WEICHTEILKNOTEN

Wie stellt der Pathologe die Diagnose?

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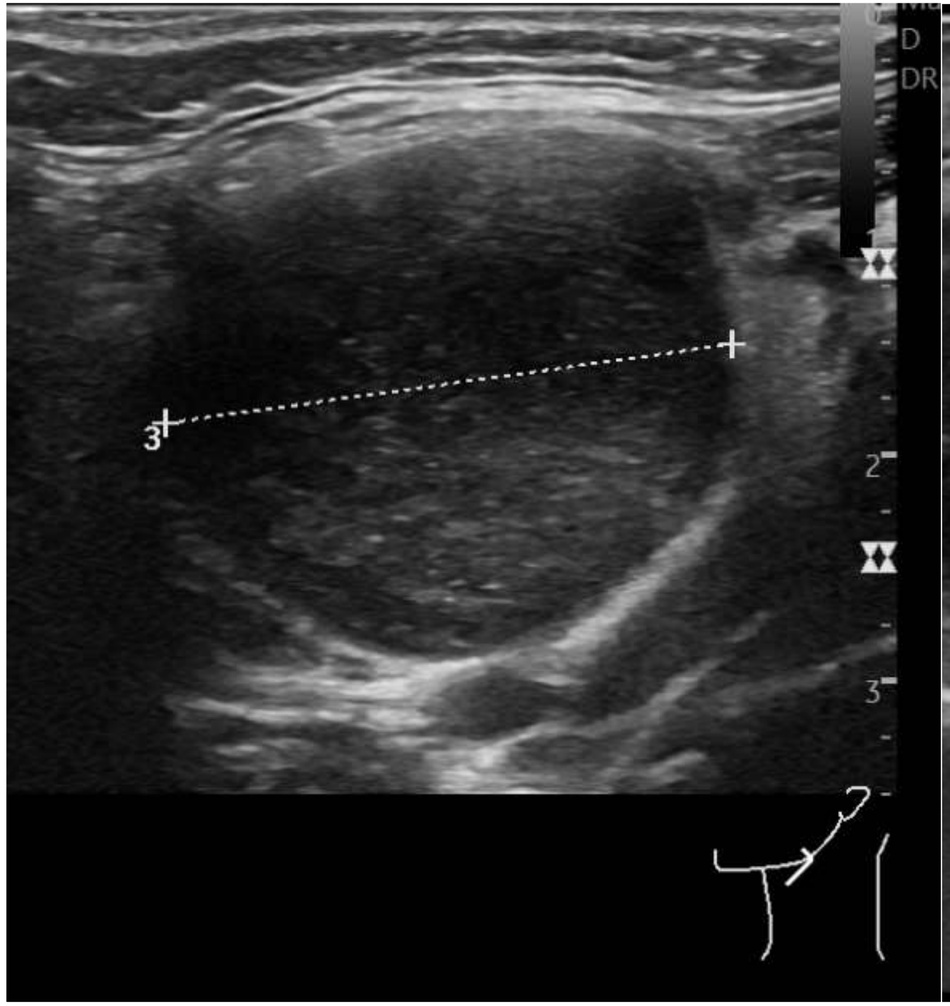
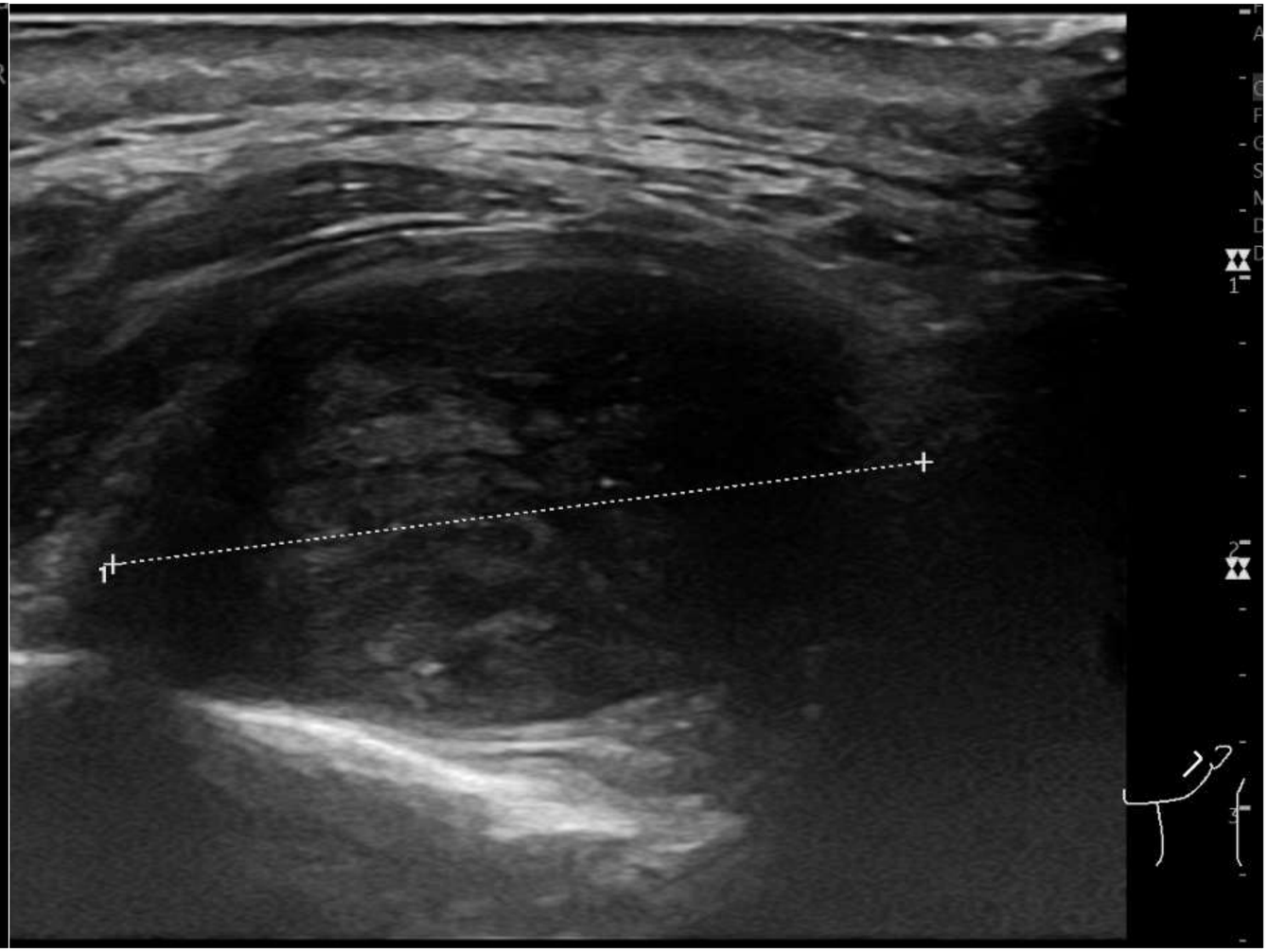
Peter Bode
Chefarzt und Institutsleiter

