## SUPPLEMENTARY DATA 3

## Supplementary data S3.1 Score form Delphi round 1

1- no contribution to diagnosis tumor predisposition syndrome
the diagnosis of known tumor predisposition syndromes in the first screening of a patient.
Please score (on a scale from 1 to 5) which morphological abnormalities would contribute to



Academic Medical Center, Amsterdam Principal investigator: Prof. dr. R. C. M. Hennekam Co-investigators: dr. J. H. M. Merks, S. M. J. Hopman Version: 22nd of February 2011

1- no contr	bution to diag	nosis tumo	or predisp	osition sy	ndrome		
2							
3							
4							
5- pathogn	monic for dia	anneis tun	nor predis	enosition s	vndrome		

Morphological abnormality	Score for contribution to diagnosis tumor predisposition syndrome		
STATURE	N/A		
Short stature, proportionate			
Short stature, short limbs			
Short stature, short trunk			
Tall stature, proportionate			
Tall stature, disproportionate/dolichostenomelia			

CRANIUM	N/A
Brachycephaly	
Dolichocephaly/scaphocephaly	
Macrocephaly	
Microcephaly	
Plagiocephaly/asymmetrical skull	
Trigonocephaly	
Flat occiput	
Prominent occiput	
Scalp defects	
Scale tumoure	

HAIR	N/A
Brittle hair	
Fine hair	
Kinky/curly hair (including pili torti)	
Coarse hair	
Abnormal secondary sexual hair	
Unusual hair whorl/pattern	

EARS	N/A
Absent auditory canal	
Narrow/atretic auditory canal	
Pits of ear helix	
Crease/pits of ear lobule	
Temporal bone aplasia/dysplasia	
Posterior helical pits	

EYES, GLOBES	N/A	
Conjunctivitis		
Conjunctival nodules		
Conjunctival telangiectasia		
Cloudy corneae/sclerocornea		
Keratoconus		
Macrocornea/megalocornea		
Visible nerve fibres on cornea		
Microcomea		
Ulceration of cornea		
Microphthalmia		
Iris atrophy/dysplasia		
Cataract		
Persistent pupillary membrane		
Nevus of Ota		
Vision, non-specific impairment		
Blindness		
Hypermetropia		
Myopia		
Nystagmus		
Photophobia		
Strabismus/gaze palsy		

EYES, ASSOCIATED STRUCTURES	N/A	
Double eyelashes		
Flame nevus of eyelids		
Ptosis of eyelids		
Blocked/absent nasolacrimal duct		
Nasolacrimal duct, abnormal position		
Ingranged toor production		

NOSE	N/A
Deviated nasal septum	
Nasal telangiertasia	

FACE	N/A
Expressionless/dull face	
Facial weakness	
Telangiectasia	
Asymmetrical maxilla	
Prominent maxilla	

MOUTH	N/A
Open mouth appearance	
Stomatitis	
Trismus	

ORAL REGION	N/A
Oral tumour	
Leukoplakia	
Oral frenula (multiple)	
Oral pigmentation	
Thick/wide alveolar ridges	
Gingivitis	
Gum hypertrophy	
Periodontoclasis/periodontitis	
Cleft palate	
Cleft uvula	
High palate	
Narrow palate	
Prominent lateral palatine ridges	
Submucous cleft palate	
Torus palatinus	
Absent or hypoplastic tongue	
Bound tongue/ankyloglossia	
Midline cleft tongue	
Furrowed tongue/prominent groove	
Glossoptosis	
Large tongue	
Lobulated tongue (including hamartomata)	
Protruding tongue	

TEETH	N/A	
Advanced tooth eruption/development		
Dental caries		
Delayed tooth eruption/development		
Enamel abnormalities		
Irregular or crowded teeth		
Large/prominent teeth		
Malocclusion of teeth		
Neonatal teeth		
Oligodontia		
Premature loss of teeth		
Abnormally shaped teeth		
Small teeth		
Single central incisor		
Talon cusp		
Supernumerary teeth		
Wide-spaced teeth		

VOICE	N/A
Hoarse voice	
Nasal speech	
High-pitched voice	
Low-pitched voice	
Speech defect/dysarthria	
Speech delay	

NECK	N/A
Branchial cleft/sinus/cysts	
Low posterior/trident hairline	
Loose skin in neck	
Torticollis, including stiff neck	

