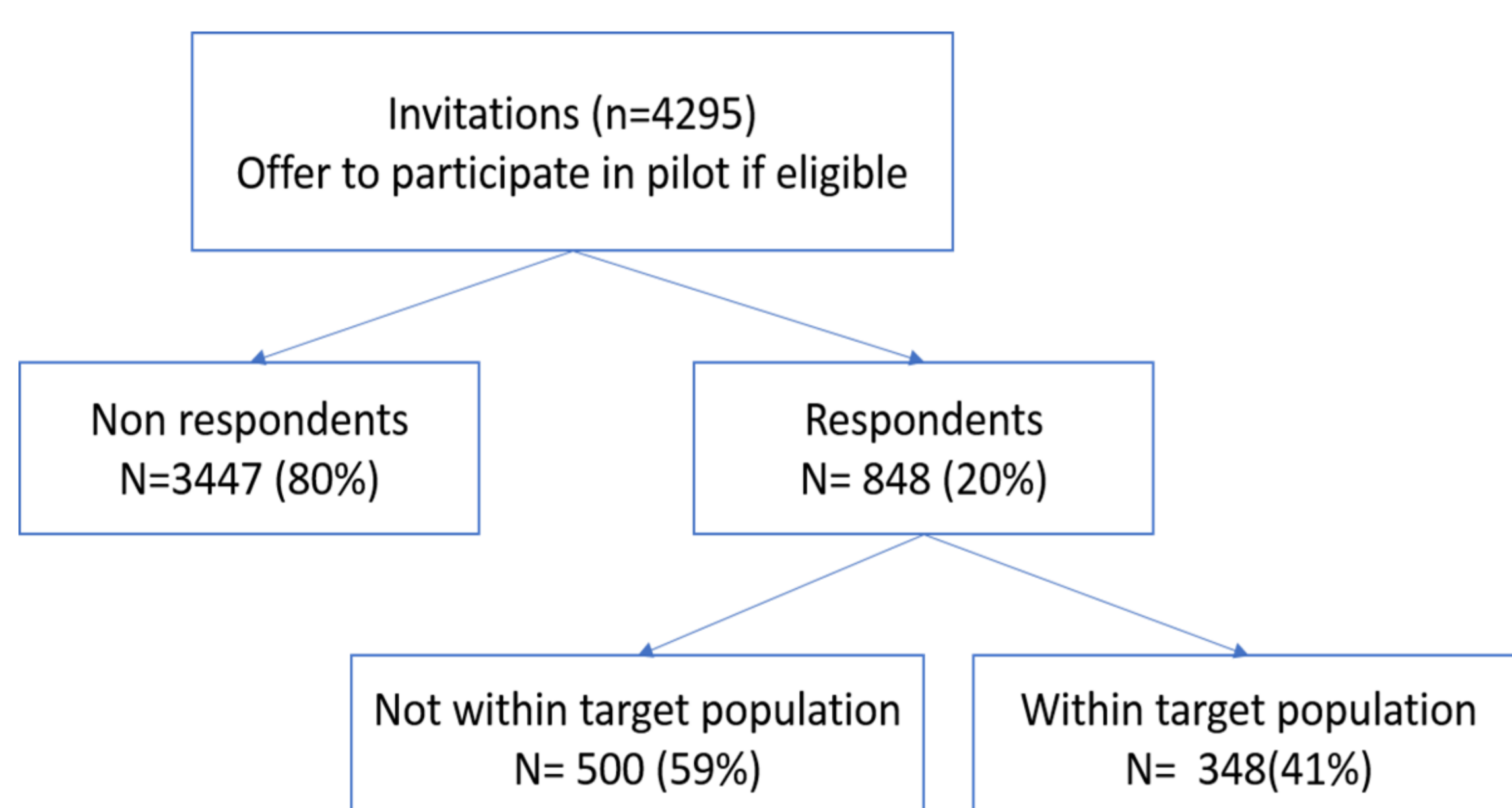


Figure 2: Flow diagram of response to invitation



3.1 Results

Uptake:

- 847/4295 (20%) women responded to the invitation and 348/847 (41%) fulfilled the inclusion criteria (see figure 2).
- 183/348 (53%) participated in the implementation study (survey study + possibility of having (free) ECS test).
- Reasons for non-participation included personal medical circumstances, religious beliefs, 'not the right timing' and over-medicalization of pregnancy.
- 130 women and their partners attended pre-test counselling and 117 women and their partners had couple-testing (see flow diagram).