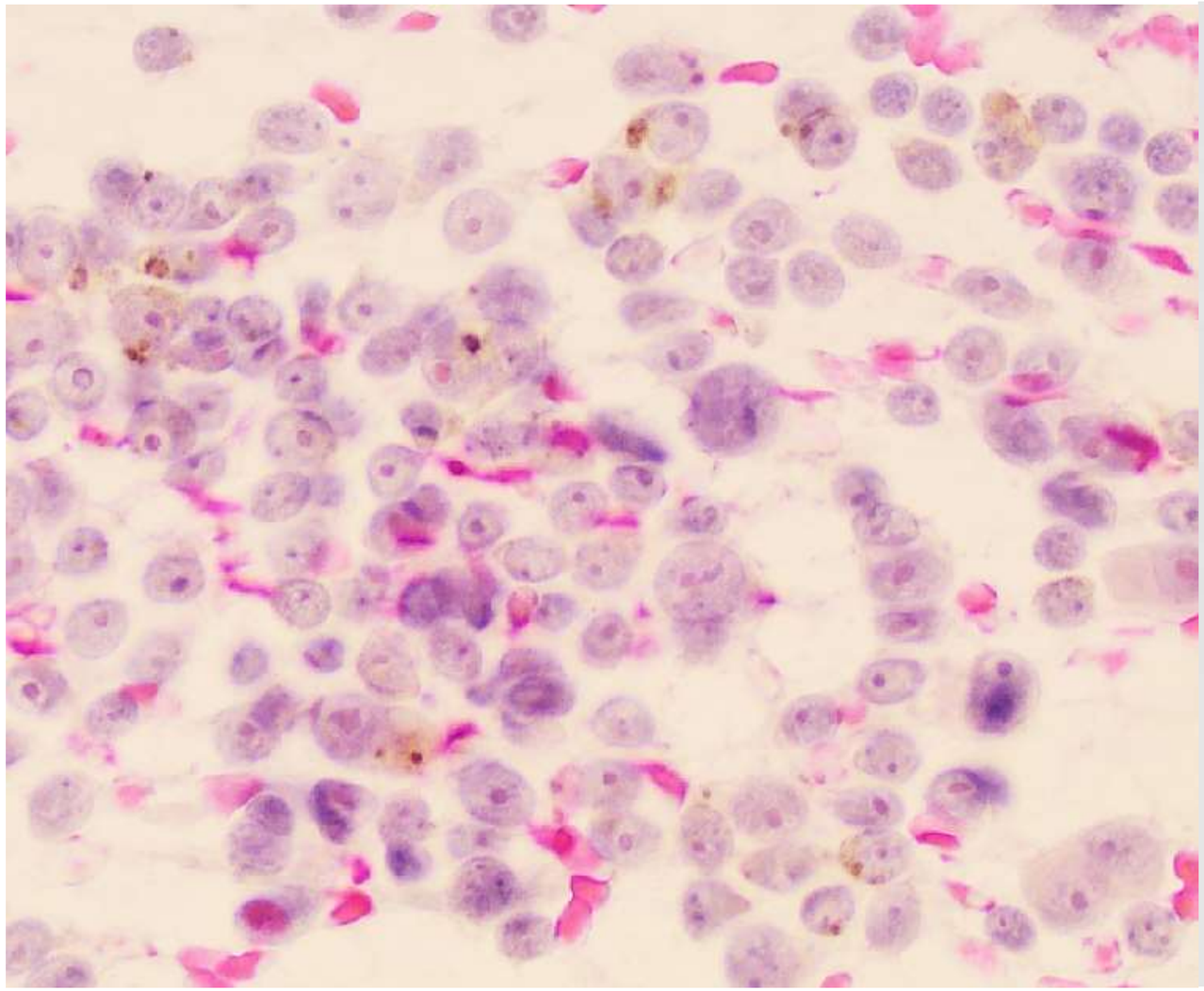
KSW Kantonsspital Winterthur

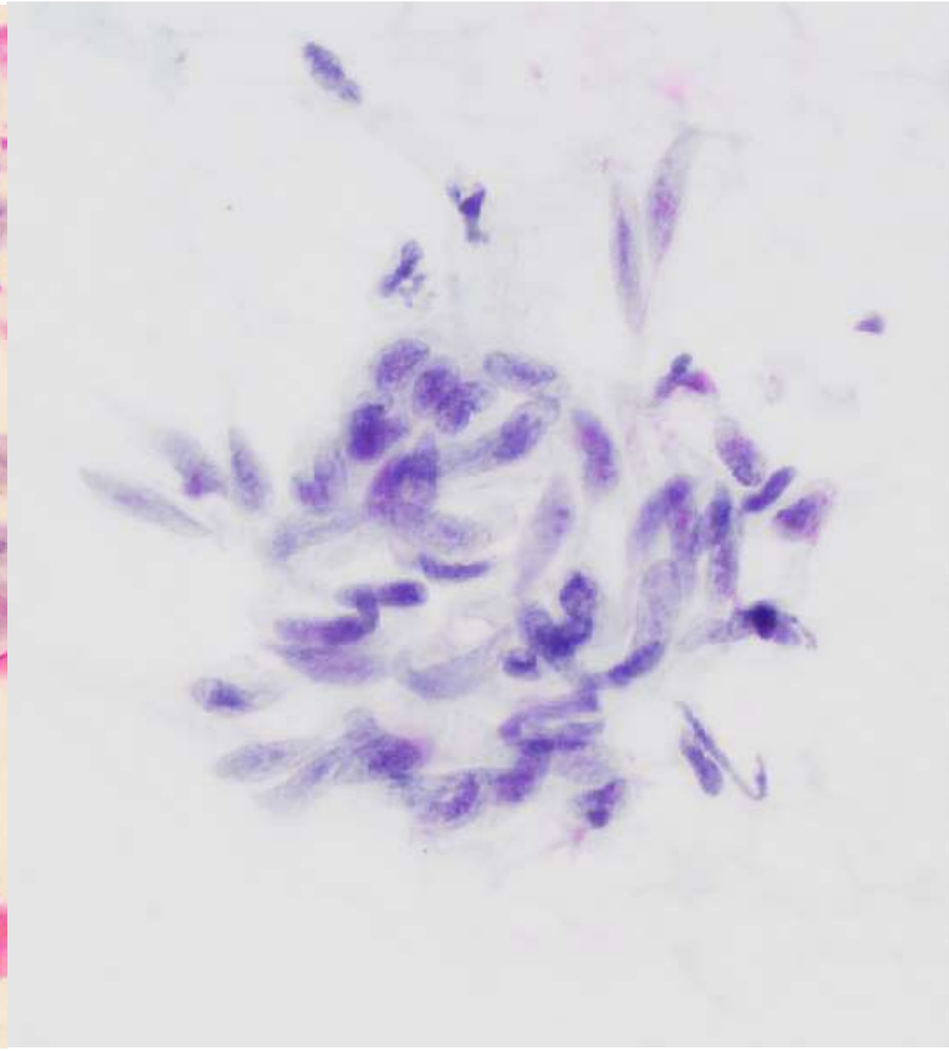
Zwei Patienten mit subkutanen Knoten: Diagnose?



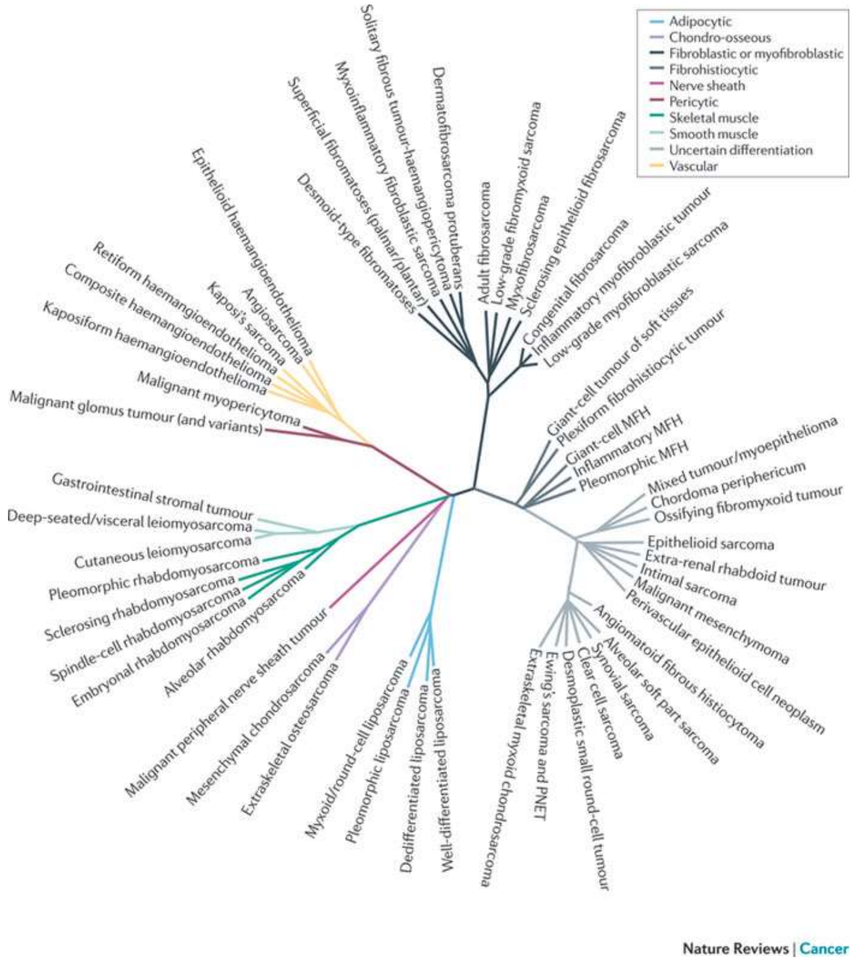


Zwei Patienten mit subkutanen Knoten: Diagnose?

