Childhood cancer: Indication for genetic counseling?*

*updated Jongmans criteria [Jongmans et al., 2016]

if at least one criterion is fulfilled, your patient may benefit from genetic counseling

1. Family history (3 generation pedigree)

O ≥2 malignancies occurred in family members before age 18 years, including index patient
O Parent or sibling with current or history of cancer before age 45 years
O ≥2 first or second degree relatives in the same parental lineage with cancer before age 45 years
O The parents of the child with cancer are consanguineous

2. One of the following Neoplasms was diagnosed:

O Adrenocortical carcinoma / adenoma
O ALL (low hypodiploid)
O ALL (ring chromosome 21)
O ALL (Robertsonian translocation 15;21)
O ALL relapse (*TP53* mutated)
O AML (Monosomy 7)
O Basal cell carcinoma
O Botryoid rhabdomyosarcoma of the urogenital tract (fusion-negative)
O Chondromesenchymal harmatoma
O Choroid plexus carcinoma / tumor
O Colorectal carcinoma
O Cystic nephroma
O Endolymphatic sack tumor
O Fetal rhabdomyoma
O Gastrointestinal stromal tumor
O Glioma of the optic pathway (with signs of NF1)
O Gonadoblastoma
O Hemangioblastoma
O Hepatoblastoma (*CTNNB1* wildtype)
O Hepatocellular carcinoma
O Infantile myofibromatosis
O Juvenile myelomonocytic leukemia
O Keratocystic odontogenic tumor
O Large cell calcifying Sertoli-cell-tumor
O Malignant peripheral nerve sheath tumor
O Medullary thyroid carcinoma
O Medulloblastoma (SHH activated)
O Medulloblastoma (WNT activated, *CTNNB1* wildtype)
O Medullary renal cell carcinoma
O Medulloepithelioma
O Melanoma
O Meningioma
O Myelodysplastic syndrome
O Myeloproliferative neoplasms (except CML)
O Myxoma
O Neuroendocrine tumor
O Paraganglioma / pheochromocytoma
O Parathyroid carcinoma / adenoma
O Pineoblastoma
O Pituitary adenoma / tumor
O Pituitary blastoma
O Pleuropulmonary blastoma
O Renal cell carcinoma
O Retinoblastoma
O Rhabdoid tumor
O Rhabdomyosarcoma with diffuse anaplasia
O Schwannoma
O Schwannomatosis
O Sertoli-Leydig cell tumor
O Sex cord stromal tumor with annular tubules
O Small cell carcin. of the ovary hypercalcemic type
O Squamous cell carcinoma
O Subependymal giant cell astrocytoma
O Thyroid carcinoma (non-medullary)
O Transient myeloproliferative disease
O Other rare cancers or cancers that typically occur in adults, unusually early manifestation age

3. O **Genetic tumor analysis reveals defect suggesting a germline predisposition**

4. O **A patient with ≥2 malignancies (**e.g. secondary, bilateral, multifocal, metachronous**)**

5. O **A child with cancer and congenital or other anomalies**

Sign	***Think of***
O Congenital anomalies	Abnormal organs, skeletal anomalies, oral clefting, abnormal teeth, urogenital anomalies, abnormal hearing or vision, etc.
O Facial dysmorphism	
O Mental impairment, developmental delay	Abnormal behavior, learning difficulties
O Abnormal growth	Height, head circumference, birth weight, hemihyperplasia, growth chart
O Skin anomalies	Abnormal pigmentation such as ≥2 café-au-lait spots, vascular lesions, hypersensitivity to sun, benign tumors, etc.
O Hematological abnormalities (not explained by current cancer)	Pancytopenia, anemia, thrombocytopenia, neutropenia, leukopenia, macrocytic erythrocytes
O Immune deficiency	Frequency of infections, lymphopenia
O Endocrine anomalies	Primary hyperparathyroidism, precocious puberty, gigantism/acromegaly, Cushing syndrome

6. O **The patient suffers from excessive toxicity of cancer therapy**