Rätselecke 29.04.2023
Fall 3

Björn Sander

Medizinische Hochschule Hannover

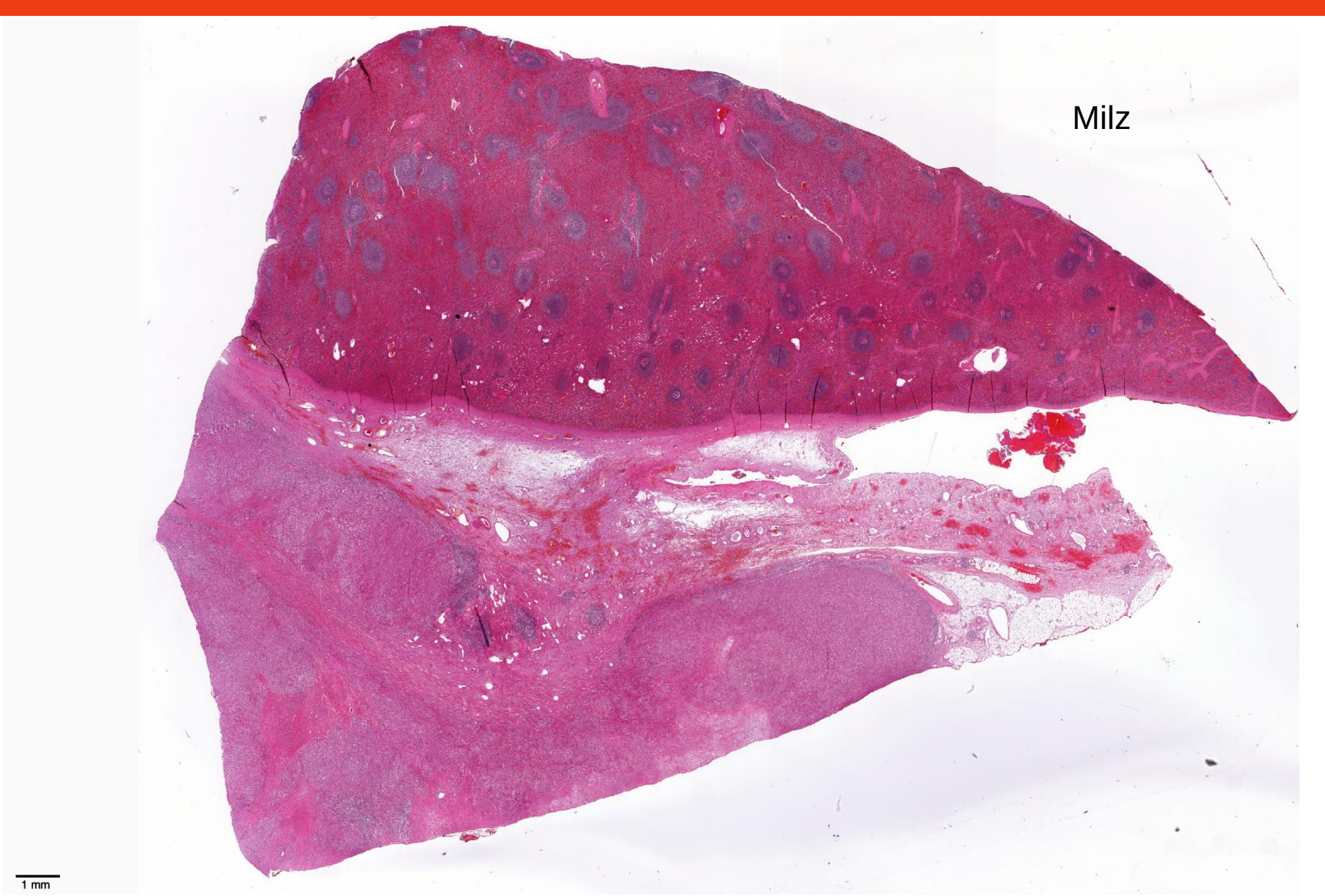
Anamnese

- **52 j. Patientin**
- Adipositas
- Z. n. Hysterektomie und Appendektomie
- Mutter: Colon-Ca. mit 39 J. Onkel mütterlicherseits: Colon-Ca. und weitere Neoplasien in jungem Alter
- Rehabilitationsmed. Behandlung bei Rückenschmerzen
- Bei Blutdruckspitzen **V. a. Phäochromozytom**
- Bildgebung: **abdomineller Tumor im linken Oberbauch**, Endokrinologie ohne wegweisenden Befund
- Bioptisch maligne Spindelzellneoplasie unklarer Differenzierung
- **Operative Resektion** des Tumors