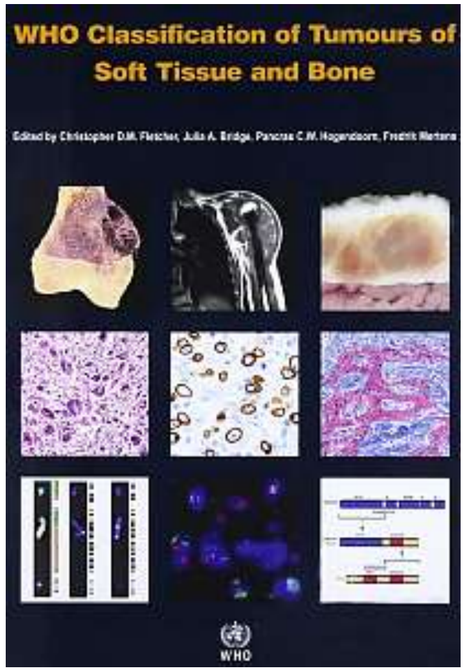




1. Herausforderung: Weichteiltumoren sind komplex!

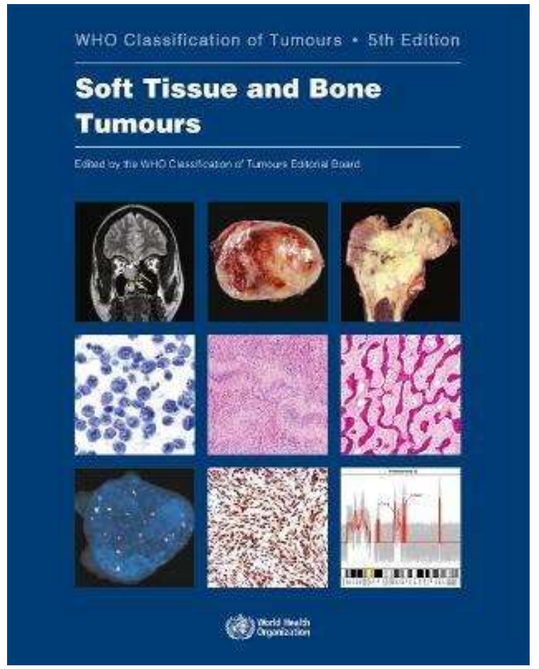


2013



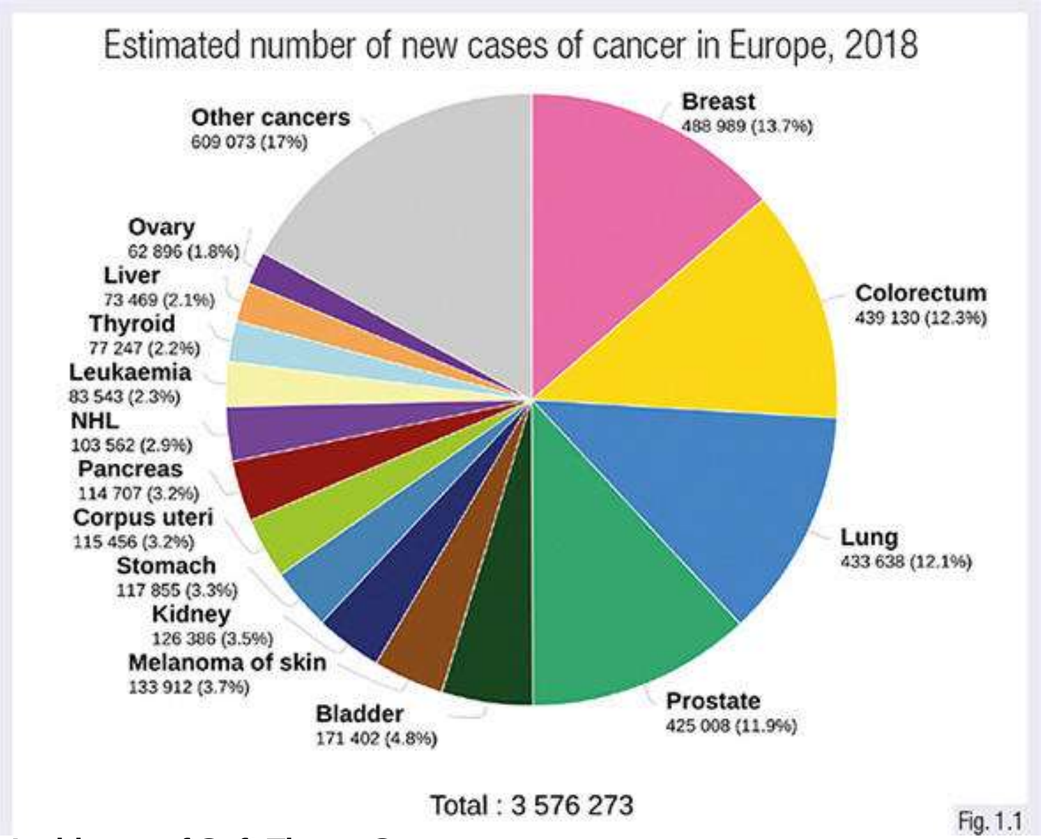
1342 g
468 Seiten

2020



1784 g (+ 33 %)
607 Seiten (+ 30 %)
> 100 Entitäten

2. Herausforderung:
Maligne Weichteiltumoren sind selten!

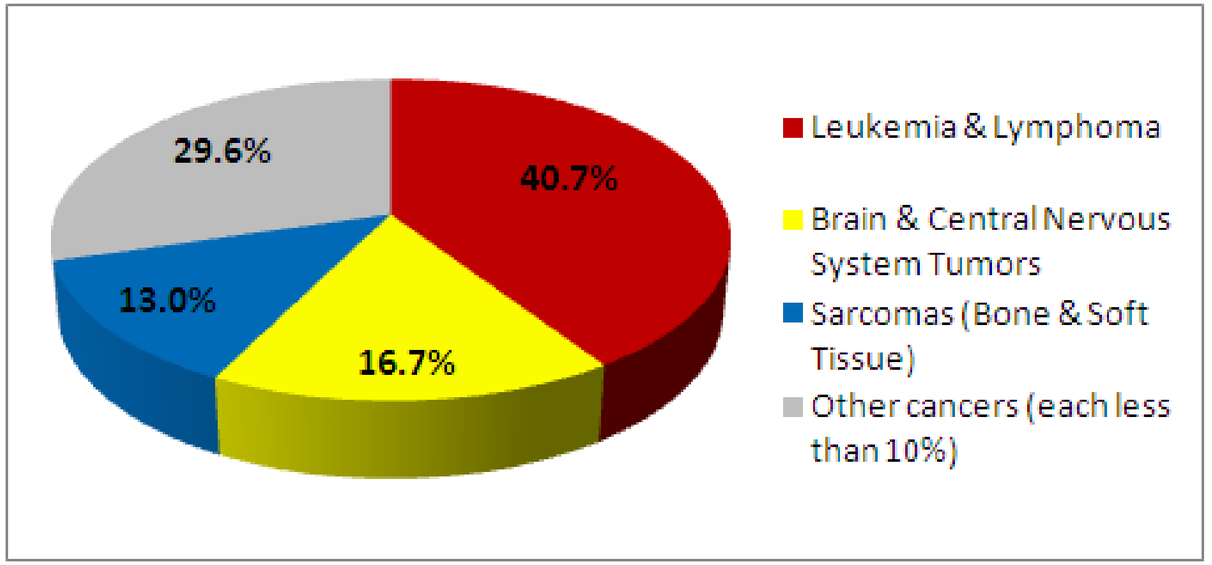


Incidence of Soft Tissue Sarcoma
2-3 new cases pro 100 000 / year
Switzerland: 200-250

Source: ESMO

< 20 Jahren

Figure 1: Percent Distribution of Childhood Cancers by Category



Based on SEER data from 1975-1995 for children less than 20 years old.

