Feasibility of Couple-Based Expanded Carrier Screening offered by the General Practitioner

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Background

- Next generation sequencing enables efficient screening of all couples planning a pregnancy for carrier status of multiple severe autosomal recessive conditions simultaneously.
- Barriers such as 1) available time, 2) adequately trained professionals within current health care systems and 3) offer by private companies raise ethical and practical concerns. This requires new strategies to implement ECS in a responsible and feasible way in regular health care.
- We have previously demonstrated positive attitudes towards such a test and that general practitioners were thought to be the most suitable provider.
- The UMCG laboratory has developed an ECS couple test including 50 severe autosomal recessive conditions.

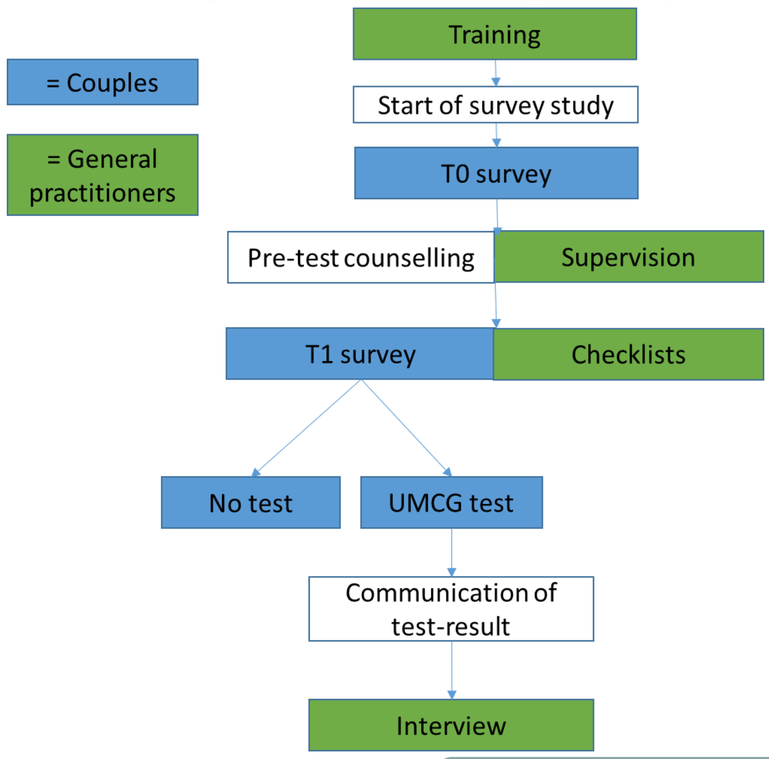
Methods

GPs invited 4295 women aged 18-40 and their partners to the **implementation study.** All couples were offered a (free) ECS test after GP pre-test counselling. For couples with a positive result a referral to clinical genetics was made. Inclusion criteria: 1) having a male partner 2) planning to have (more) children 3) not being pregnant.

- The test-offer was considered feasible if 1): ≤ 20% of GPs needed additional supervision; 2) ≤ 20% of normal risk couples were referred for additional counselling to clinical genetics 3) ≥ 80% of consultations within time-span of double consultation.
- We used a mixed methods approach. 1: Semi-structured interviews with GPs, and checklists (response rate 83%); 2: Couples: Longitudinal survey, response rates 99% (T0) and 92% (T1)).
- Qualitative interviews were analysed using framework analysis.

See poster P01.034B for the results about uptake of the test-offer

Flow diagram: Feasibility study



Overview participating GPs and number of pre-test counselling sessions and couple-tests

Participating practices	No conducted counselling/GPs attended training	Type of practice	No of GPs participated in interview	No of pre-test counselling sessions	Couple-tests results communicated
No 1	1/1	City	1	24	23
No 2	1/1	City	1	12	12
No 3	1/2	Rural	1	4	3
No 4	4/6	Urban countryside	2	23	20
No 5	2/3	Urban countryside	2	27	25
No 6	1/1	Urban countryside	1	18	14
No 7	1/1	Rural	1	5	5
No 8	1/2	Urban countryside	1	5	4
No 9	1/2	City	0	12	11
Total	13/19		10	130	117

GP Training

1: Before the study commenced, GPs followed a training designed by the research team and given by professionals the UMCG Clinical Genetics department.
All GPs were asked to fill out an online evaluation and knowledge test and provided with further training if knowledge was lacking.
2: Each GP was supervised by a genetic counsellor twice during pre-test counselling
3: Several supportive material, such as an information booklet and informative website were provided.

Feasibility measures

Feasibility topics	Quantitative	Qualitative
	Instrument and time-point (measures)	*Interview topics*
1.Training	Training and supervision at T0	Interview (GP) Training, supervision and additional preparation
2. ECS-test offer: counselling aspects	Checklist at T1(GP) Survey at T1 (couples) *Patient satisfaction* *7-item counselling satisfaction scale* (α 0.920)	Interview (GP) Barriers and facilitators
3. ECS-test -offer care: organizational aspects	*Checklist at T1(GP)* *Duration of consultation* *(aim within 20 minutes= double consultation)* *No of normal risk referrals*	Interview (GP) Barriers and facilitators
4.Other factors influencing the provision of the test by the GP	Survey at T1 (couples) *Barriers to participate*	Interview (GP) Barriers and facilitators
5.Views about implementation		Interview (GP)

Results

Training

"I particularly liked the training course, which was essential. It would be difficult to provide the PCS test without doing the training course first." GP practice 8

All GPs felt capable of providing pre-test counselling after training, supervision and with supportive materials. 0/13 GPs needed additional supervision.

Counselling Aspects

- Patient satisfaction: ≥ 90% scored ≥ 4/5 (1-5 scale).
- Genetic counselling satisfaction scale mean 4.7 (SD 0.5) (1-5 scale).

Organisational Aspects

- 58% of consultations ≤ 20 minutes; median (IQR) 20.
- GPs did not see an indication to refer normal risk couples to clinical genetics for additional pre-test counselling.
- GPs reported that, from their experience, negative test-results were satisfactorily communicated in various ways such as by telephone or email.

"I noticed that I got more and more relaxed, that it was easy for me ... (...)The consultations were really good, very relaxed ... Yes, it worked, it worked well." GP practice no 1

Barriers and Facilitators

- Only 7% of couples partly and 4% completely agreed that participating in this ECS-test takes much time or effort.
- About 20% of couples did not like having to book a GP appointment.
- Interviews demonstrated that GPs thought a higher prior knowledge level influenced the consultation positively, because couples were already well informed.

Views on Future Implementation

- These experienced GPs also considered themselves as most suitable to provide ECS to couples from the general population.
- Education of test-providers was considered essential.
- GPs considered pre-test counselling with both partners present important.

"Yes, the GP is of course suitable because he or she knows the people best and because there's a low threshold to seeing him, and also because he might know the families." GP practice no 5

Conclusion and recommendations

Our research demonstrates that if GPs are trained to provide couple based- ECS this is acceptable and also feasible in most cases. We are now looking into ways of reducing time-costs for GPs, through, for example, provision of information prior to couples' attendance.