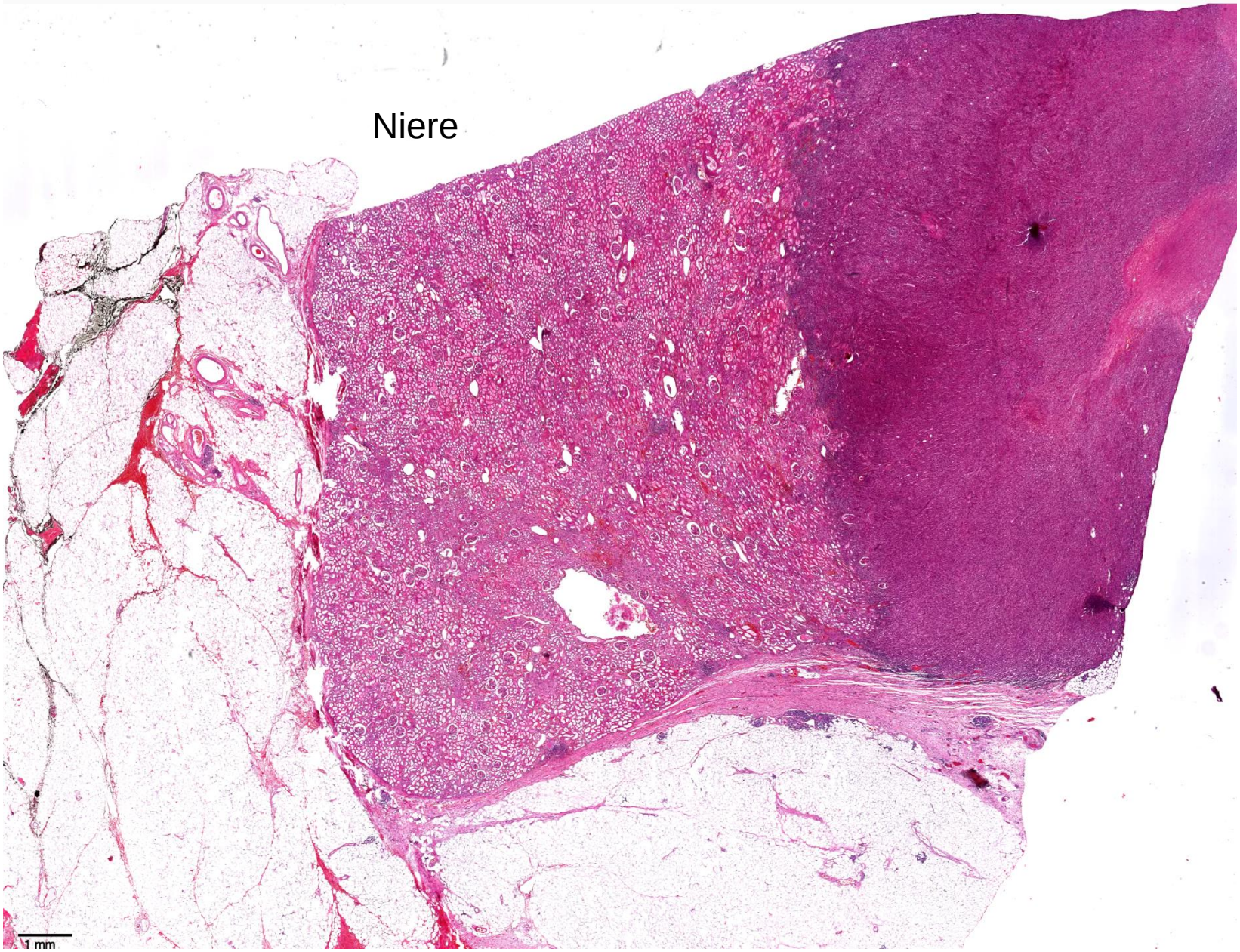
Makroskopie

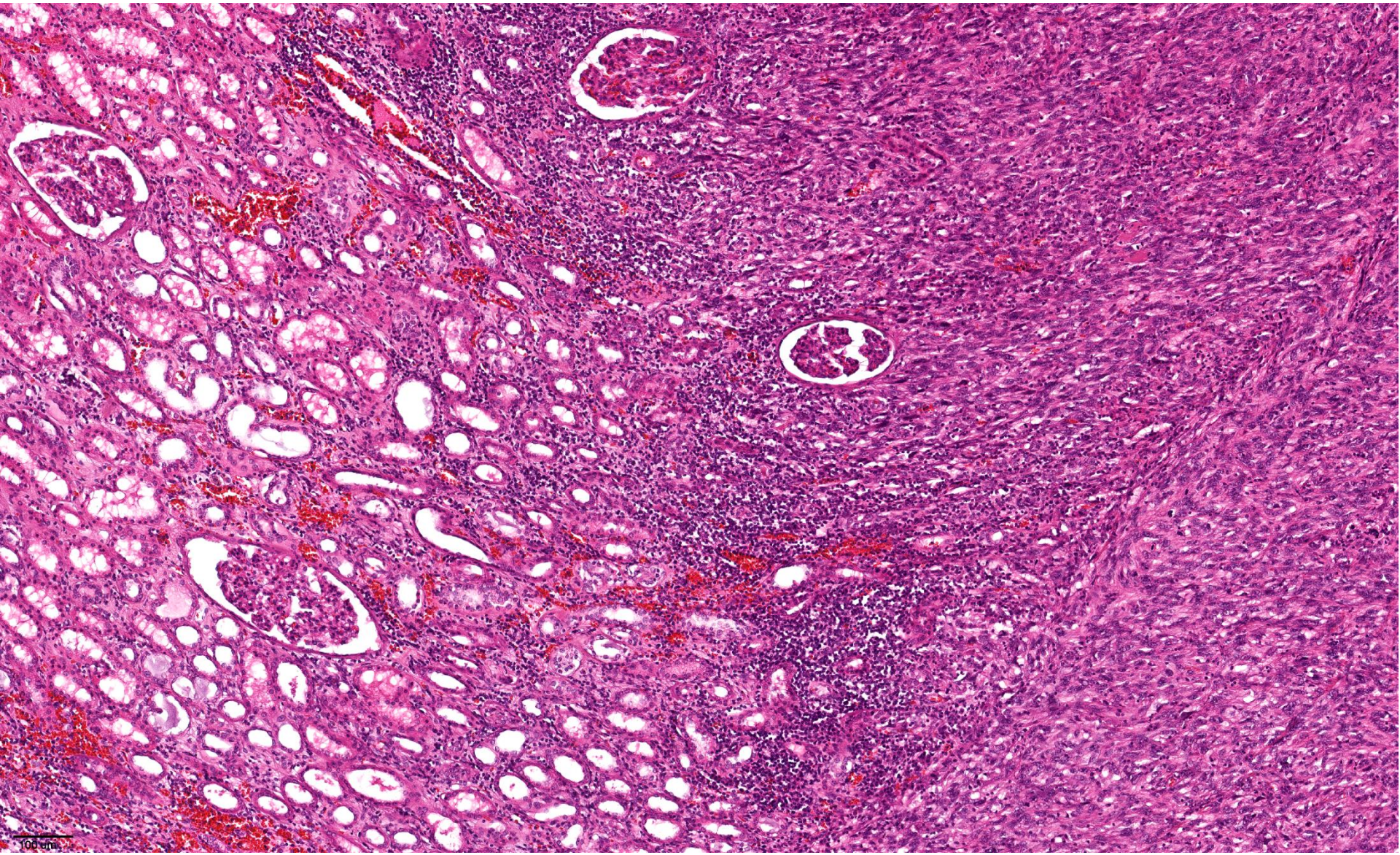
- En-bloc-Resektat Pankreasschwanz, Milz, linke Niere
- Maximal 14 cm durchmessender retroperitonealer Tumor zwischen Milz, linker Niere und Pankreasschwanz