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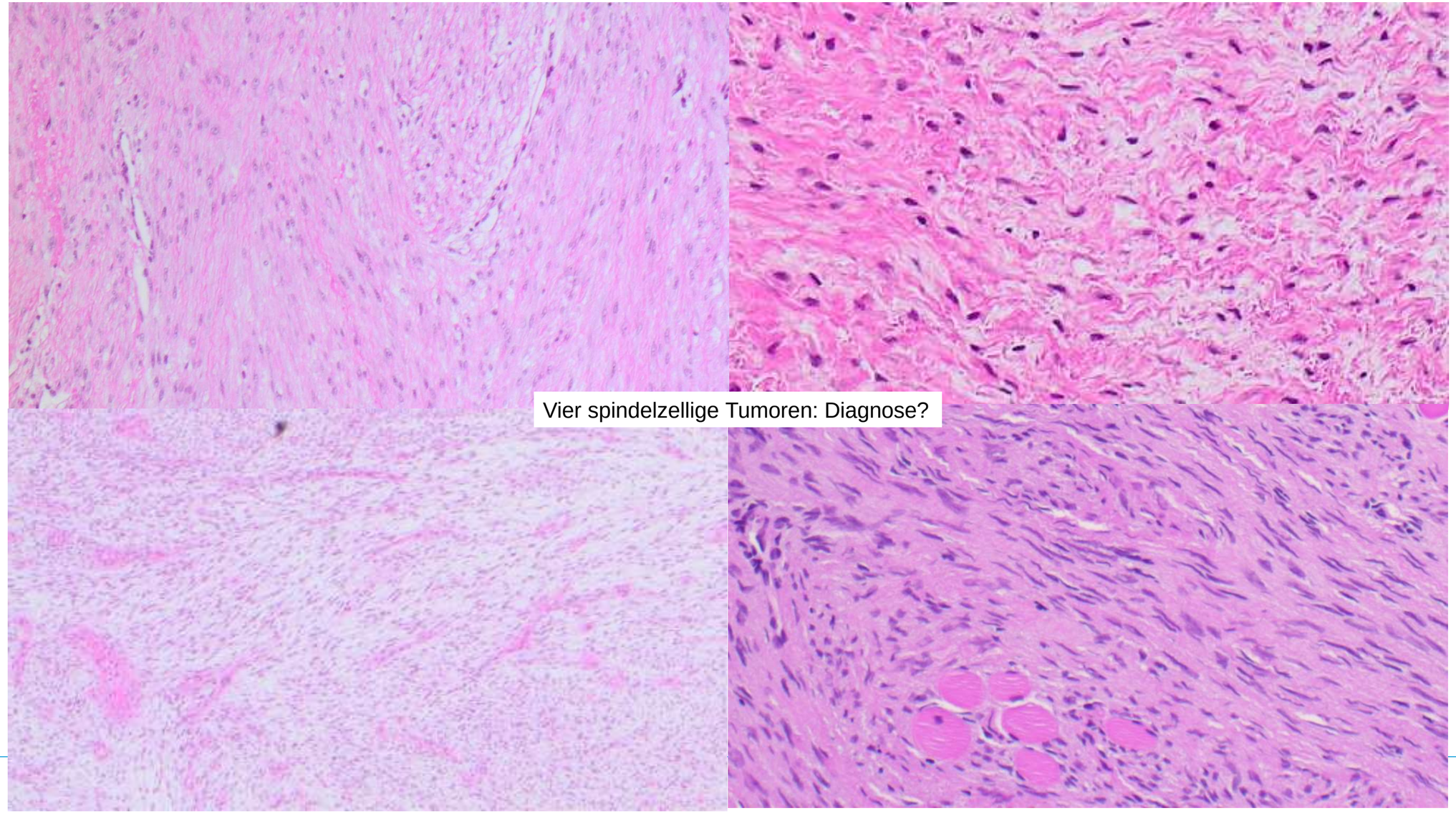
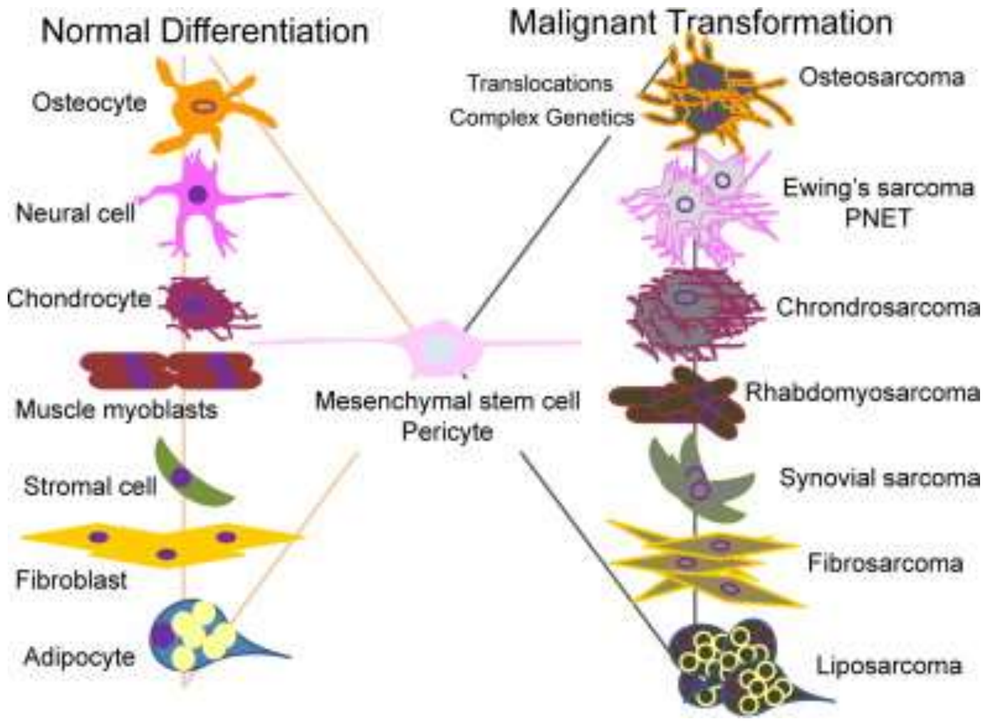
3. Herausforderung:
Weichteiltumoren sind morphologisch ähnlich!



Vier spindelzellige Tumoren: Diagnose?

Wie werden Weichteiltumoren / Sarkome klassifiziert?

Histology and Immunophenotype



From Teicher, Biochemical Pharmacology, 2012

Molecular Genetics

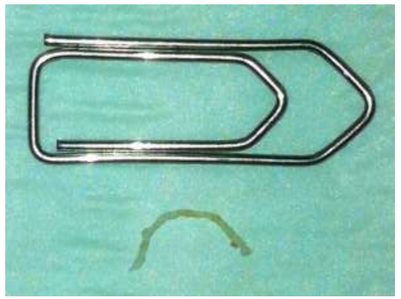
1.Near diploid karyotypes and simple genetic alterations

- Translocations / gene fusions
- specific activating mutations (e.g. *c-kit* in gastrointestinal stroma tumor / GIST)
- Deletions (e.g. *SMARCB1* in malignant rhabdoid tumor)
- Amplifications

2. Complex and unbalanced karyotypes

- Genomic instability
- Multiple genomic aberrations
- Heterogeneity of aberrations across tumor type

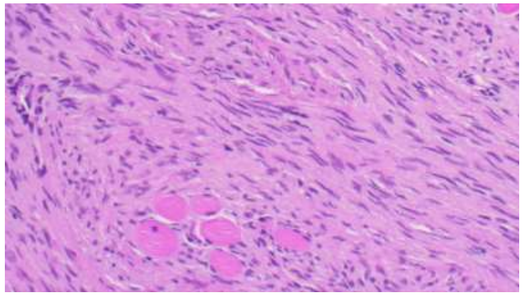
Welche Werkzeuge uns zur Verfügung?

