Tailored approach to inform family members at risk of inherited cardiac diseases: a RCT

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Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Interventie onderzoek

Samenvatting

ID

NL-OMON27057

Bron
Nationaal Trial Register

Aandoening

Predictive genetic testing, inherited cardiac diseases, inform family members at risk, tailored

Ondersteuning

Primaire sponsor: Academic Medical Center, University Medical Center Groningen & University Medical Center Utrecht
Overige ondersteuning: Dutch Heart Foundation

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

The difference between the intervention- and the control group, in uptake of genetic counselling and testing by family members at risk of inherited cardiac diseases in the first

year after detection of the disease-causing mutation in the index patient, compared to the total number of at-risk family members.
Conditional uptake, defined as the number of family members that is genetically tested relative to the number of family members attending genetic counselling in the first year after detection of the disease-casing mutation.

Toelichting onderzoek

Achtergrond van het onderzoek

Rationale: Predictive genetic testing for family members at risk of inherited cardiac diseases has become part of genetic clinical practice. In the Netherlands, family members at risk of inheriting the mutation are generally informed by the index patient, supported by a family letter of the genetic counsellor. Previous research shows that with this approach on average only half of family members at risk of inherited cardiac diseases attend genetic counselling. To prevent fatal consequences of these diseases, uptake should be optimal.

Objectives: Therefore, this study aims to investigate uptake of genetic counselling and testing using a tailored approach towards informing family members at risk, in which index patients may decide which family members they will inform themselves at first and which family members they prefer to be informed by the genetic counsellor directly, compared to uptake using the usual approach. Secondary aims are to evaluate acceptance of the used approach and impact on family relationships and psychological functioning.

Methods: This study is a multicenter randomized clinical trial. Adult index patients with an inherited cardiac disease and a pathogenic mutation are eligible to participate. In addition, first-degree (or second-degree in case of a deceased connecting first-degree family member affected or suspected to be affected) family members at risk of inheriting the mutation of enrolled index patients will be invited to participate as well. Uptake of genetic counselling and testing of family members in the first year after detection of the pathogenic mutation in the index patient will be compared to the total number of at-risk family members. Participants will be asked to complete questionnaires on appreciation of the used approach, perceived impact on family relationships and impact on psychological functioning (i.e., worrying and feelings of fear and depression).

Doel van het onderzoek

In the Netherlands, family members at risk of inherited cardiac disease are generally informed by the index patient (i.e., the first in the family diagnosed with an inherited cardiac disease) about the advice of cardiac monitoring and predictive genetic testing, supported by a family letter written by the genetic counsellor. However, previous research shows that the uptake of genetic counselling and testing is relatively low. Ideally, this percentage should be higher to prevent fatal consequences of the disease. This study aims to investigate a tailored approach towards informing at risk compared to usual care (i.e., by the index patient). We

hypothesize that uptake of genetic counselling and testing with a tailored approach will be improved.

Onderzoeksopzet

Index patients:

T1: One month after receiving genetic test result

T2: Nine months after receiving genetic test result

Family members:

T1: Directly after attending genetic counselling/genetic testing

Onderzoeksproduct en/of interventie

In this study, a tailored approach of informing family members at risk of inherited cardiac diseases will be investigated, in which index patients will be informed about the risk for their family members and will be offered the choice which family members will be informed by the index patients themselves at first and which family members they prefer to be directly informed by the genetic counsellor. In both cases, a family letter with information written by the genetic counsellor is provided. After one month, family members informed by the index patients will receive an additional letter of the genetic counsellor with consent of the index patient. In addition, a website-link with disease specific information will be provided for both index patients and family members. In the control group, the standard approach will be used, in which the index patient is asked to inform their family members, supported by a family letter written by the genetic counsellor.

Contactpersonen

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

The study population consists of:

(1) Index patients with an inherited cardiac disease and a putative pathogenic mutation (i.e., class 4 or 5 mutation): Inclusion criteria: (a) Index patients who are the first in their family to attend pre-test genetic counselling about genetic testing for inherited cardiac diseases, (b) Index patients that have at least one alive adult family member at risk of inheriting the mutation, (c) Index patients who are aged 18 years or older, (d) Index patients who are able to read and write Dutch. For final enrolment: Index patients who have a putative pathogenic mutation detected at the DNA test.
(2) Their first-degree, and second-degree family members in case of a deceased connecting first-degree family member that is affected or suspected to be affected (in case of sudden cardiac death), who are supposed to have a 50% risk of inheriting the disease-causing mutation. Inclusion criteria: (a) Family members who are aged 18 years and older, (b) Family members who are able to read and write Dutch.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

Exclusion criteria: (1) Index patients and family members who have cognitive functioning problems and therefore are not able to provide informed consent.

Onderzoeksopzet

Opzet

Type:	Interventie onderzoek
Onderzoeksmodel:	Parallel
Toewijzing:	Gerandomiseerd
Blindering:	Open / niet geblindeerd
Controle:	Actieve controle groep

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	01-10-2017
Aantal proefpersonen:	425
Type:	Verwachte startdatum

Ethische beoordeling

Positief advies

Datum:	24-05-2017
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

Geen registraties gevonden.

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL6341

Register	ID
NTR-old	NTR6657
Ander register	: 2017_145, MEC AMC

Resultaten