BACK AND SPINE	N/A
Pilonidal cyst/sinus/dimple	
Scoliosis	

THORAX	N/A	
Asymmetric thorax		
Short thorax		
Short sternum		
Stridor		
Asternia or bifid sternum		
Gynaecomastia		
Supernumerary nipples		
Wide-spaced nipples		
Absent or hypoplastic pectorals		
Winged scapulae		

ABDOMEN	N/A	
Diastasis recti		
Inguinal hernia		
Umbilical hernia		
Abdominal muscle hypoplasia/aplasia		
Abnormal placement of anus		
Anal fistula		

GENITALIA	N/A	
Ambiguous/absent genitalia		
Female pseudo hermaphroditism		
Male pseudo hermaphroditism		
True hermaphroditism		
Hypogonadism		
Abnormal genital pigmentation		
Prominent clitoris		
Abnormal labia		
Early puberty in females		
Late puberty in females		
Hydrocele/lymphedema of testis		
Hypospadias		
Large penis		
Small penis (including micro)		
Early puberty in male		
Late puberty in male		
Hypoplastic scrotum		
Overriding scrotum		
Absent testis		
Cryptorchid testes		
Large testes		
Small testes		
Scrotum, bifid		

UPPER LIMBS	N/A	
Asymmetric arms		
Mesomelia of upper limbs		
Rhizomelia of upper limbs		
Proportionate short arms		
Limited movement of shoulder		
Cubitus valgus		
Dislocated elbow		
Limited movement/flexion deformity of elbow		
Madelung deformity		
Restriction of supination/pronation		
HANDS	N/A	
Large hands		
Small hands		
Small hands Thenar hypoplasia		

HANDS	N/A	
Large hands		
Small hands		
Thenar hypoplasia		
Abnormal dermatoglyphic patterns		
Hyper-mobile/extensible fingers		
Post-axial polydactyly of fingers		
Limited movement of fingers		
Osseous syndactyly of fingers		
Short/hypoplastic metacarpals		
Synostosis of metacarpals		
Hypoplastic phalanges		
Short phalanges		
Wide phalanges		
Abducted/hitchhiker thumb		
Fetal finger pads		

NAILS	N/A
Thickened nails	
Thin/hrittle naile	

LOWER LIMBS	N/A	
Asymmetric lower limbs		
Hypertrophy of lower limb		
Mesomelia of lower limbs		
Oedema of lower limbs		
Rhizomelia of lower limbs		
Proportionate shortening of lower limb		
Flexion deformity of hip		
Coxa vara		
Coxa valga		
Absent or hypoplastic patella		
Dislocated patella		
Dislocated knee		
Hyper-extensible knees		
Limited movement of ankle		

FEET	N/A	
Flat arches of feet		
High arches of feet (pes cavus)		
Abnormal/deep plantar creases		
Abnormal dermatoglyphics on feet		
Large feet		
Small feet		
Vertical talus (see also rocker-bottom feet)		
Hyper-mobile toes		

ENDOCRINE	N/A
Ectopic/enlarged thyroid	

MUSCLES	N/A		
Muscle atrophy			
Myotonia			
Muscle weakness/myopathy			

JOINTS	N/A
Contractures (including arthrogryposis)	
Laxity	
Stiffness/arthritis	

NEUROLOGY	N/A	
Anosmia		
Ataxia		
Cranial nerve palsies		
Dementia/psychosis		
Extra-pyramidal disorder		
Hemiplegia		
Hypotonia		
Mental retardation/developmental delay		
Spasticity/increased tendon reflex		
Tremors		

SKELETAL	N/A
Hemi-hypertrophy	

SKIN	N/A		
Cutis laxa			
Dimples			
Hypohidrotic or dry skin			
Hyperelastic skin			
Hyperkeratosis			
Keratosis pilaris			
Seborrhoea			
Soft skin			
Thick/stiff skin			
Thin skin/generalized skin atrophy			
Scleroderma			
Axillary freckles			
Mongolian spot			
Adenoma sebaceous			
Cysts			
Lipomata			
Neurofibromas/schwannomas			
Pedunculated skin lesions/skin tags			
Xanthomas			

		cannot realiably be disting	
a known tumor predisposit	on syndrome		

Please check whether you have put in a score for each item

## Supplementary data S3.2 Score form Delphi round 2

Please score for each item if you think this item should be included in a screening tool for known tumor predisposition syndromes or not

Please note:

\*This list contains only morphologic abnormalities which are not (reliable) visible on 2D or 3D picture series

\*Consider the morphological abnormalities as part of a tumor predisposition syndrome, not as the result of the malignancy

\*You can enter 0 or 1 as many times as you think is applicable, no minimum or maximum amount is set for both scores