Mikroskopie

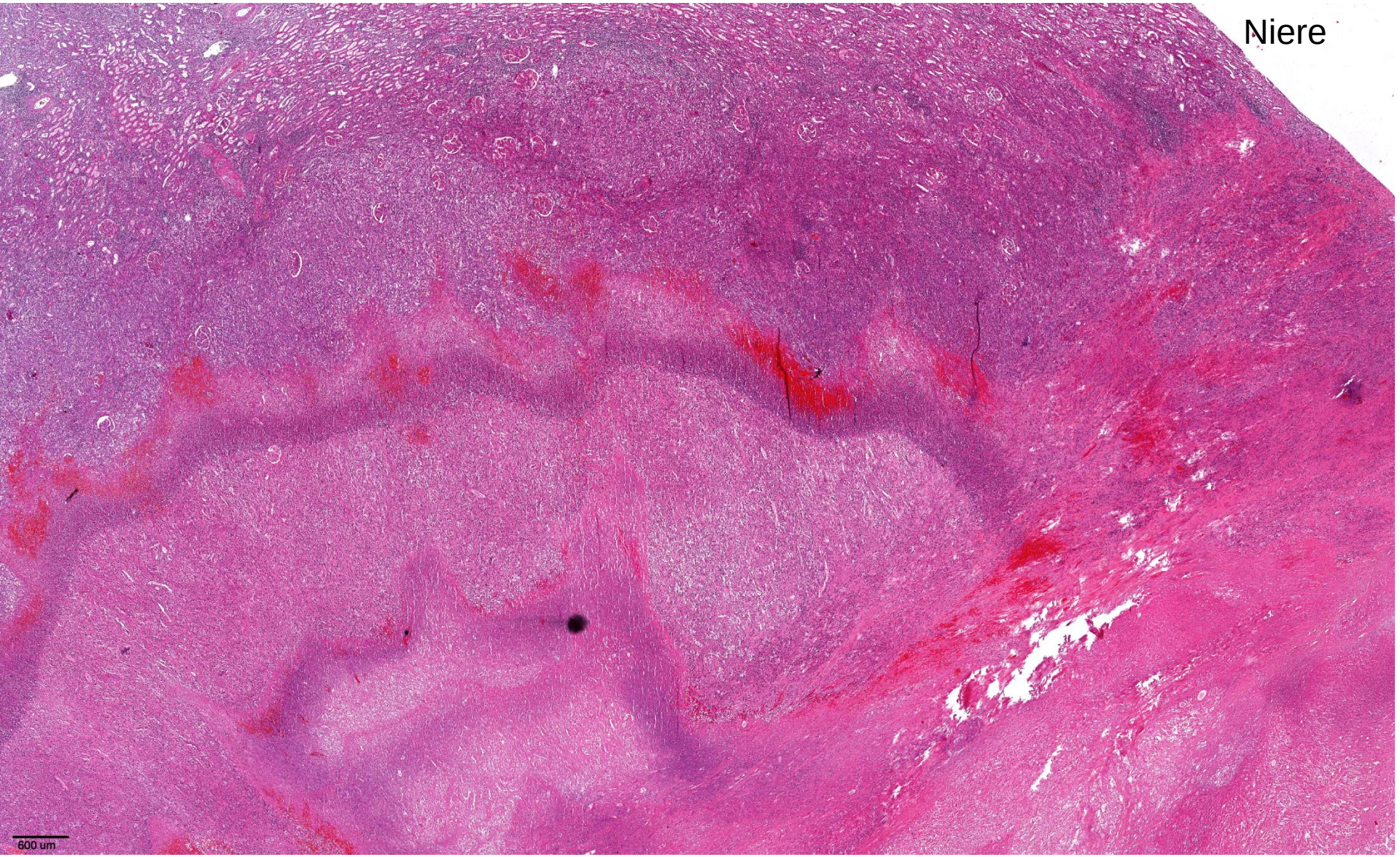

Milz
1 mm

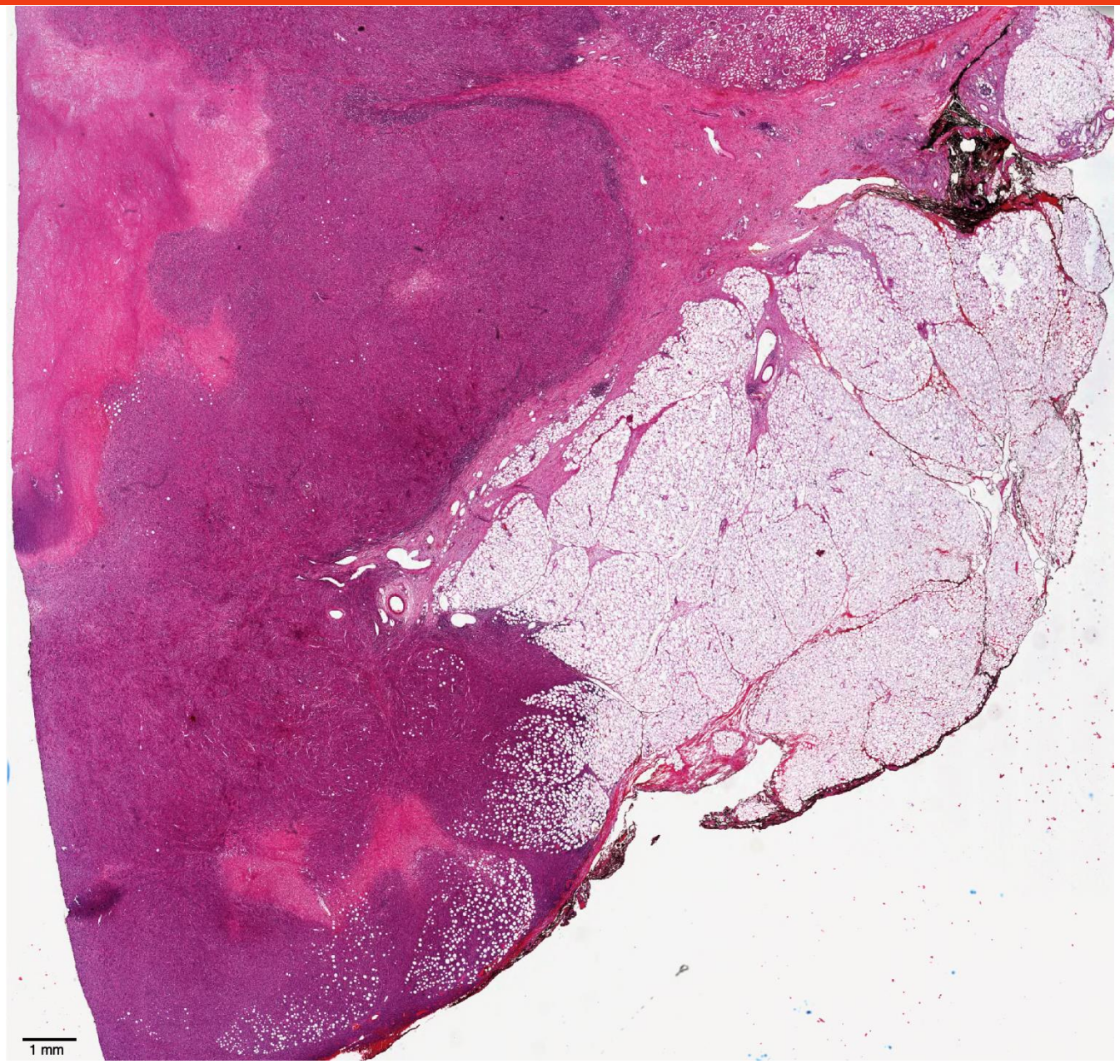
Niere




100 µm

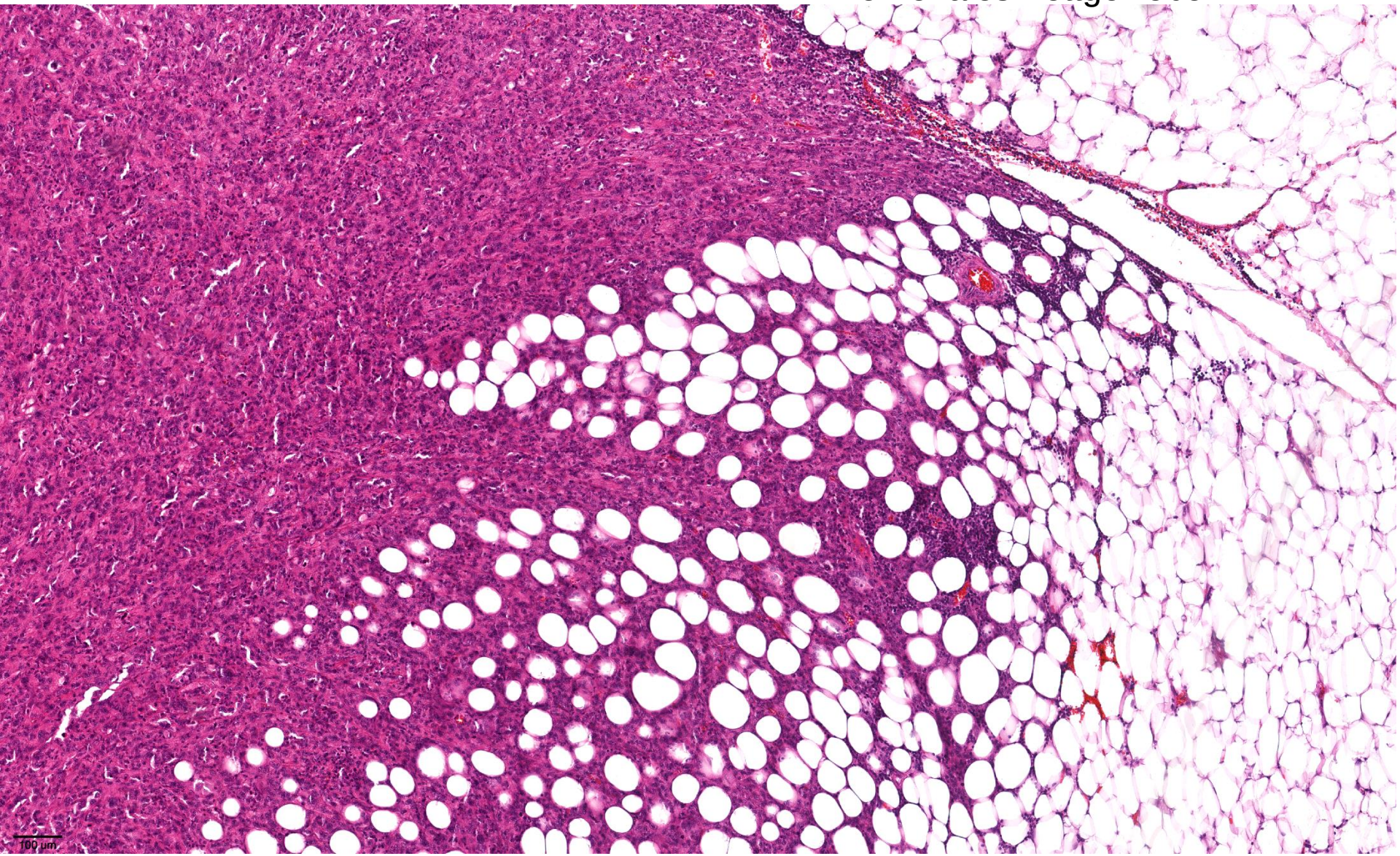
Niere



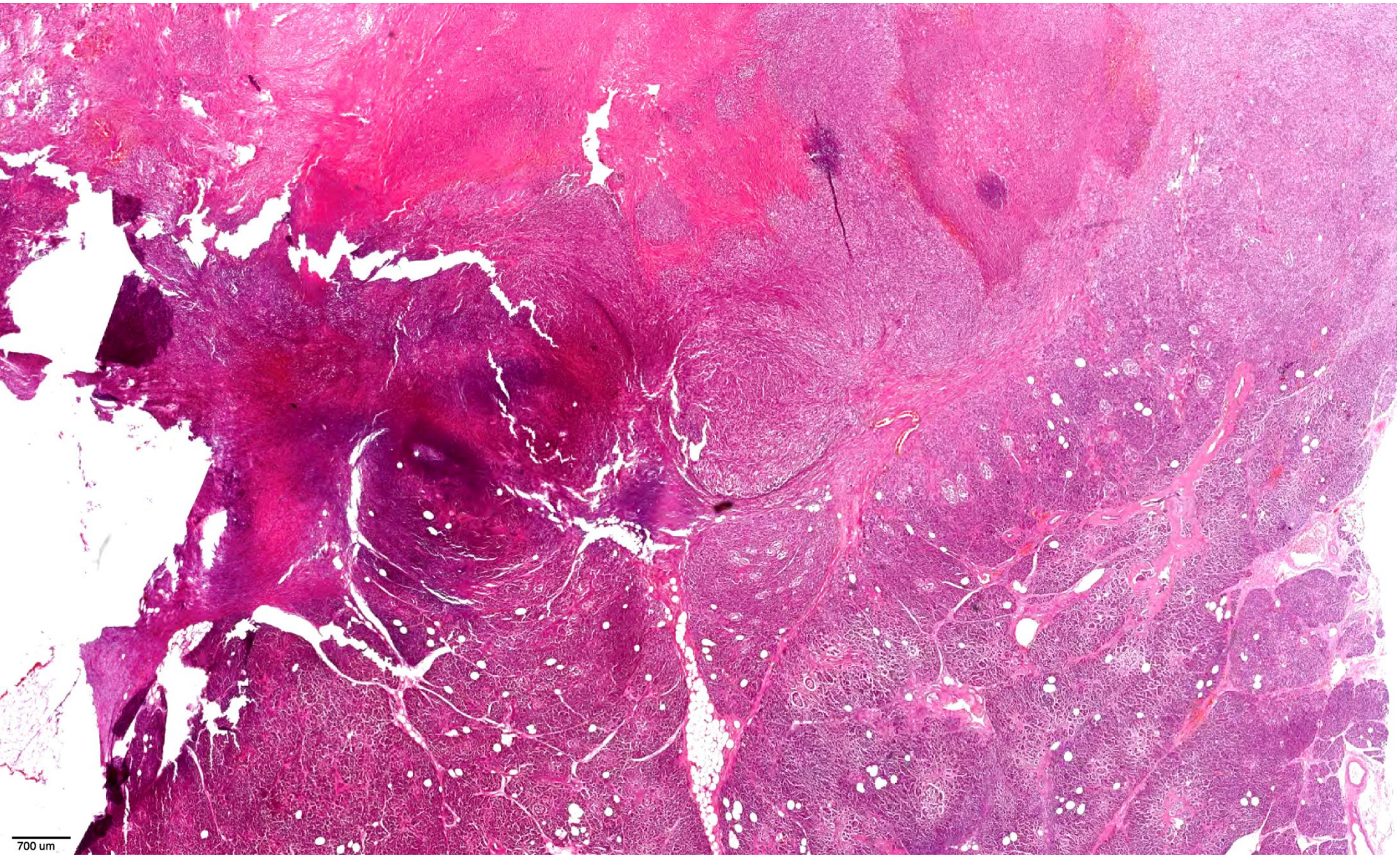


