Psychosocial Distress and Quality of Life in CDKN2A Pathogenic Variant Carriers and their first-degree relatives

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To determine 1) the psychological distress and quality of life, and 2) the psychosocial impact of the pathogenic germline CDKN2A variant on lifestyle, genetic testing, skin, and pancreatic surveillance, and family planning in PV carriers and their…

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational non invasive

Summary

ID

NL-OMON53436

Source
ToetsingOnline

Brief title
PSI-CDKN2A

Condition

- Congenital and hereditary disorders NEC
- Anxiety disorders and symptoms
- Family issues

Synonym
CDKN2A P16-Leiden

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum

Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: CDKN2A, Hereditary cancer, Psychological, Quality of Life

Outcome measures

Primary outcome

First, the questionnaire aims to explore the psychological distress and quality of life by validated questionnaires including CWS, HADS, and SF-12. Second, the psychosocial impact on lifestyle, genetic testing, skin and pancreatic surveillance, and family planning is examined by both self-developed and validated questionnaires (PAHC, DCS).

Our primary objectives are:

- To investigate the quality of life and psychological distress in CDKN2A PV carriers and their first-degree relatives;

- To assess the psychosocial impact of the CDKN2A PV on daily life, with regards to the following four themes: 1) lifestyle, 2) genetic testing, 3) skin and pancreatic screening, and 4) family planning.

Secondary outcome

- To investigate whether psychosocial outcomes differ between CDKN2A PV carriers and first-degree relatives;

- To identify individuals who may benefit from additional psychological support.

- To develop a website and decision aid to help PV carriers and first-degree relatives with complex choices associated with the PV, such as engagement in

genetic testing and cancer screening, lifestyle, and family planning.

Study description

Background summary

Individuals with a germline CDKN2A pathogenic variant (PV) have an increased lifetime risk of 70% to develop melanoma of the skin and 20% to develop pancreatic cancer. While skin surveillance is offered to CDKN2A PV carriers and their first-degree relatives, who are at a 50% risk of carrying the mutation, pancreatic screening is reserved for those with a proven pathogenic CDKN2A variant. Screening for cancer may be psychologically burdensome and lead to increased cancer worries. In addition, genetic testing and knowledge about one*s genetic risk profile may have a negative psychosocial impact on daily life. Accordingly, PV carriers and their first-degree relatives may experience impaired quality of life with psychological distress about their own and relatives* cancer risk. Literature on the psychosocial impact of the CDKN2A mutation carriership is scarce.
A recent focus group study conducted by our work group among PV carriers and their first-degree relatives demonstrated different perceptions and attitudes toward four themes; 1) lifestyle, 2) genetic testing, 3) skin and pancreatic surveillance, and 4) family planning. However, a quantitative analysis of quality of life and psychosocial distress in a large cohort of proven and potential PV carriers has never been conducted.

Study objective

To determine 1) the psychological distress and quality of life, and 2) the psychosocial impact of the pathogenic germline CDKN2A variant on lifestyle, genetic testing, skin, and pancreatic surveillance, and family planning in PV carriers and their first-degree relatives. The results will be used to develop a website with an online decision-aid tool for PV carriers and their family members.

Study design

Observational cross-sectional questionnaire-based study.

Study burden and risks

Individuals are asked to complete a questionnaire at home, which will take around 45 minutes. No invasive procedures or clinical tests are required for this study. Consequently, no risks are associated with participation.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

- 18 years or older
- Written informed consent
- Proven CDKN2A PV carrier (confirmed by positive genetic test result)
OR
- CDKN2A risk carrier (50% chance of carrying the PV; genetic status not [yet] confirmed by genetic testing)
- Participation in the LUMC skin and/or pancreatic surveillance program

Exclusion criteria

Individuals that are not able to read or understand Dutch will be excluded from

participation in this study.

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Prevention

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	03-02-2023
Enrollment:	500
Type:	Actual

Ethics review

Approved WMO

Date:	25-01-2023
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

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	NL82652.058.22