Academic Medical Center, Amsterdam Principal investigator: Prof. dr. R. C. M. Hennekam Co-investigators: dr. J. H. M. Merks, S. M. J. Hopman Version: September 18th 2011

0- do NOT include in screening tool for known tumor predisposition syndromes 1- include in screening tool for known tumor predisposition syndromes

Morphological abnormality	Score for inclusion in screening tool
Short stature, proportionate	
Tall stature, disproportionate/dolichostenomelia	
Brittle hair	
Fine hair	
Iris atrophy/dysplasia	
Flame nevus of eyelids	
Asymmetric thorax	
Asternia or bifid sternum	
Supernumerary nipples	
Umbilical hernia	
Ambiguous/absent genitalia	
Prominent clitoris	
Early puberty in females	
Early puberty in male	
Large hands	
Tremors	
Cysts	
Microcephaly	
Pits of ear helix	
Cataract	
Asymmetrical maxilla	
Protruding tongue	
Loose skin in neck	
Cryptorchid testes	
Post-axial polydactyly of fingers	
Abnormal/deep plantar creases	
Cranial nerve palsies	
Hyperkeratosis	
Thin skin/generalized skin atrophy	
Tall stature, proportionate	
Scalp tumours	
Posterior helical pits	
Lobulated tongue (including hamartomata)	
Female pseudo hermaphroditism	
True hermaphroditism	
Asymmetric arms	
Lipomata	
Pedunculated skin lesions/skin tags	
Macrocephaly	
Kinky/curly hair (including pili torti)	
Crease/pits of ear lobule	
Visible nerve fibres on cornea	
Nasal telangiectasia	
Telangiectasia	
Oral tumour	
Leukoplakia	
Oral pigmentation	
Large tongue	
Male pseudo hermaphroditism	
Asymmetric lower limbs	
Hypertrophy of lower limb	
Ectopic/enlarged thyroid	
Ataxia	
Hemi-hypertrophy	

Additional items	Score for inclusion in screening tool			
Photosensitivity skin				
Palmar pits				
Mucosal neurinomas				
Hyperpigmentation				
Abnormal oral mucosa (cobblestone)				
Papilloma periorifical				
Discourse				

Scores round 1 (highlighted is own score) 1= no contribution to diagnosis tumor predisposition syndrome
5= pathognomonic for diagnosis tumor predisposition syndrome

1	1	3	2	3	3	2	3
3	3	3	2	1	2	2	4
1	2	3	1	4	4	1	3
1	1	3	2	3	4	3	1
1	1	2	4	3	5	4	1
3	2	3	1	2	3	3	1
3	1	1	2	3	3	1	3
3	1	3	3	4	2	1	1
							4
3	1	2	2	5	3	1 2	
2	3	3	4	2	3		2
1	3	2	2	5	5	4	1
1	1	3	2	3	3	3	1
1	2	3	2	4	4	1	3
2	2	3	2	4	4	1	3
2	2	2	3	3	3	4	1
3	1	2	2	3	3	1	3
3	2	5	2	3	3	2	1
1	3	4	2	3	4	3	4
1	1	5	2	4	4	4	1
3	2	2	2	4	4	3	3
3	1	3	1	4	3	1	5
3	1	2	4	3	5	2	5
1	2	3	3	3	4	3	1
2	1	3	2	3	3	3	5
2	3	3	1	3	3	4	3
2	1	3	3	4	4	4	1
4	4	2	2	4	5	1	1
3	2	2	3	3	3	1	5
1	3	3	2	4	4	3	5
3	3	4	2	3	4	4	4
4	3	5	2	4	5	1	3
1	1	5	3	4	4	4	1
4	3	4	1	5	5	1	1
1	3	4	2	5	5	2	5
1	3	4	3	5	5	1	5
4	3	4	3	4	3	3	4
4	3	4	1	3	3	4	4
4	3	3	4	4	3	2	5
4	4	3	4	3	4	4	5
2	1	4	4	4	4	4	3
1	4	4	4	4	4	4	1
4	5	5	4	5	4	1	1
4	4	4	2	2	4	4	1
4	4	4	4	4	4	4	4
2	4	4	4	4	4	3	3
1	4	5	2	5	4	3	5
4	4	5	3	4	4	4	5
3	4	3	4	5	5	4	5
1	3	4	2	5	5	4	5
4	1	4	3	4	4	3	4
4	3	4	4	4	5	4	4
4	4	5	4	3	4	4	3
4	4	2	2	4	5	3	5
4	4	4	4	4		4	5