

**SarcBOP Registerstudie**

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## Checklist genetic risk

Dear patient,

With this letter, we would like to inform you about the background and benefits of the checklist for genetic risk in sarcoma patients, and the options available to you in the case of an existing genetic risk.

### The questionnaire asks five questions:

The **first** question relates to the diagnosis and exact name of the sarcoma. The following questions are asked:

- a. Rhabdomyosarcoma - a sarcoma that originates in the skeletal muscles
- b. Osteosarcoma - a sarcoma that originates in the bone
- c. Sarcoma of the cervix uteri - A sarcoma in women that originates in the cervix
- d. pleuropulmonary blastoma - an extremely rare, primary sarcoma in the thoracic cavity that occurs in childhood and originates from the pleura or lung tissue
- e. MPNST (malignant peripheral nerve sheath tumour) - a sarcoma that originates from tissue surrounding nerves.
- f. abdominal fibromatosis **without** evidence of a CTNNB1 mutation - a sarcoma that occurs in the abdomen **and lacks of a specific genetic alteration**
- g. gastrointestinal stromal tumor **without** evidence of a KIT or PDGFRA mutation – Gastrointestinal stromal tumors are rare soft tissue sarcomas in the gastrointestinal tract that often exhibit specific genetic alterations, such as KIT or PDGFRA mutations. Only if these **genetic alterations are absent** should this option be marked with a cross
- h. intracranial sarcoma – A sarcoma located in the brain or the meninges

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The **second** question concerns your medical history:

- a. Two bone or soft tissue tumors, one of which is  $\leq 45$  years of age (first diagnosis) - have you been diagnosed with two different sarcomas and did one occur at or below the age of 45?
- b. Occurrence of multifocal bone and soft tissue tumors - did the sarcoma occur simultaneously at different sites at the time of diagnosis?

The **third** question concerns other changes in your body:

- a. Skin changes, e.g., café-au-lait spots (pigment changes of the skin), pits (pilonidal sinuses), skin tumors, etc.
- b. Growth abnormalities, e.g., macrocephaly (large skull), asymmetries, exostoses (bone overgrowths), myxomas (benign heart tumors), osteochondromas (benign tumors near growth plates consisting of bone and cartilage tissue)
- c. Intellectual developmental disorder
- d. Congenital malformations
- e. Syndromal (distinctive) facial features, such as those observed in Down syndrome or Turner syndrome

The **fourth** question aims at detecting genetic alterations in the sarcoma that indicate a general genetic change in your body (germline variant, inherited cancer predisposition, e.g., Li-Fraumeni syndrome).

The **fifth** question focuses on diseases in your family:

- a.  $\geq 1$  **first- or second-degree** relative diagnosed with cancer in childhood or adolescence
- b.  $\geq 1$  **first-degree** relative with the same tumor or with a cancer diagnosis  $\leq 45$  years of age
- c.  $\geq 2$  **second-degree** relatives with the same tumor or with a cancer diagnosis  $\leq 45$  years of age
- d.  $\geq 1$  **first- or second-degree** relative with breast cancer  $\leq 45$  years, brain tumor or rare cancer (e.g. adrenocortical carcinoma, paraganglioma).  
<https://www.rarecancerseurope.org>

How do I determine the degree of relationship?

**1st degree** = parents and children

**2nd degree** = grandparents and grandchildren; siblings

**3rd degree** = great-grandparents, great-grandchildren, uncle/aunt, nephew/niece

**4th degree** = great-great-grandparents, great-great-grandchildren, great-uncle/great-aunt, cousin (cousin)/cousin (base), son/daughter of nephew/niece

**5th degree** = grandson/granddaughter of nephew/niece

As soon as you provide an answer other than "Not applicable" to any question, genetic counseling should be considered. Please discuss this with your treating oncological medical team. They can provide you with a referral for genetic counseling.

If you have any further questions regarding the genetic checklist, please speak with your treating oncological medical team or contact us by phone (06221-56-36235) or email ([SarcBOP@NCT-heidelberg.de](mailto:SarcBOP@NCT-heidelberg.de)).

Thank you and best regards,  
Your SarcBOP Study Team