

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

who participates?

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The authors do not declare conflicts of interest



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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

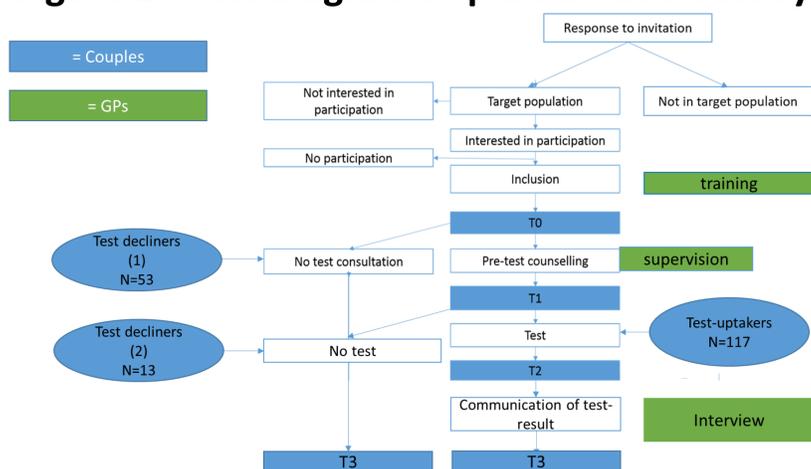
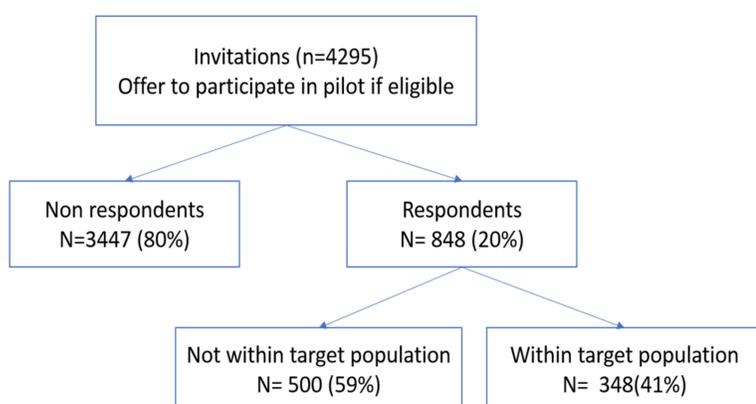


Figure 2: Flow diagram of response to invitation



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.