Implementation study on population based carrier screening via the general practitioner in the northern part of the Netherlands. Research on the implementation of a clinically validated population based carrier screening test for couples with a child wish.

Published: 02-11-2015
Last updated: 19-04-2024

This study aims to investigate whether the clinically validated population based expanded preconception carrier screening (PCS) test, developed by the Department of Genetics of the UMCG, can be implemented responsibly via the general practitioner as...

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational non invasive

Summary

ID

NL-OMON42389

Source
ToetsingOnline

Brief title
PCS implementation study

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
population based preconceptional carrier screening, preconceptionscreening

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: genetics, population based carrier screening, preconception care, preconception screening

Outcome measures

Primary outcome

1. psychological impact and informed choice

2. uptake

3. practical feasibility (GP)

Ad 1.

Psychological impact and informed choice

Receiving information about the PCS test may, raise a couple*s awareness of the

risk of having a child with a severe disorder. This awareness, in addition to

having to make a decision whether to take the test or not may evoke adverse

psychological reactions with both an emotional (anxiety, distress) and/or

cognitive (rumination, decisional conflict) nature.

Both couples who take the test as well as those who decide not to, immediately,

or after consultation of the GP, may experience adverse psychological

reactions.

The broader literature on receiving genetic risk information and genetic counselling and testing shows that in general the psychological impact is moderate or at best short lived (Haga et al, 2014). To date, however, no studies have been conducted examining the psychological reactions of prospective parents without a positive family history to expanded PCS.

In general, implementation a test, such as the PCS test, is acceptable from a psychological point of view, if psychological effects on population level are limited and if the level of knowledge of the population that gets the information is such that the decision on whether or not to take the test is based on an informed one. This implementation study is meant to investigate the impact on those couples who are offered the test, might take the test, but who are not a carrier couple. This concerns the large majority of our study population, since only 1-3 couples are expected to be a carrier couple. The impact on this group is relevant to measure, since the positive consequences of the PCS offer and taking the test, (increasing reproductive autonomy), for a small group (1 in 150 couples who take the test) need to be balanced against the burden of offering the test to the whole population that gets the offer.

Informed Choice:

Similar as in studies on prenatal screening, in a study which investigates the implementation of a preconception screening test, informed choice is an important objective (Marteau et al., 2001). The aim of this offer of PCS screening is not to reach the highest uptake as possible, as is the case in for

example newborn screening. On the contrary, its aim is to increase reproductive autonomy, by letting the couples decide for themselves whether or not they want to participate in the test. This choice to take the test or not should optimally be an informed choice, meaning that both partners have the correct knowledge on the essential aspects of the test and have an attitude towards screening which corresponds to their choice whether or not to participate

Methods:

When examining the psychological impact of PCS, the study aims are:

a) to examine short term psychological reactions to receiving information about the offer of PCS testing (T0)

b) to examine short term psychological reactions to having received counselling from the GP (T1)

c) to examine short term psychological reactions to having taken the test (T2)

d) to examine long term psychological reactions six months after having received information/having had GP counselling/having taken the test

Measures:

Positive and Negative Affect Schedule (PANAS; Watson et al, 1988)

Short version of the State-Trait Anxiety Inventory Scale (STAI-6; Marteau et al, 1992)

Decisional Conflict Scale (Koedoot et al., 2001)

Knowledge and informed choice will be measured with specifically designed self-constructed items for the PCS test, based on the literature. This will be

measured after the GP consultation, irrespective of whether or not the couple decides to take the test.

All patients who are asked to participate in the research will also be offered a free PCS test. They are asked to fill in a maximum of four questionnaires, taking about 15-20 minutes each.

T0: after the offer of the PCS test (all)

T1: after the GP consultation (only those couples who visited their GP)

T2: after having done the PCS (only those couples who took the test)

T3: 6 months after having filled in the last questionnaire (all respondents at T0)

Ad 2.

To measure the uptake, the number of participants per step in the protocol will be recorded and related to the total number of women and their partners who were invited to participate.

1) What percentage of invited women/couples reads the information leaflet and what percentage visits the website?

2) What percentage of couples attends a GP consultation?

3) What percentage of invited women/couples takes the test?

Ad 3: Practical feasibility

To study the practical feasibility, the PCS offer by the GP within the primary health care system will be evaluated. In order to investigate this, GPs will be asked to fill in a survey and they will also be asked to participate in an in depth interview on the following aspects:

- Time spent on the selection of patients, inviting the patients, counseling (pre-test counseling and communication of the test result)
- What are the GP*s experiences concerning their own pre-test counseling skills (after having participated in the training and if needed after additional training during the study)
- What are their opinions on taking up the offer of PCS in their regular practice within the standard care of the preconception consultation. What are, according to them, the barriers and benefits of this offer of PCS via the GP, and what can be done to overcome these barriers, and what could be solutions to problems that may arise.