The UMCG ECS Test

- Criteria for conditions in panel
 - Early onset
 - No treatment available
 - Very severe conditions (either severe physical abnormalities or severe developmental delay) OR premature death OR severe pain
- 1:150 risk for positive **carrier couple** result
Example: Epidermolysis Bullosa

3.2 Results

- Test-acceptors differed significantly from test-decliners in attitude towards ECS (more positive), timing of pregnancy (less soon), (more) perceived benefits and (more) perceived behavioural control (see table 1).
- No significant differences were found in (sufficient) knowledge, age, religion or educational level. (see table 1&2).
- 88% of test-acceptors, 81% (1) and 77% (2) of test-decliners showed sufficient knowledge ($\geq 3/5$ correct answers) before pre-test counselling (see table 2).
- 99% of test-acceptors and 100% of test-decliners showed sufficient knowledge after pre-test counselling (see table 2). This did not differ significantly between test-acceptors and test-decliners.

Table 1: Differences between female test-uptakers and test-decliners

| Total cohort N= 183 | Test-uptakers (n=117) n(%) | Test-decliners (1) (n= 53) n(%) p value | Test-decliners (2) (n=13) n(%) p value |
|---|----------------------------------|--|---|
| Age mean (SD) | 27.9 (5.1) | 27.5 (4.8) 0.621 | 28.0 (5.0) 0.968 |
| Religious beliefs (yes) | 37 (27.4) | 10 (19) 0.256 | 3(24) 0.517 |
| Educational status | | | |
| - Low | 5 (4.3) | 5 (9) | 0 (0) |
| - Intermediate | 48 (41.0) | 27 (51) | 9 (69) |
| - High | 64(54.7) | 21 (40) 0.127 | 4 (31) 0.138 |
| Children (yes) | 14(12.0) | 13 (25) 0.044* | 1 (7) 0.540 |
| Planned timing of pregnancy | | | |
| - As soon as possible (within 6 months) | 15 (12.8) | 12 (23) | 6 (46) |
| - 6 months -2 years | 33 (28.2) | 22 (42) | 1(8) |
| - 2-5 years | 50 (42.7) | 11 (21) | 2 (15) |
| - > 5 years | 10 (8.5) | 5 (4) | 4 (31) |
| - Not sure | 9 (7.7) | 3 (6) 0.053 | 0 0.001* |
| Perceived behavioural control | | | |
| - Very difficult | 1 (1) | 1 (2) | 0 (0) |
| - Difficult | 7 (6) | 11 (21) | 0 (0) |
| - Not difficult/not easy | 29 (25) | 6 (11) | 5 (38) |
| - Easy | 58(50) | 24(45) | 2 (15) |
| - Very easy | 22 (19) | 10 (19) 0.024* | 6 (46) 0.068 |
| Attitude towards termination of pregnancy Mean (SD) | 18.4 (6.0) | 17.1 (5.7) 0.204 | 17.7 (7.5) 0.686 |
| Attitude towards participation in the UMCG ECS-test mean(SD) | 24.2 (3.0) | 22.3 (4.5) 0.008* | 22.4 (4.6) 0.195 |
| Perceived benefits | 18.5 (2.7) | 17.0 (2.8) 0.001* | 18.9 (3.8) 0.632 |

Table 2: Level of knowledge before and after pre-test counselling

| Total cohort N= 183 | Test-uptakers (n=117) n(%) | Test-decliners (1) (n= 53) n(%) p value | Test-decliners (2) (n=13) n(%) p value |
|--|----------------------------------|--|---|
| Sufficient knowledge before pre-test counselling | 103 (88) | 43(81) 0.343 | 10 (77) 0.377 |
| Sufficient knowledge after pre-test counselling | 115 (99) | | 6 (100) 1.0 |

Response rate T1: test-uptakers= 99%; test-decliners (2)=46%

4. Conclusions

- Our results show that couples from the general population are interested in couple-based ECS.
- Most couples who attended pre-test counselling proceeded with couple testing.
- Most female participants had sufficient knowledge which increased after pre-test counselling.
- Timing of a planned pregnancy might explain why some couples declined testing after pre-test counselling.