

SUPPLEMENTARY DATA 3

Supplementary data S3.1 Score form Delphi round 1

Please score (on a scale from 1 to 5) which morphological abnormalities would contribute to the diagnosis of known tumor predisposition syndromes in the first screening of a patient.

1- no contribution to diagnosis tumor predisposition syndrome
2
3
4
5- pathognomonic for diagnosis tumor predisposition syndrome



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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	
Scalp tumours	

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 cornea/sclerocornea	
Keratoconus	
Macrocornea/mesalocornea	
Visible nerve fibres on cornea	
Microcornea	
Ulceration of cornea	
Microphthalmia	
Iris atrophy/dysplasia	
Cataract	
Persistent pupillary membrane	
Nevus of Ota	
Vision, non-specific impairment	
Blindness	
Hypermetropia	
Miopia	
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	
Increased tear production	

NOSE	N/A
Deviated nasal septum	
Nasal telangiectasia	

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	
Periodontoclasia/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	
Glossoposis	
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/ident hairline	
Loose skin in neck	
Torticollis, including stiff neck	

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

THORAX	N/A
Asymmetric thorax	
Short thorax	
Short sternum	
Stridor	
Asteria 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	
Overshining 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	
Thenar hypoplasia	
Abnormal dermatoglyphic patterns	
Hyper-mobile/extendible 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/witchhiker thumb	
Fetal finger pads	

NAILS	N/A
Thickened nails	
Thin/brittle nails	

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-extendible 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 arthrogyposis)	
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	
Seborrhea	
Soft skin	
Thick/stiff skin	
Thin skin/generalized skin atrophy	
Scleroderma	
Axillary freckles	
Mongolian spot	
Adenoma sebaceus	
Cysts	
Lipomata	
Neurofibromas/schwannomas	
Pedunculated skin lesions/skin tags	
Xanthomas	

Space for additional items (not listed above) which cannot reliably be distinguished on the 2D and/or 3D picture series, that you feel would contribute to the diagnosis of a known tumor predisposition syndrome

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

Thank you very much for your help!