Niere

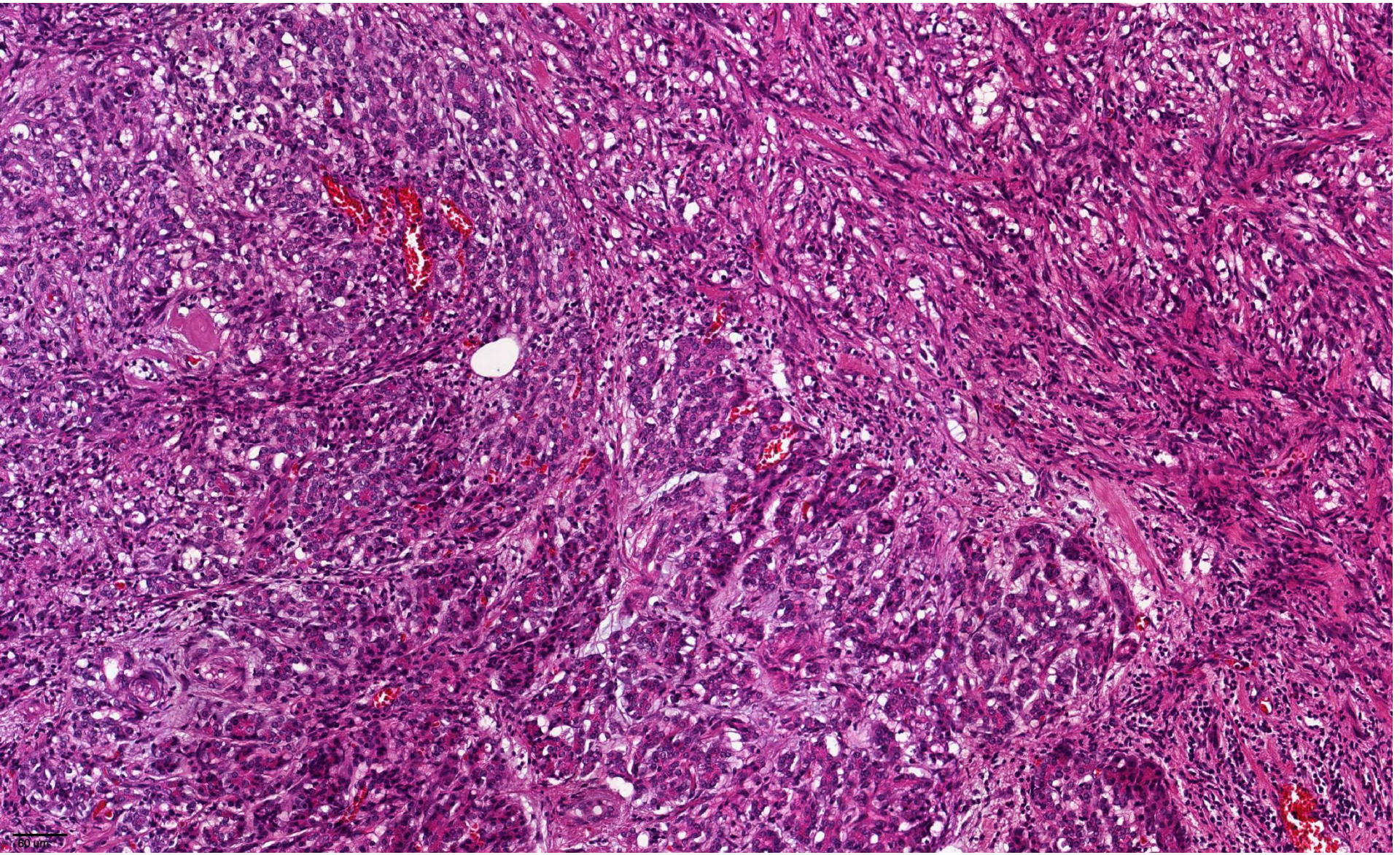
Perirenales Fettgewebe

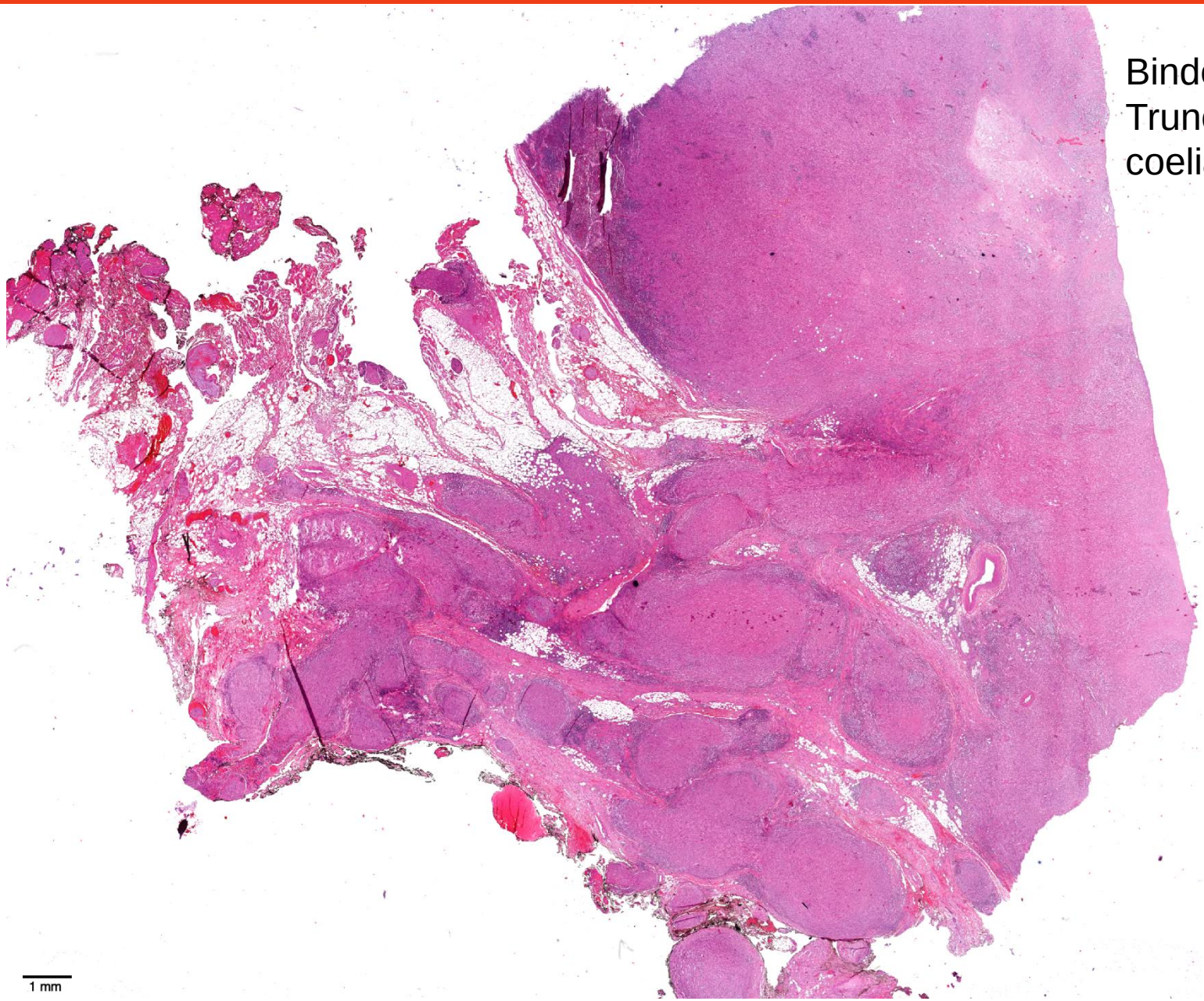


Pankreas

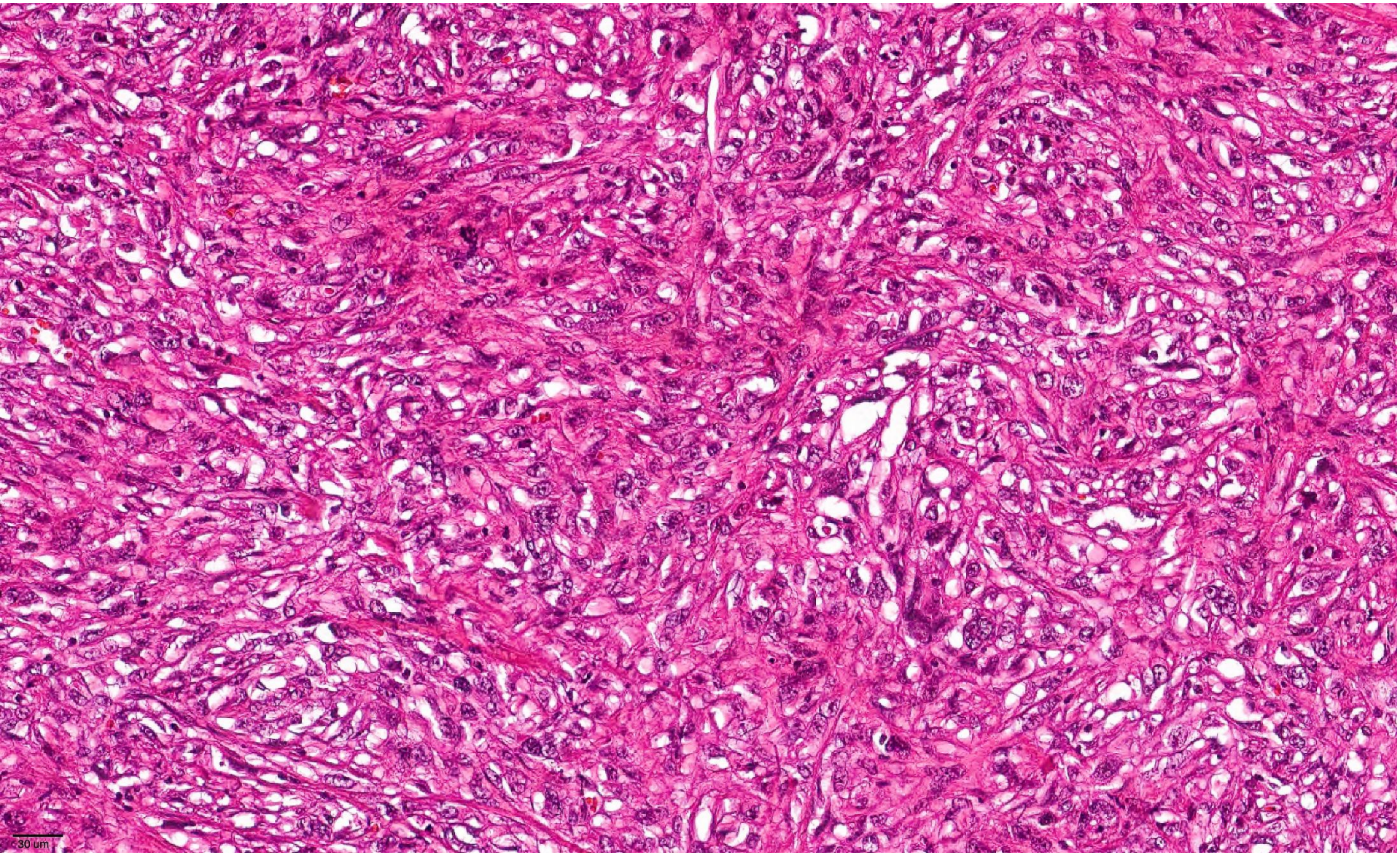


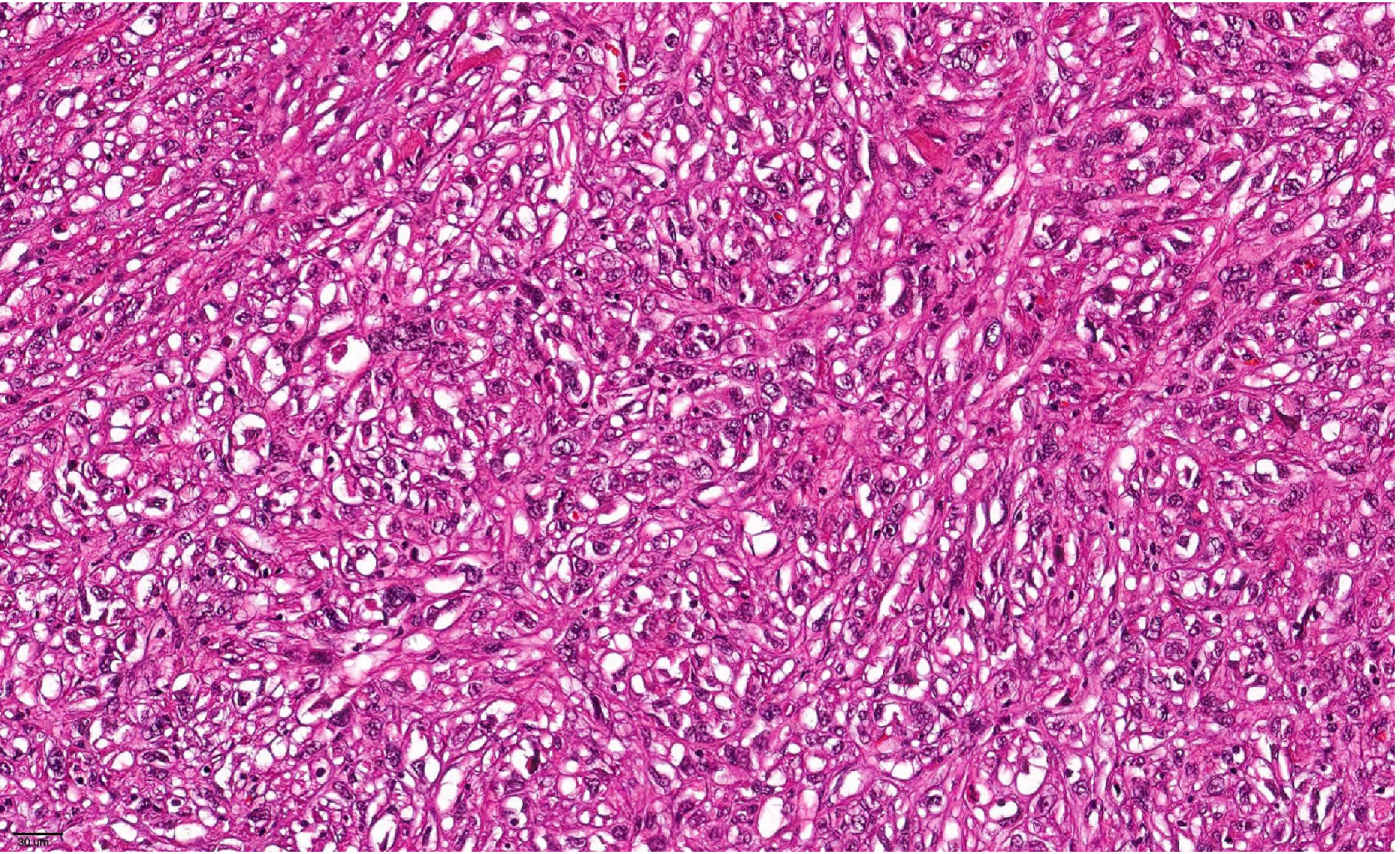