Furthermore, the proportion of GP*s who need additional supervision from the genetic counselor during the study will be recorded as well as the proportion of couples who request additional counseling by the genetic counselor after having had the GP consultation

Secondary outcome

not applicable

Study description

Background summary

The department of Genetics of the UMCG has developed a PCS test which has not yet been implemented in either the Dutch or European regular health care system as offer to the general population. The PCS tests that are currently offered, usually test for one or a few conditions, such as Tay Sachs and Cystic Fibrosis. Due to the introduction of next generation sequencing (NGS) it is, however, now possible to screen for many genes or conditions simultaneously while the costs of this type of screening are declining rapidly.
A PCS test for the whole population that screens for several conditions simultaneously is currently not routinely offered in Dutch health care, nor in surrounding countries. The test provides information about carrier status of 70 autosomal recessive conditions. The PCS test only includes those diseases which are characterized as very severe, early onset and untreatable. The test will be provided to the couples with a child wish from the general population by their general practitioner.

The UMCG*s PCS test will detect an increased risk (both partners of a couple being a carrier for the same disease) in approximately 1 per 150 couples. These couples have a 1 in 4 risk with each pregnancy that their child will have one of the severe autosomal recessive conditions being screened for. These carrier couples can, if they so wish, avoid the birth of an affected child for example by making use of IVF with embryo selection (pre-implantation genetic diagnosis, PGD) or prenatal diagnostics. Other options include making use of a gamete donor or to refrain from having children with this partner. Given the fact that the offer of the, technically validated, PCS test to the general population is new, little is known about the uptake, feasibility and psychological impact of the test for potential participants and providers, in this case the GPs.
GPs have been selected to be the preferential provider of this test by both prospective participants as well as prospective providers of the test due to various reasons. The implementation study aims to investigate all three abovementioned aspects, uptake, psychological impact and practical feasibility. Given the interest for these kinds of screening tests both from the society at large as well as from politics, and therefore the high probability that these will be implemented in the national healthcare system, it is necessary to get experience on the implementation of these test in a small setting first, before recommendations can be given for nation-wide implementation. The research protocol of this implementation study is based on prior research and recommendations from investigations by our research group on the ethical, psychological and practical aspects, as well as on studies from the literature on similar, but slightly different PCS tests.

Study objective

This study aims to investigate whether the clinically validated population based expanded preconception carrier screening (PCS) test, developed by the Department of Genetics of the UMCG, can be implemented responsibly via the general practitioner as part of preconception care for couples wishing to have a child. Responsible implementation of this test will be evaluated by studying:

a) psychological impact
b) uptake
c) practical feasibility.

Study design

It is an observational study. Participants will be asked to fill in a minimum of two to a maximum of four questionnaires at various defined time points during the study period. These questionnaires consist of validated as well as self-constructed items. Participants are also offered to have the PCS test (for free).

The implementation study will take place within the GP practices of 10-15 GPs in the northern part of the Netherlands, who have been trained for this before the start of the study.
The test will be offered as part of preconception care, which is already part of the GP standard care (NHG standard preconceptiezorg) (het *preconceptieconsult*). Couples who wish to have a child together are given advice aimed at creating a healthy pregnancy.

All female patients between the age of 18-40 of the participating GP practices will receive an offer to have the PCS test (for free) with their partner. The couples will receive an information leaflet as well as a link to the website www.dragerschapstest.umcg.nl where they can find additional information and can ask questions concerning participation in the research.
If they are interested in taking the test, they will first need to make an appointment for a consultation with the GP to discuss the offer. In case the GPs need assistance or require supervision for these pre-test counseling sessions, a genetic counselor will be available from the department of Clinical Genetics. After the GP consultation, a couple can have their blood drawn for the PCS test. Results will be sent to the GP within 8 weeks and are disclosed to the couple by the GP, after which the couple can be directed to the department of Clinical Genetics if indicated (if both partners turn out to be a carrier for the same disease or if they need additional post-test counseling).

Study burden and risks

The burden of participation is minimal for the majority of the study population (2 questionnaires with an average duration of 15-20 minutes). The burden is

slightly higher for those respondents (approximately 10-25% of study population) who visit their GP to talk about the decision whether or not to take the PCS test (3 questionnaires, 1 consult with GP). For the respondents who decide to take the PCS test (approximately 5-20% of study population), the burden is highest (4 questionnaires, 1 consult with GP, 1 blood test).The physical risks attributed to the blood test are negligible. Based on the literature, the psychological impact is expected to be moderate and short lived, however, since this will be the first time such a population based expanded carrier screening test is offered, measuring the psychological impact will be part of the investigation.

Contacts

Public
Universitair Medisch Centrum Groningen

Antonius Deusinglaan 1
groningen 9713 AV
NL
Scientific
Universitair Medisch Centrum Groningen

Antonius Deusinglaan 1
groningen 9713 AV
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

1 be a female patient from one of the participating GPs
2 aged 18-40
3 have a partner
4 have a wish to have children with that partner
5 and: male partners of these females

Exclusion criteria

women who are already pregnant

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Health services research

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	14-01-2016
Enrollment:	2000
Type:	Actual

Ethics review

Approved WMO

Date:	02-11-2015
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL54060.042.15