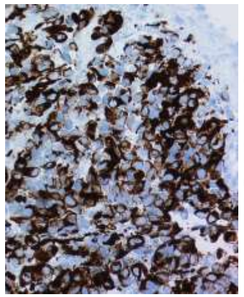


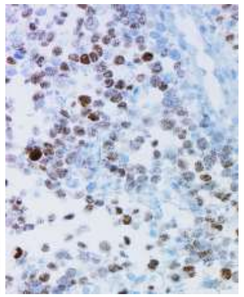
1-2 Tage

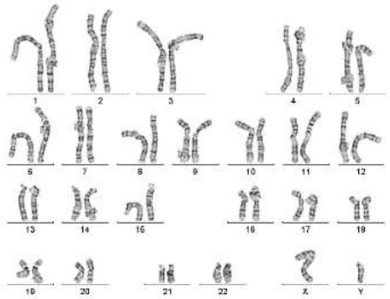
+ 3-4 Tage

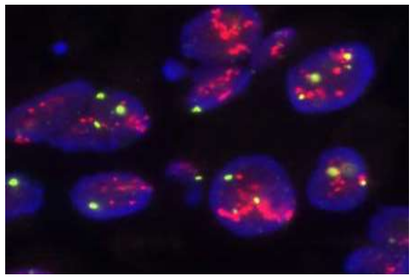
+ 7-10 Tage

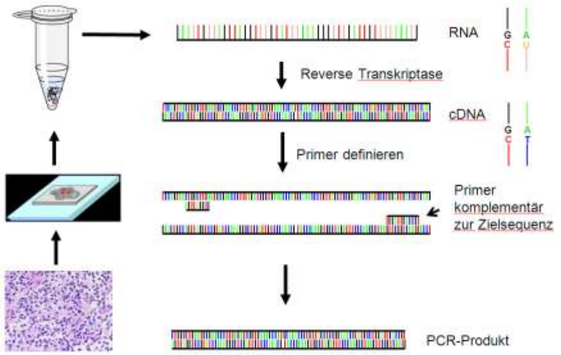










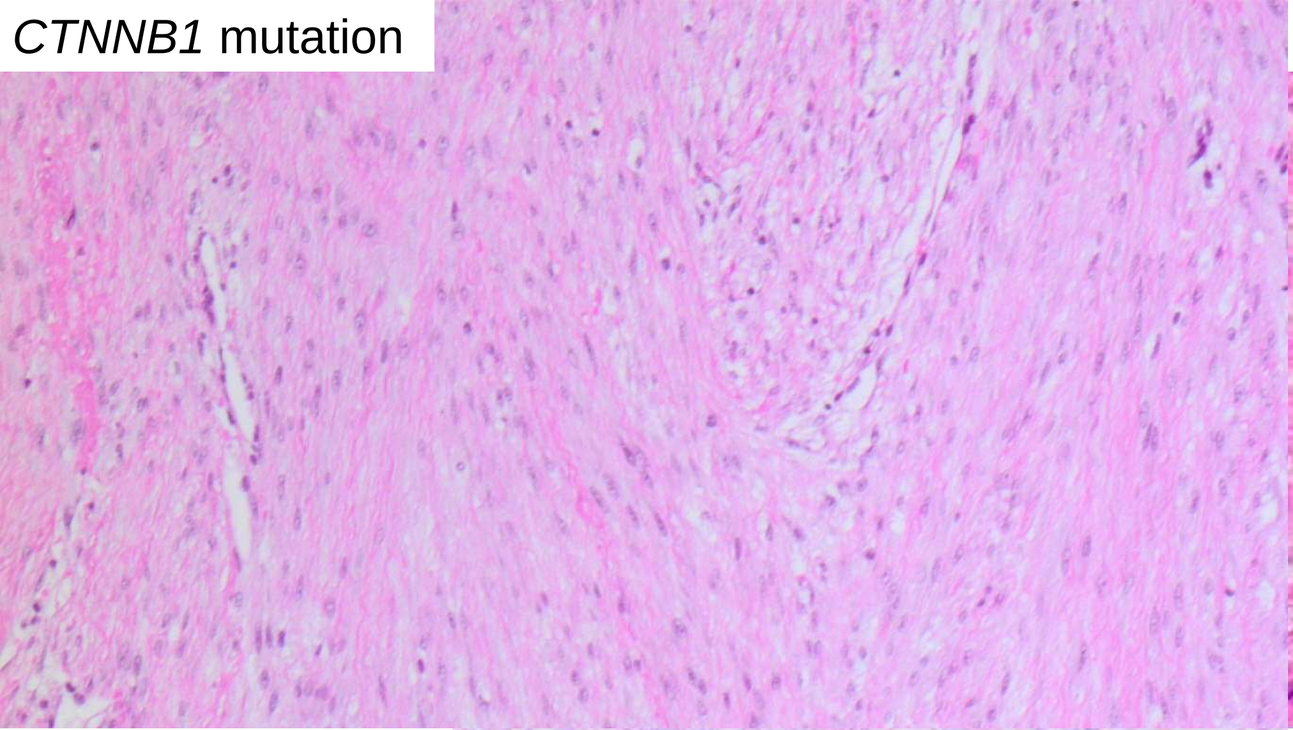


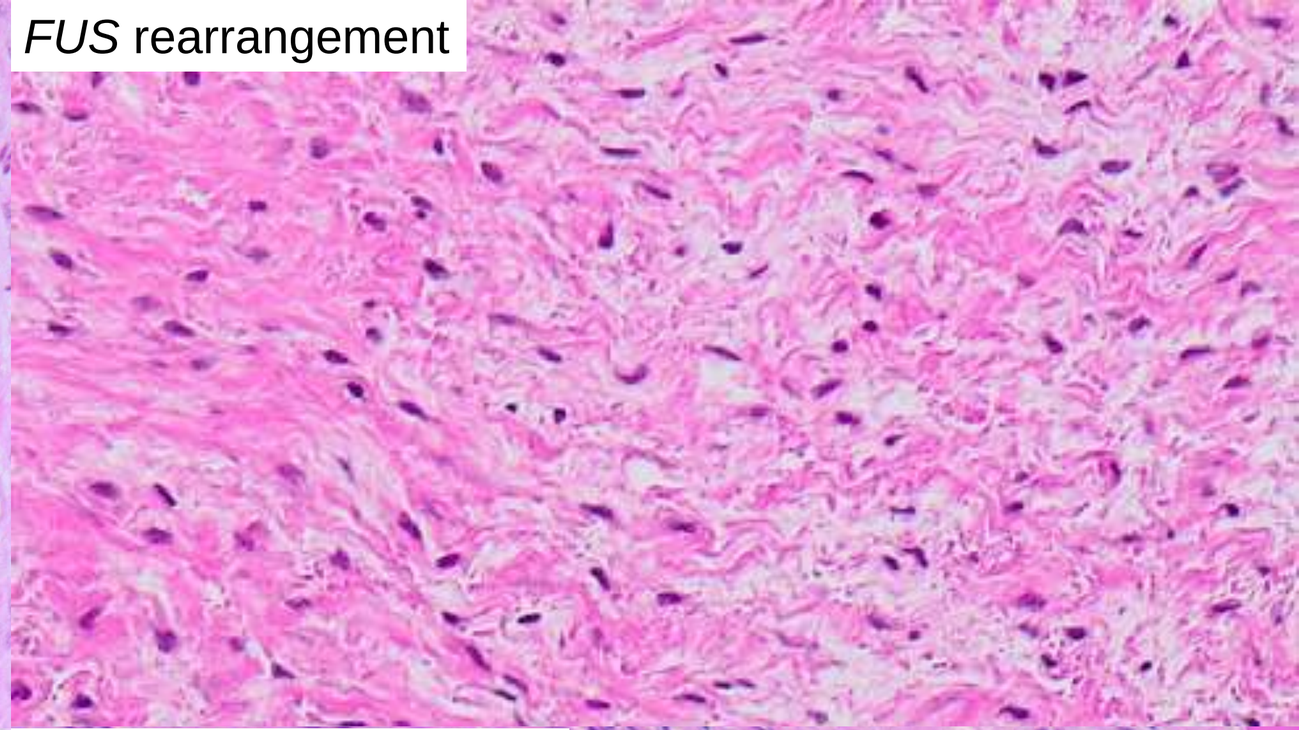
Konventionelle Morphologie
- Mustererkennung
- "Blickdiagnosen" vs Differentialdiagnosen

