



Version 14th of May 2013

**Tumor Predisposition Syndromes in Childhood Cancer Screening Instrument****Part I: Screening form**

Please note that this screening form is part of a screening instrument which has been subject to an initial, limited validation. Prudence is called for preliminary use in clinical practice

**Patient characteristics**

Name:

Date of birth:

Sex:

Self-reported ethnicity:

Date of examination:

Diagnosis:

Year of diagnosis:

Therapy: Chemo:

Surgery:

Radiotherapy:

Relapse: Y/N Year:

Therapy (*please specify*):Perinatal history:

term labour (weeks):

weight at birth (grams):

complications:

Development:

Motor:

Cognition:

Puberty:

Other medical history (hospital admissions, problems requiring specialist medical care):**Family history**

(Please make a note of relatives using French nomenclature: M= mother, P= father, F= brother, S= sister, Fs= son, Fe= daughter. For example for a cousin who is the son of the brother of mother (=son of uncle mother's side): FsFM)

Tumor(s)/Leukemias (please indicate age of onset):Morphological abnormalities:**Physical examination****ANTHROPOMETRICS**

Height (m):

Weight (kg):

Head circumference (cm):

**HEAD****Cranium**

Scalp tumors:

yes/no

Brittle hair\*:

yes/no

**Eyes**

Cataract*	yes/no
Visible nerve fibres on cornea*	yes/no
Photosensitivity*	eyes /skin/no

**Ears**

Crease/pits of ear lobule*	yes/no
(Posterior) helical pits of ear helix*	yes/no

**Mouth/oral region**

Leukoplakia*	yes/no
Tongue*	large/lobulated/protruding/normal
Oral pigmentation*	yes/no
Oral tumor	yes/no
Abnormal oral mucosa (cobblestone)*	yes/no
Mucosal neurinomas*	yes/no
Papilloma peri-orificial*	yes/no

**THORAX**

Supernumerary nipples*	yes/no
------------------------	--------

**ABDOMEN**

Umbilical hernia*	yes/no
-------------------	--------

**EXTREMITIES**

Asymmetry* (length, width or both)	arms/legs/both /no
Palmar pits*	yes/no

**GENITALIA**

Abnormal genital pigmentation*	yes/no
Ambiguous genitalia*	yes/no

**SKIN**

Teleangiectasia*	conjunctival/ nasal/oral/no
Tumors*	yes/no
(e.g. adenoma sebaceous/lipomata/neurofibromas/schwannomas/skin appendages)	
Blue naevus*	yes/no
Axillary freckling*	yes/no
Thin skin/generalized skin atrophy*	yes/no

**NEUROLOGICAL**

Ataxia	yes/no	if yes, please specify....
Cranial nerve palsies	yes/no	if yes please specify....

**ENDOCRINE**

Enlarged thyroid	yes/no
------------------	--------

Score form submitted by physician (name and function):

Hospital:

Date:

\*: Illustrating picture available (Tumor Predisposition Syndromes in Childhood Cancer Screening Instrument, part II: Definitions and pictures)

# Tumor Predisposition Syndromes in Childhood Cancer Screening Instrument

part II: Definitions and pictures

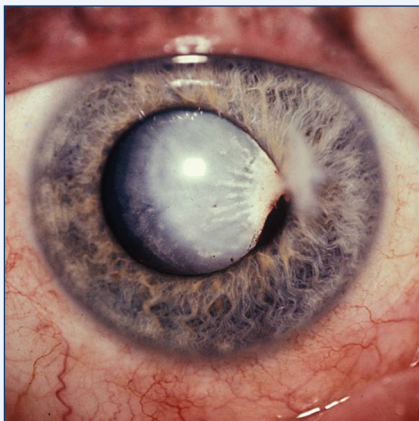


# Brittle hair



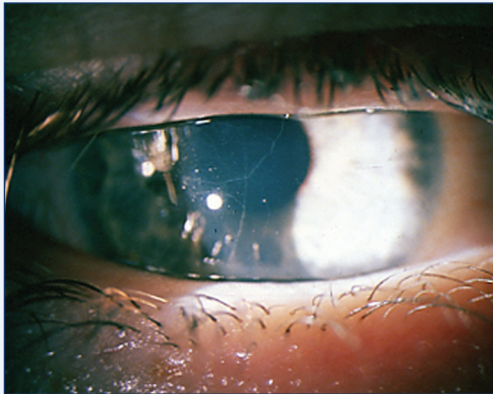
Easily breaking  
(often also thin) hair

