Delineation of novel tumor predisposition syndromes is patients with childhood cancer and identification of the underlying molecular defects

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1. Further delineation of four newly identified tumor predisposition syndromes in childhood cancer patients using 3D-analysis of facial morphology. 2. Identification of molecular genetic defects responsible for the tumor predisposition syndromes...

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

Summary

ID

NL-OMON32364

Source
ToetsingOnline

Brief title
Tumor predisposition syndromes in childhood cancer patients

Condition

- Congenital and hereditary disorders NEC
- Miscellaneous and site unspecified neoplasms malignant and unspecified

Synonym
cancer syndromes, tumor predisposition syndromes

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Stichting Kindergeneeskundig Kankeronderzoek

Intervention

Keyword: childhood cancer, dysmorphology, tumor predisposition syndrome

Outcome measures

Primary outcome

1. In order to objectify and specify craniofacial abnormalities of the four new syndromes, three- dimensional (3D) analysis of facial (dys)morphology will be performed using dense surface models.

2. In order to further investigate and identify molecular defects underlying the four new tumor predisposition syndromes, karyotyping, array CGH experiments and/or SNP array will be performed to identify copy number changes and microdeletions. Karyotyping will be performed in order to identify balanced translocations.

Secondary outcome

no

Study description

Background summary

Genetic syndromes can be associated with an increased risk for tumor development. Previous studies in our center showed a significantly increased incidence of morphological abnormalities and presence of (known or suspected) syndromes in a cohort of 1,073 long-term survivors of childhood cancer and pediatric patients with cancer.
Furthermore, statistically significant patterns of co-occurring morphological

abnormalities suggestive of new tumor predisposition syndromes were found. Since many dysmorphic syndromes involve craniofacial abnormality, techniques for objective analysis of facial abnormality such as three-dimensional (3D) imaging of the face are relevant for supporting clinical diagnosis. Karyotyping and molecular genetic techniques such as array based comparative genomic hybridization (aCGH) and single nucleotide polymorphism (SNP) arrays may help to elucidate the underlying molecular defects of the newly identified tumor predisposition syndromes.

Study objective

1. Further delineation of four newly identified tumor predisposition syndromes in childhood cancer patients using 3D-analysis of facial morphology.

2. Identification of molecular genetic defects responsible for the tumor predisposition syndromes using karyotyping and the molecular techniques SNP array and/or array-CGH.

Study design

Observational cohort study

Study burden and risks

The 3D imaging of the face is non-invasive and will be performed using photographic devices; there are no associated risks or clear burden for the patients and their parent(s) at participation. In order to perform karyotyping, aCGH and/or SNP array experiments, peripheral blood (PB) (15 ml) will be drawn. The associated risk and burden will be that associated with a single PB puncture. Further delineation of the new tumor predisposition syndromes and identification of the molecular defects underlying these syndromes can be of high importance in further care and counseling for the patient and his/her family, and for other patients having the same new syndrome.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

-Patients diagnosed with pediatric cancer and identified to show 1 of the 4 patterns of morphological abnormalities indicative of novel tumorpredisposition syndromes (blepharophimosis (BP) pattern, asymmetric lower limbs (ALL)pattern, epicanthal folds (EF) pattern, Sydney creases (SC) pattern)
-Written informed consent by patient when aged 18 years or older, by patient and parents when 12 years or older but younger than 18 years, by parents when younger than 12 years, and written informed consent by parent(s).

Exclusion criteria

zie inclusie criteria

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-08-2008
Enrollment:	300
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Amsterdam UMC

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	NL23474.018.08