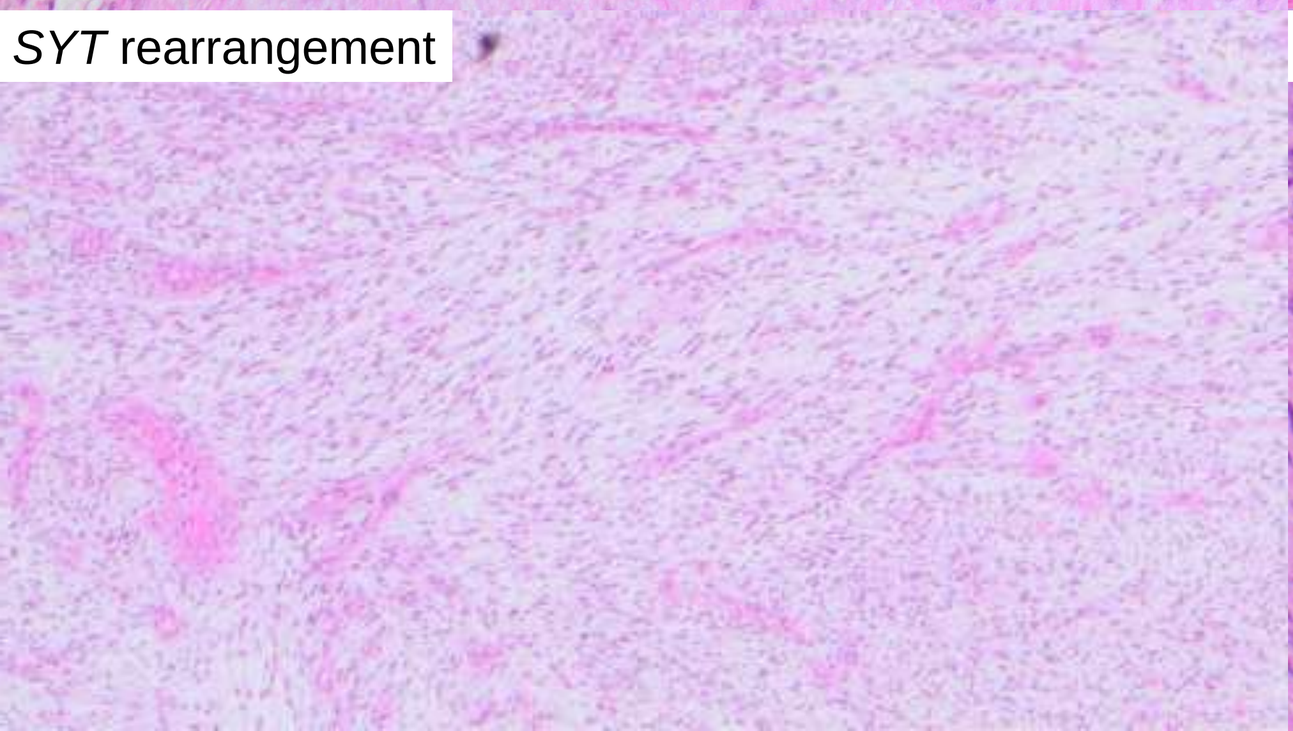
Immunhistochemie
- Liniendifferenzierung
- Surrogatmarker für genetische Aberrationen

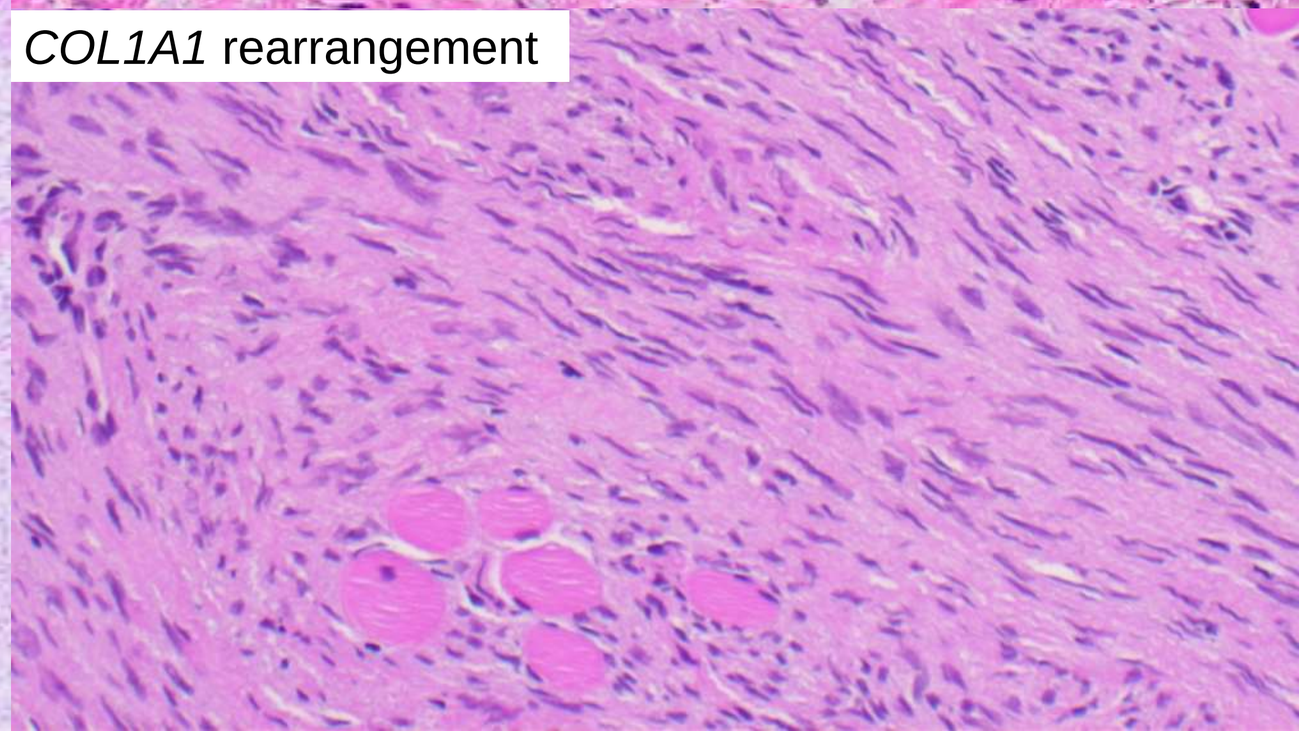
Fluoreszenz-in-situ Hybridisierung:
- Translokationen
- Amplifikationen / Deletionen

Analysen auf DNA- und RNA-Ebene:
- Translokationen
- Amplifikationen / Deletionen
- Mutationen