# Cataract



Lens opacities of all types

# Visible nerve fibres on cornea



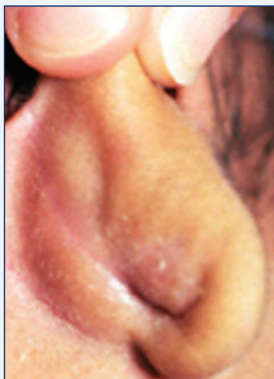
Prominent nerve fibres on the cornea

# Photosensitivity



Hypersensitive reaction to sunlight of eye and/or skin

# Crease/pits of ear lobule



Sharply demarcated, typically linear and approximately horizontal indentations in the ear lobe



## (Posterior) pits of ear helix



Permanent indentation on the posteromedial aspect of the helix that may be sharply or indistinctly delineated

# Leukoplakia



White, slightly raised plaques on the mucosal surface



# Tongue (large, protruding, lobulated)



- Large tongue: Increased length and width of the tongue
- Lobulated tongue: Multiple indentations and/or elevations on the edge and/or surface of the tongue producing an irregular surface contour
- Protruding tongue: Tongue extending beyond the alveolar ridges or teeth at rest

# Oral pigmentation



Increased pigmentation,  
either focal or generalized,  
of the oral mucosa

# Abnormal oral mucosal (cobblestone)



Thickened oral mucosa with irregular surface

# Mucosal neurinomas



Painful elevations on the edge or surface of the tongue

# Papilloma peri-orificial



Small epithelial tumor with a wart-like appearance

# Supernumerary nipples



Uni/bilateral additional nipple(s), either complete or as indentation



# Umbilical hernia



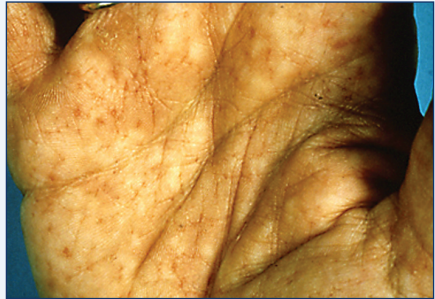
Protruding swelling including the umbilicus

## Asymmetry of extremities (length, width or both)



Difference in the length,  
width or both of arms  
and/or legs

# Palmar pits



Pin-point indentations in the palm(s) of the hand

# Abnormal genital pigmentation



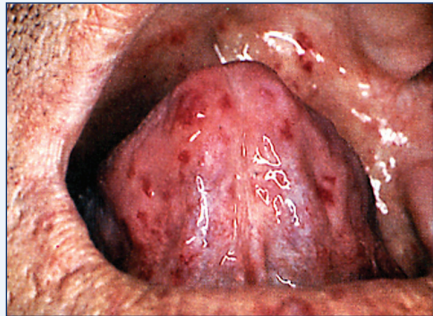
Localized or  
generalized increased  
or decreased genital  
pigmentation

# Ambiguous genitalia



External genitalia without either typical male or female appearance

# Teleangiectasia (conjunctival, nasal, oral)



Dilated tangled small blood vessels on the conjunctivae or nasal/oral mucosa

# Tumor

(e.g. lipomata, adenoma sebaceous, neurofibromas, schwannomas, skin appendages)