Pankreas





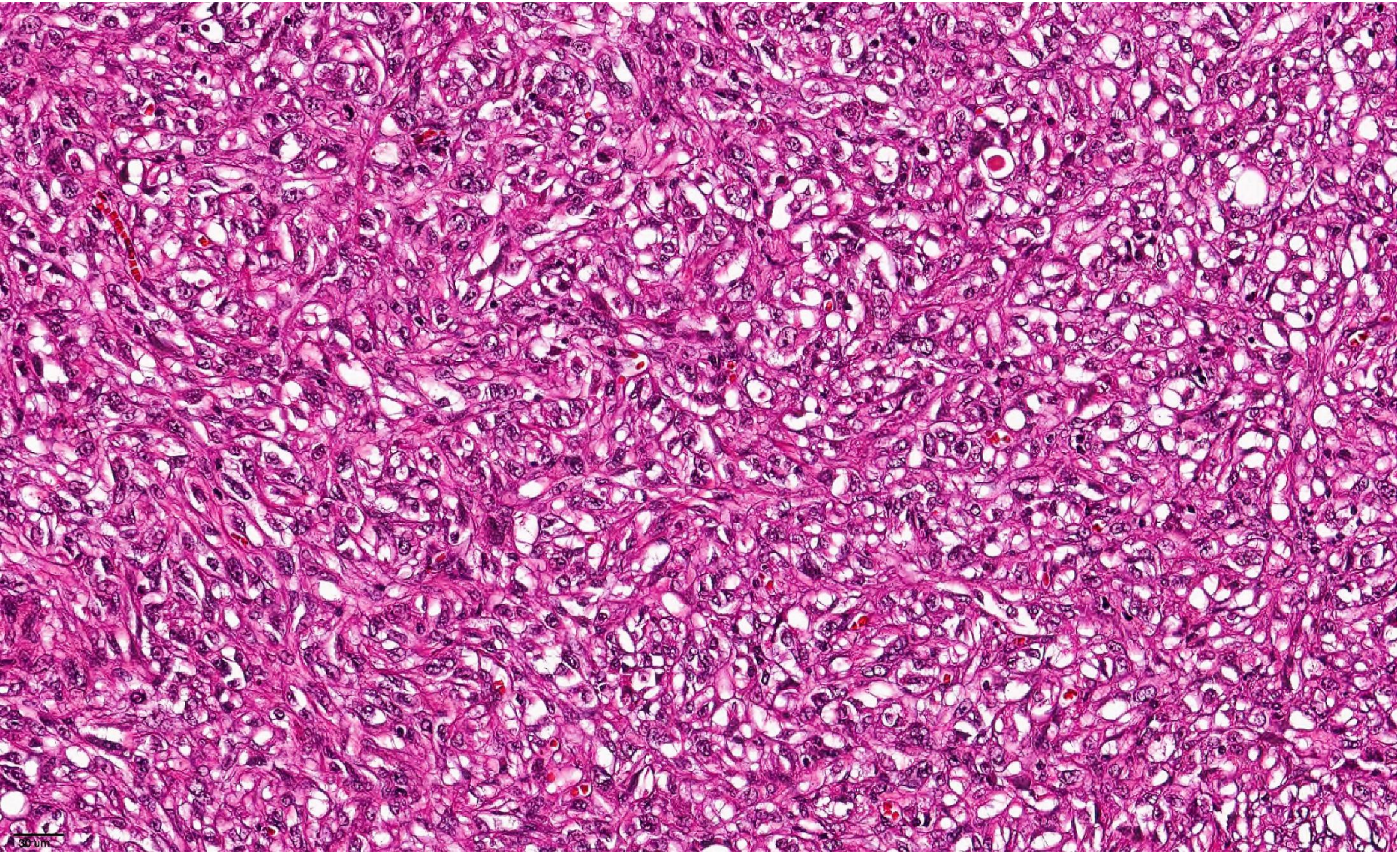
Bindegewebe am Truncus coeliacus


30 um


30 µm

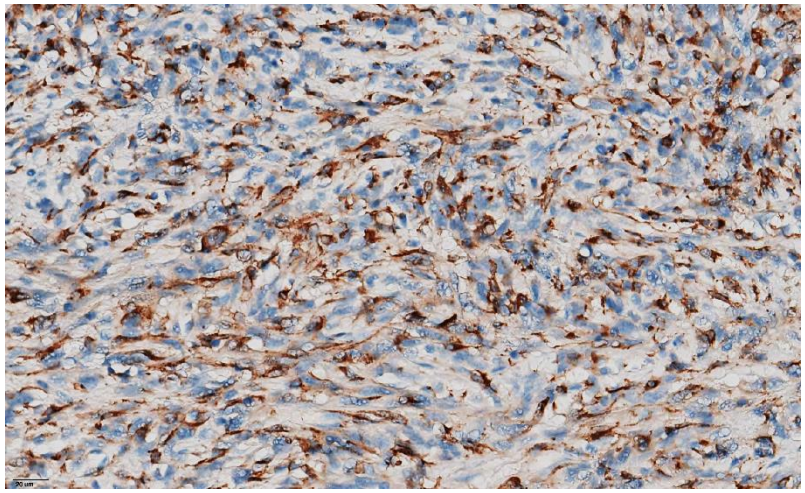
Immunhistochemie

- CK8/18, CK5/14, CK7, CK20, p40, GATA3, Uroplakin III negativ
- PAX8 negativ
- DOG1, CD117, CD34 negativ
- INSM1 negativ
- Melan A, SOX10, S100 negativ
- ERG negativ
- CD20, CD3, CD30, ALK, TdT, MPO, CD123 negativ
- MDM2-FISH negativ