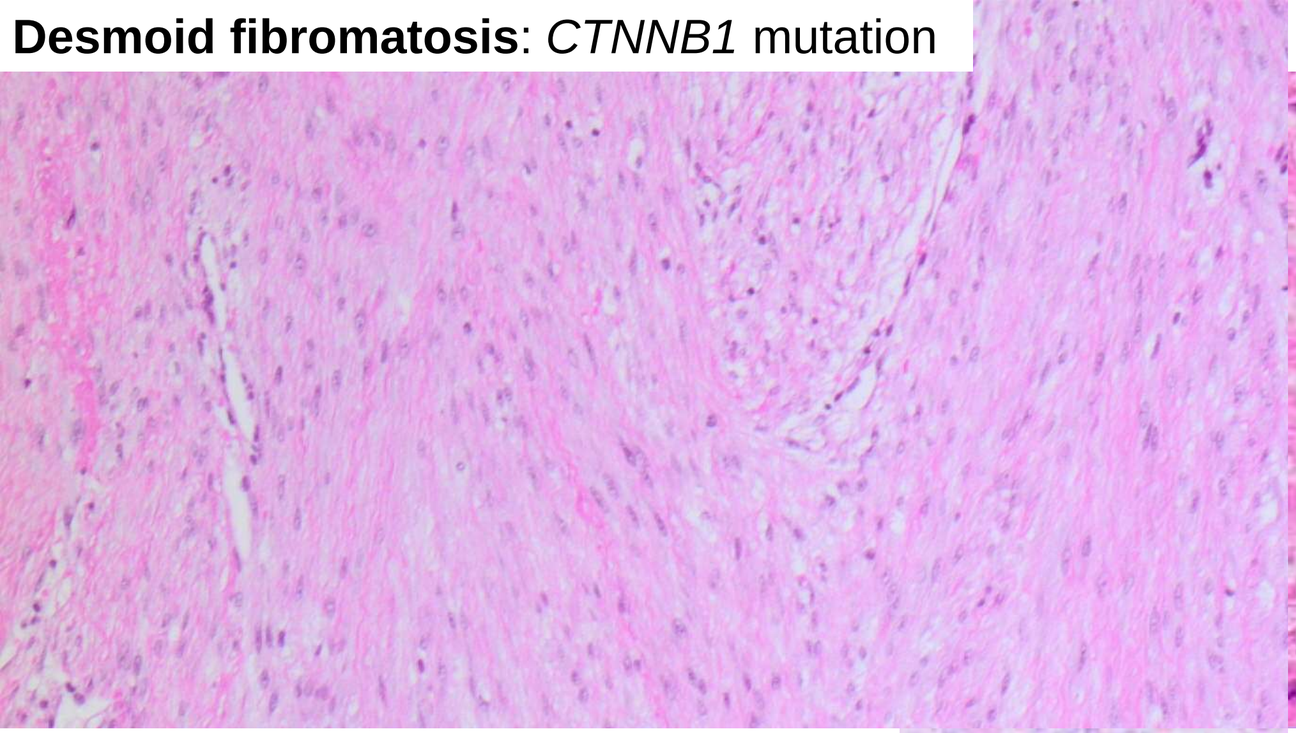

CTNNB1 mutation


FUS rearrangement

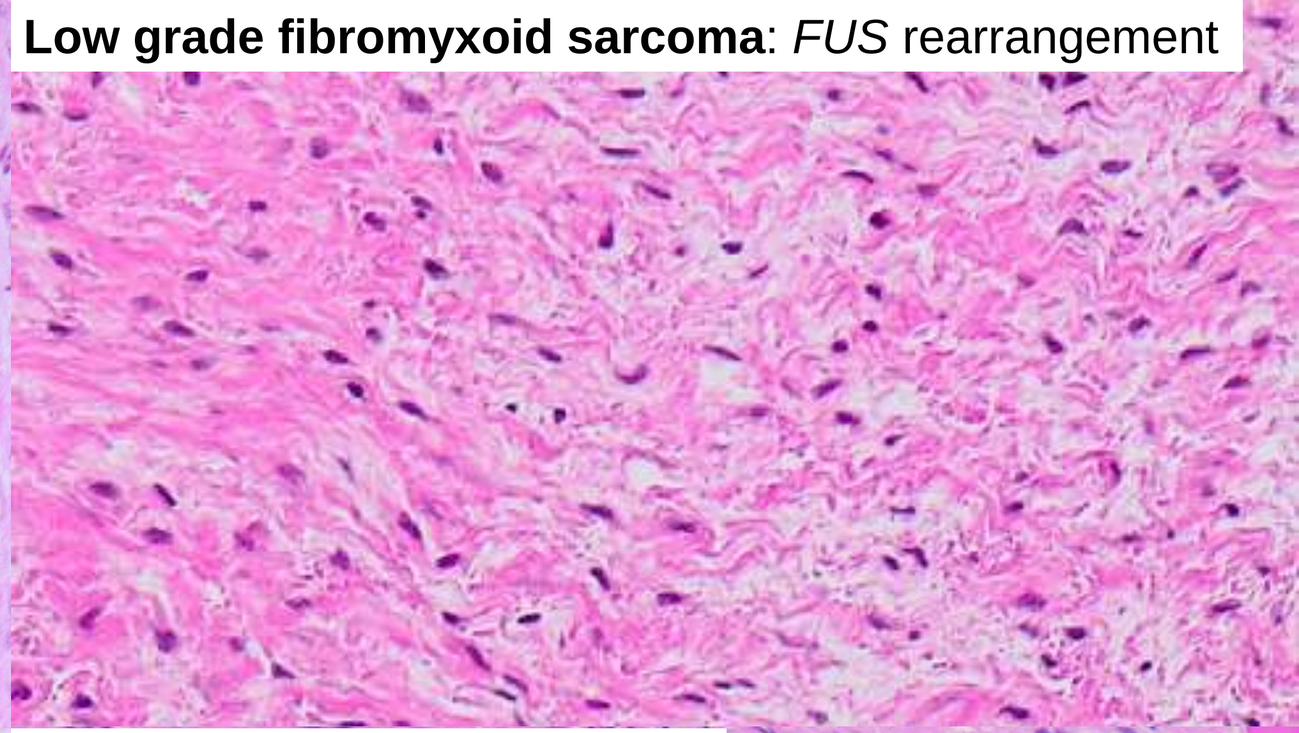

SYT rearrangement


COL1A1 rearrangement

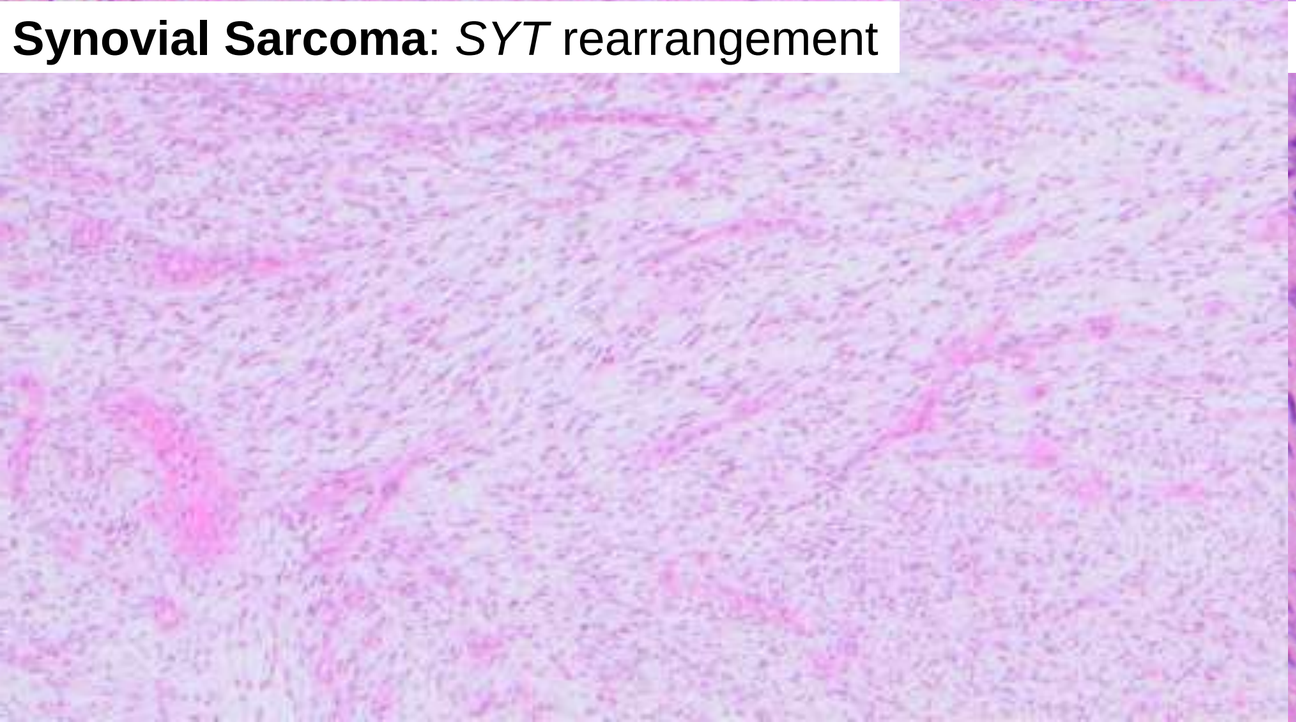
Desmoid fibromatosis: *CTNNB1* mutation



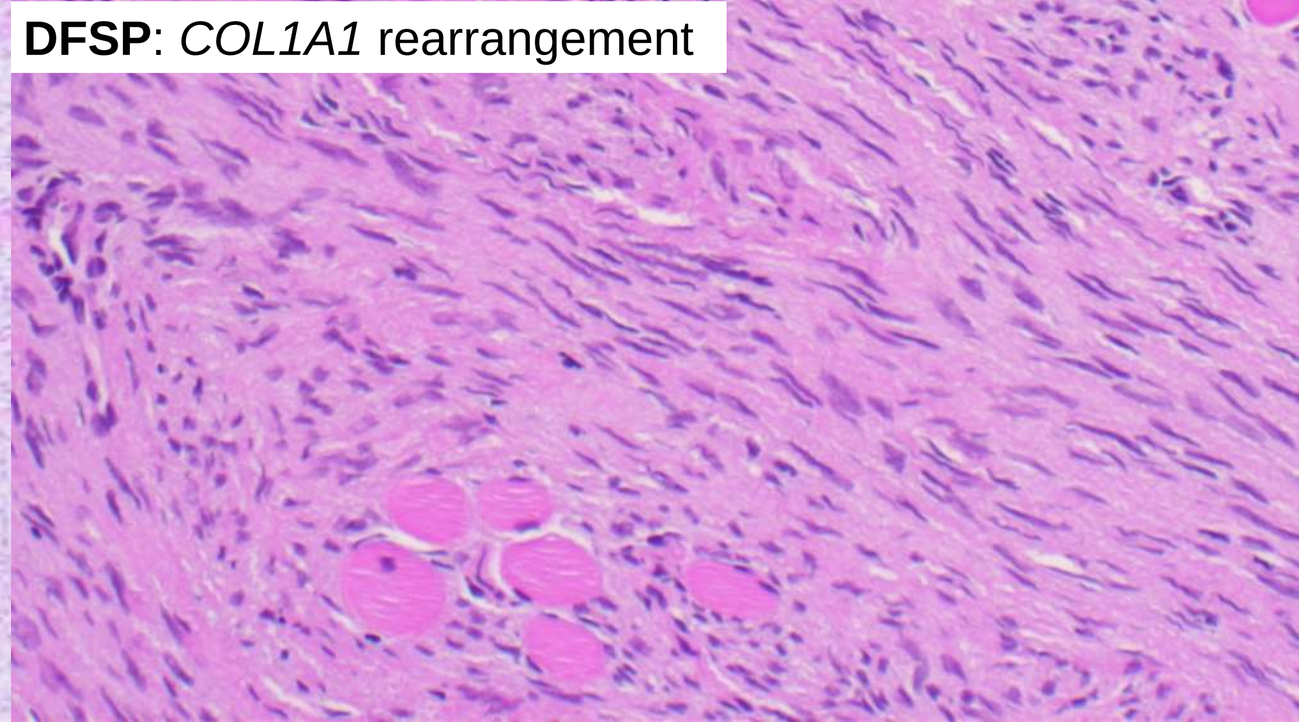
Low grade fibromyxoid sarcoma: *FUS* rearrangement



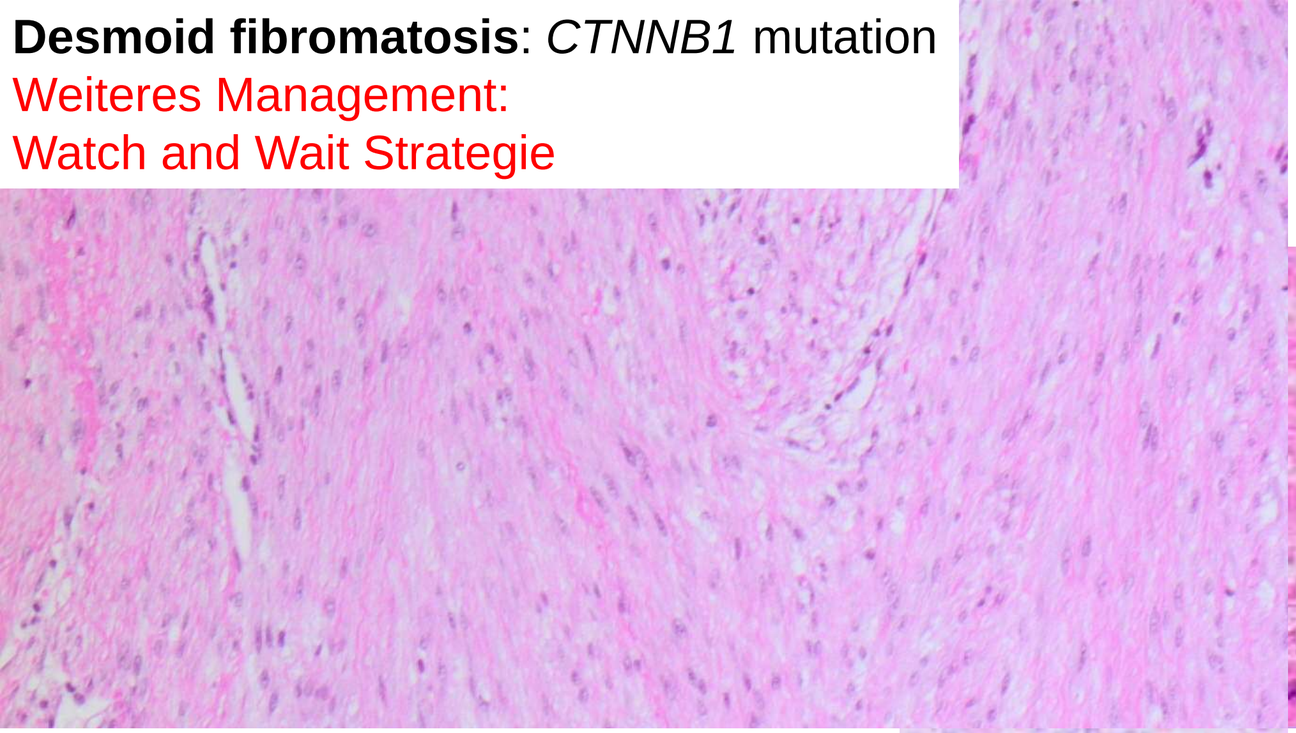
Synovial Sarcoma: *SYT* rearrangement



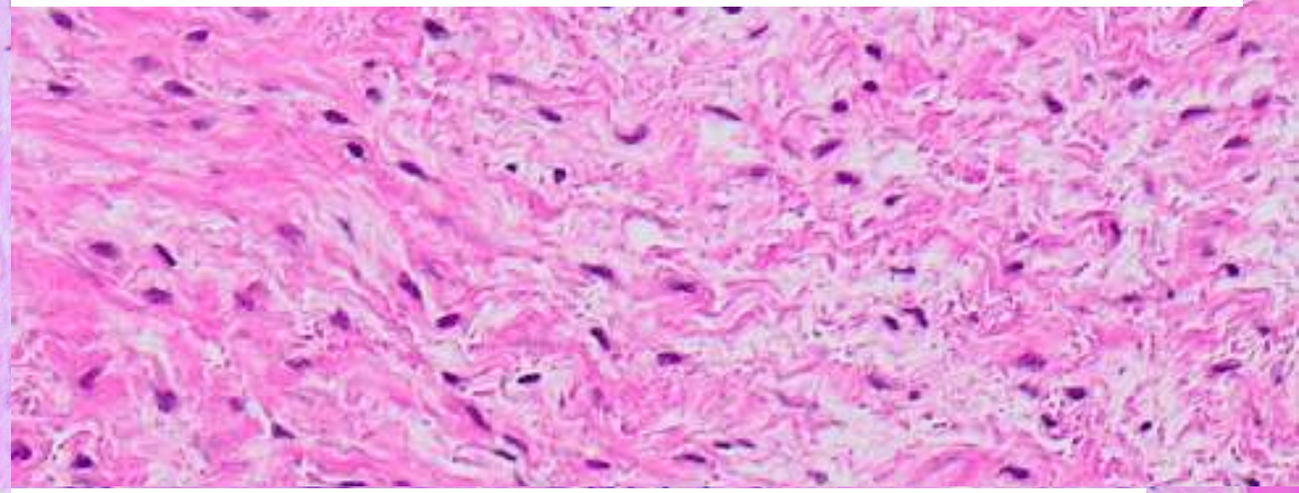
DFSP: *COL1A1* rearrangement



Desmoid fibromatosis: *CTNNB1* mutation
Weiteres Management:
Watch and Wait Strategie



Low grade fibromyxoid sarcoma: *FUS* rearrangement
Weiteres Management:
Staging und onkochirurgische Resektion
Langjähriges Followup



Synovial Sarcoma: *SYT* rearrangement
Weiteres Management:
Staging
Neoadjuvante (System-) Therapie
Onkochirurgische Resektion
Followup

DFSP: *COL1A1* rearrangement
Weiteres Management:
Onkochirurgische Resektion
Zielgerichtete Therapie (Tyrosinkinase-Inhibitoren)?

Zusammenfassung

1. Weichteiltumoren sind komplex, Sarkome selten

2. **Molekularpathologische Diagnostik** wichtig für die abschliessende Diagnose und das weitere Management

3. Repräsentative Biopsien und **Materialtriage** als Grundlage einer effizienten Diagnostik

4. **Interdisziplinäre Zusammenarbeit** (Onkologen, Chirurgen, Radiologen und Pathologen) als Basis für eine erfolgreiche Behandlung von Weichteiltumoren