New strategies to detect cancers in carriers of mutations in RB1: blood tests based on tumor-educated platelets, or extracellular vesicles.

Gepubliceerd: 10-09-2019 Laatst bijgewerkt: 18-08-2022

Possibly the baseline in Rb survivors and Rb children is different from control patients.

Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON28055

Bron
Nationaal Trial Register

Verkorte titel
NIRBTEST

Aandoening

Retinoblastoma

Ondersteuning

Primaire sponsor: None
Overige ondersteuning: This project is financially suported by TRANSCAN

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

1. Determine the non-cancerous baseline in adult RB1-mutation carriers (Rb-survivors).
2. Contribute to the biobanking of blood and cancerous tissues from RB1-mutation carriers with SPMs.

Toelichting onderzoek

Achtergrond van het onderzoek

Rationale: Individuals with a cancer predisposition due to a mutation in the paradigm tumor suppressor gene RB1, have a high risk to develop the childhood cancer retinoblastoma (Rb). Biopsies are not possible in Rb, before treatment selection. Heritable Rb patients have also a high risk to develop other types of second primary, either childhood or adult, malignancies (SPMs), notably sarcomas and melanomas. Remarkably, SPMs are now the leading cause of death in heritable-Rb-survivors. Unfortunately, there are no well-developed regular surveillance protocols for SPMs in Rb survivors available right now. Recently, new non-invasive cancer test have been developed, based on either RNA-sequencing data from platelets (ThromboSeq), or on extracellular membrane vesicles (EVs) derived from tumor cells present in blood.
Objective:
- Determine the non-cancerous baseline in adult RB1-mutation carriers (heritable-Rb-survivors).
- Contribute to the biobanking of blood and cancerous tissues from RB1-mutation carriers with SPMs.
- The development of blood-based tests, either platelet or EV-based, for the detection of (the type of) tumors in RB1-mutation carriers.
Study design: Cross-sectional multicenter trial.
Study population:
- 40 Rb patients (children),
- 40 controls (children),
- 153 Rb survivors (adults),
- 153 controls (adults),
- 10 Rb survivors with SPM (children/adults).
Main study parameters/endpoints:
- Determine the non-cancerous baseline in adult RB1-mutation carriers (heritable-Rb-survivors).
- Contribute to the biobanking of blood and cancerous tissues from RB1-mutation carriers with SPMs.
Nature and extent of the burden and risks associated with participation, benefit and group relatedness:
Two blood samples totalling 10ml blood will be collected for every participant. Additionally, a short questionnaire has to be filled in concerning their and their family’s cancer history. Blood draws will be done, when participants are already present in the hospital for other appointments, and thus no extra visits are required. For all children, blood will be collected through an already present IV, and so no extra venepuncture is required. Children have to be

included because Rb is a tumor only present in this patient group.

Doel van het onderzoek

Possibly the baseline in Rb survivors and Rb children is different from control patients.

Onderzoeksopzet

1 timepoint and in case of developped SPM a second timepoint

Onderzoeksproduct en/of interventie

None

Contactpersonen

Publiek

Amsterdam UMC
Armida Fabius

+31204440285

Wetenschappelijk

Amsterdam UMC
Armida Fabius

+31204440285

Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

criteria:
- Adult:
o Group 1: germline mutation RB1.
o Group 2 (control): no germline mutation RB1.
- Pediatric:

o Group 1: somatic or germline mutation RB1
o Group 2 (control): no mutation RB1.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

- Adult:
o Group 1: concomitant heritable (inherited) disorder other than caused by monoalleic mutation of RB1.
o Group 2 (control): cancer or already known cancer predisposition syndrome.
- Pediatric:
o Group 1: concomitant heritable (inherited) disorder other than caused by monoalleic mutation of RB1.
o Group 2: cancer or already known cancer predisposition syndrome.

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Anders
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	13-12-2018
Aantal proefpersonen:	396
Type:	Verwachte startdatum

Voornemen beschikbaar stellen Individuele Patiënten Data (IPD)

Wordt de data na het onderzoek gedeeld: Nog niet bepaald

Ethische beoordeling

Positief advies

Datum:	10-09-2019
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	NL8013
Ander register	METC VUMC : METC2018.095

Resultaten