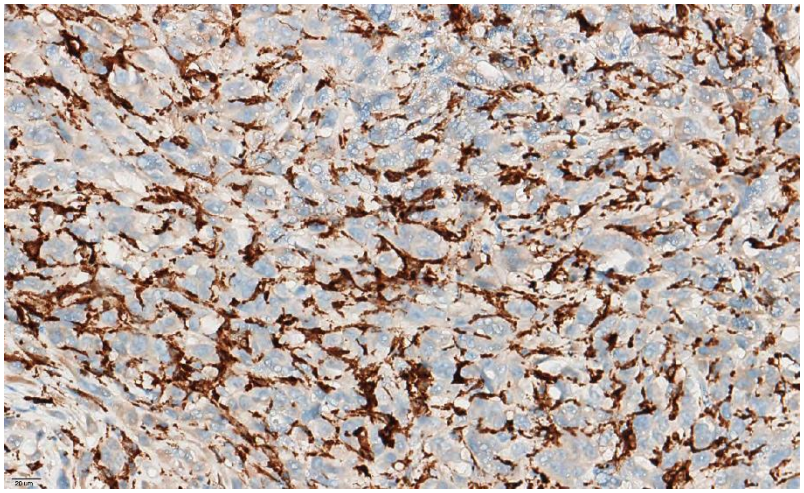

30 µm

Immunhistochemie

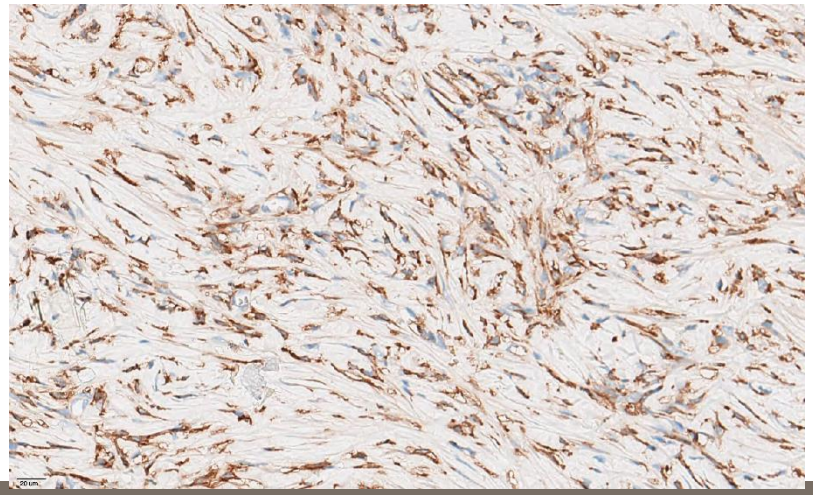
CD68 (PGM1)



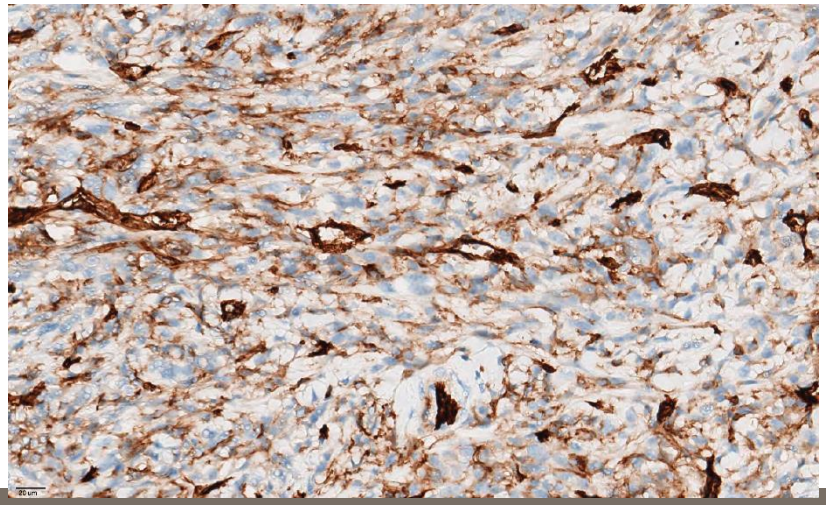
CD163



CD14



CD31



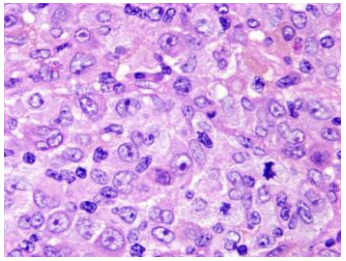
Histiozytäres Sarkom – Differentialdiagnosen

Histiozytäres Sarkom

CD68+, CD163+, CD14+, CD31+, Lysozym+, CD4+, S100-

Langerhanszell-Histiozytose/Sarkom

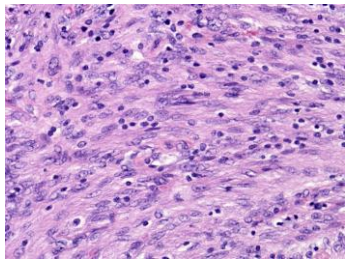
S100+, CD1a+, Langerin/CD207+



LHS
WHO 5. ed.

FDC Sarkom

CD21+, CD23+, CD35+



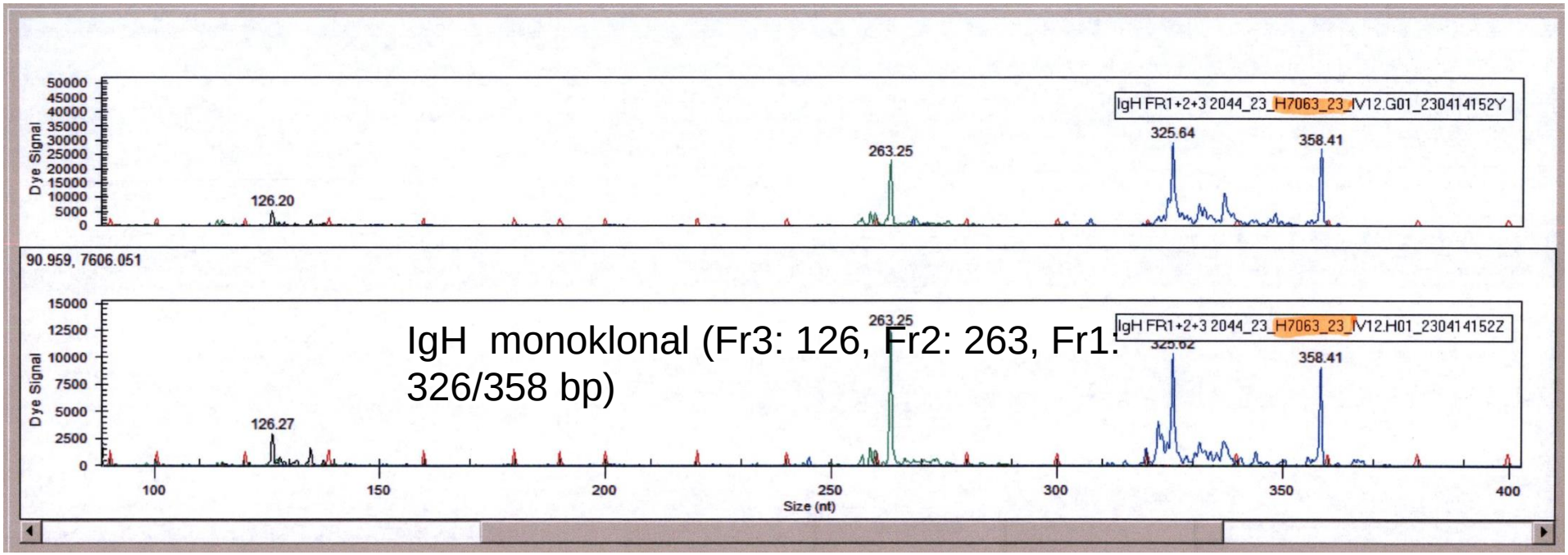
FDC-Sarkom
WHO 5. ed.