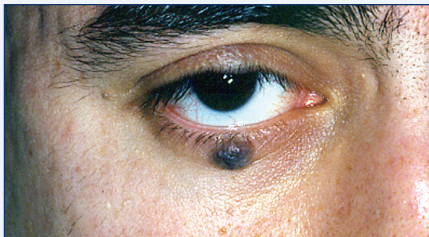


Tumors on or just below the skin varying in consistency, size, and colour

# Blue naevus



Round, bluish-purple and somewhat elastic swelling





# Axillary freckles



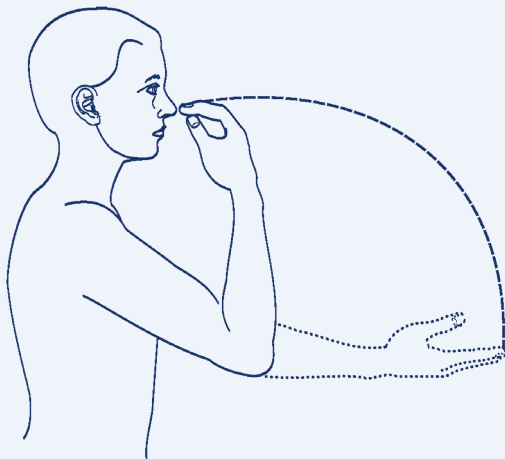
Freckles in the arm pits

# Thin skin/generalized skin atrophy



Thin, almost translucent skin, often with decreased subcutaneous (fat) tissue

# Ataxia



1. When bringing finger to nose, patient cannot touch the tip of the nose in a single fluent motion (dysmetria, either hypo- or hypermetria), or has increasing tremor in the vicinity of the target (intention tremor)
2. When walking, the patient is not able to follow a straight line

When the patient scores “yes” for one of these items, please score ataxia as “yes” on the score form and please clarify which item was disturbed (1, 2, or combination)

# Cranial nerve palsies (I)

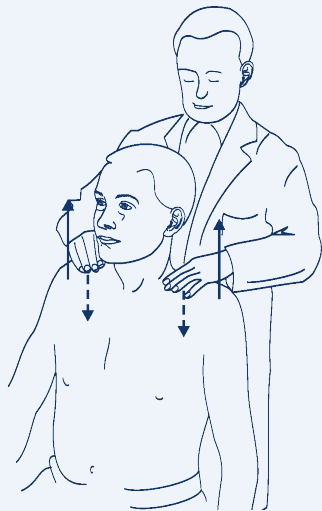
1. Patient has asymmetric pupils or has one or two eyelids overhanging part of the pupil
2. Patient has jerky eye movements when following finger or disturbance in the normal balance of eye control (nystagmus)
3. Patient has a squint (strabismus) or double vision (diplopia)
4. Sensation to light touch is abnormal on either side of the face
5. Patient's face shows asymmetry when raising eyebrows, closing eyes or showing teeth

When the patient scores “yes” for one of these items, please score cranial nerve palsies as “yes” on the score form and clarify which item was disturbed (1, 2, 3, 4, 5 or combination)

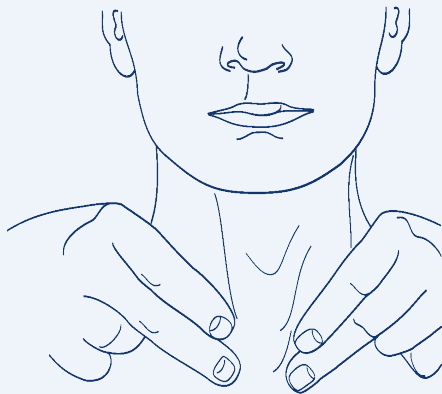
## Cranial nerve palsies (2)

6. Patient has a hearing disability or vertigo
7. Patient has difficulty swallowing, a hoarse voice or a dysarthria
8. Patient cannot hold shoulders in shrugged position against resistance or rotate head against resistance
9. Tongue of the patient deviates when protruding or shows increased folds and/or wasting (atrophy)

When the patient scores “yes” for one of these items, please score cranial nerve palsies as “yes” on the score form and clarify which item was disturbed (6, 7, 8,9 or combination)



# Enlarged thyroid



Thyroid visibly enlarged or enlarged on palpation