IDC Sarkom

S100+, CD45+, CD4+, CD43+, CD21-, CD23-, CD35-, CD1a-, Langerin-

Andere Histiozytosen (Erdheim-Chester, ALK-positive Histiozytosen)
(systemisch, ALK-Testung)

IgH Klonalitätsanalyse



IgH monoklonal (Fr3: 126, Fr2: 263, Fr1: 326/358 bp)

Assoziation mit indolentem B-Zell-Lymphom?

Klonale Rearrangements in soliden hämatologischen Tumoren

Huang et al. High frequency of clonal Ig and T-cell receptor gene rearrangements in histiocytic and dendritic cell neoplasms. Oncotarget. 2016 Nov 29;7(48)

Langerhans-Zell-Histiozytosen (n=8):

ca. 75 % IgH und/oder IgKappa

FDC Sarkome (n=9):

ca. 78 % IgH und/oder IgKappa

Histiozytäre Sarkome (n=3):

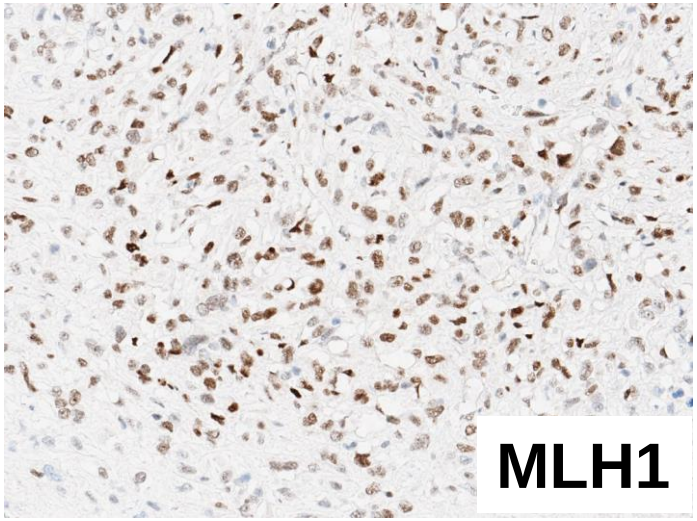
ca. 33 % IgKappa

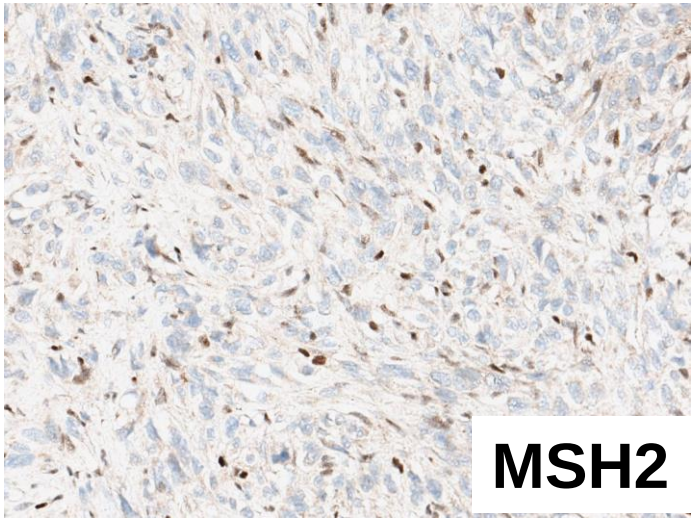
Sollte eine Sequenzierung bei histiozytären Sarkomen erfolgen?

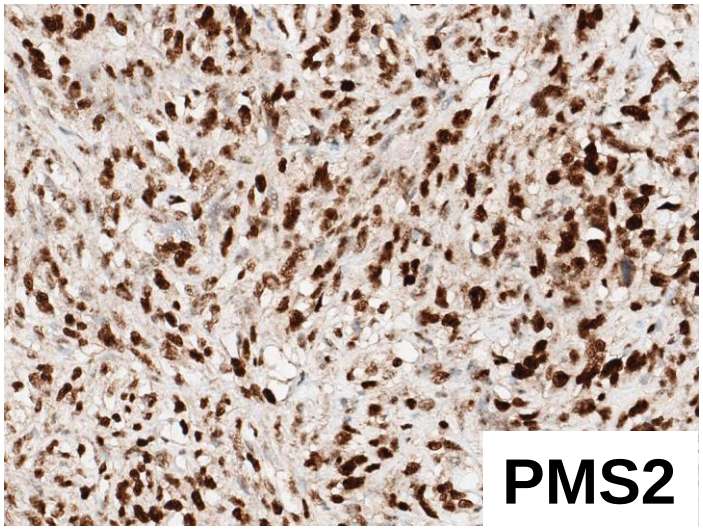
- Hauptsächlich MAPK-Gene betroffen (*KRAS*, *NRAS*, *BRAF*, *NF1* etc.)
- Zudem *PI3K/AKT/mTOR*-Weg
- *CDKN2A/B*-Deletionen

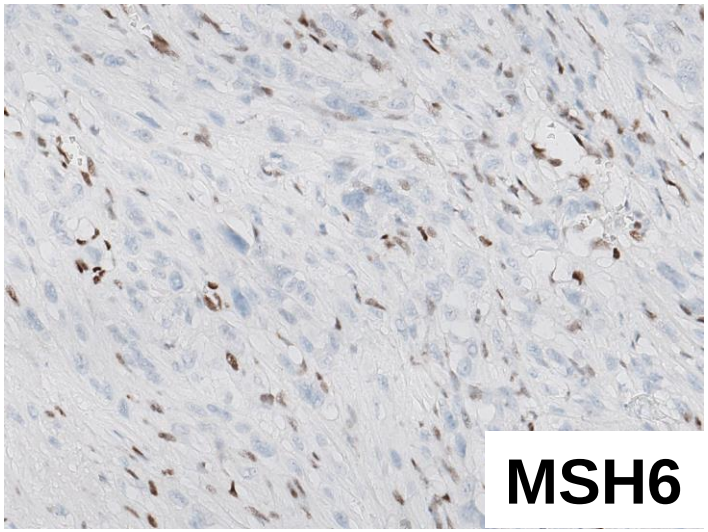
Egan C, Lack J, Skarshaug S, Pham TA, Abdullaev Z, Xi L, Pack S, Pittaluga S, Jaffe ES, Raffeld M. The mutational landscape of histiocytic sarcoma associated with lymphoid malignancy. Mod Pathol. 2021 Feb;34(2):336

Mikrosatellitenanalyse









BAT-25
BAT-26
NR-21
NR-24
NR-27
MSI, 3 von 5 Markern instabil

Diagnose

Mikrosatelliten-instabiles histiozytäres Sarkom mit Nachweis eines klonalen IgH-Rearrangements

Verdacht auf Lynch-Syndrom