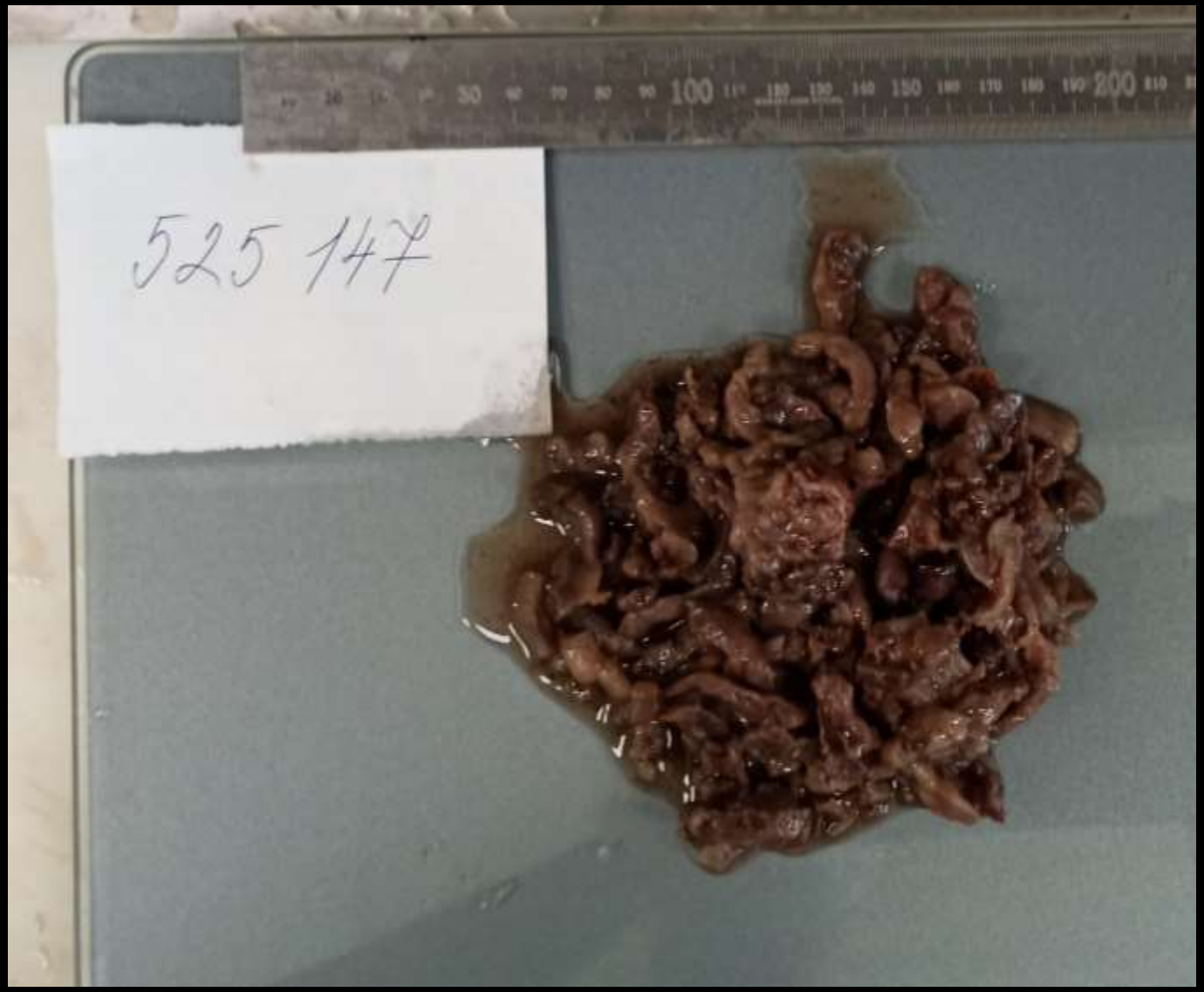
Prípad SD-IAP 768

B. Rychlý, Unilabs Bratislava

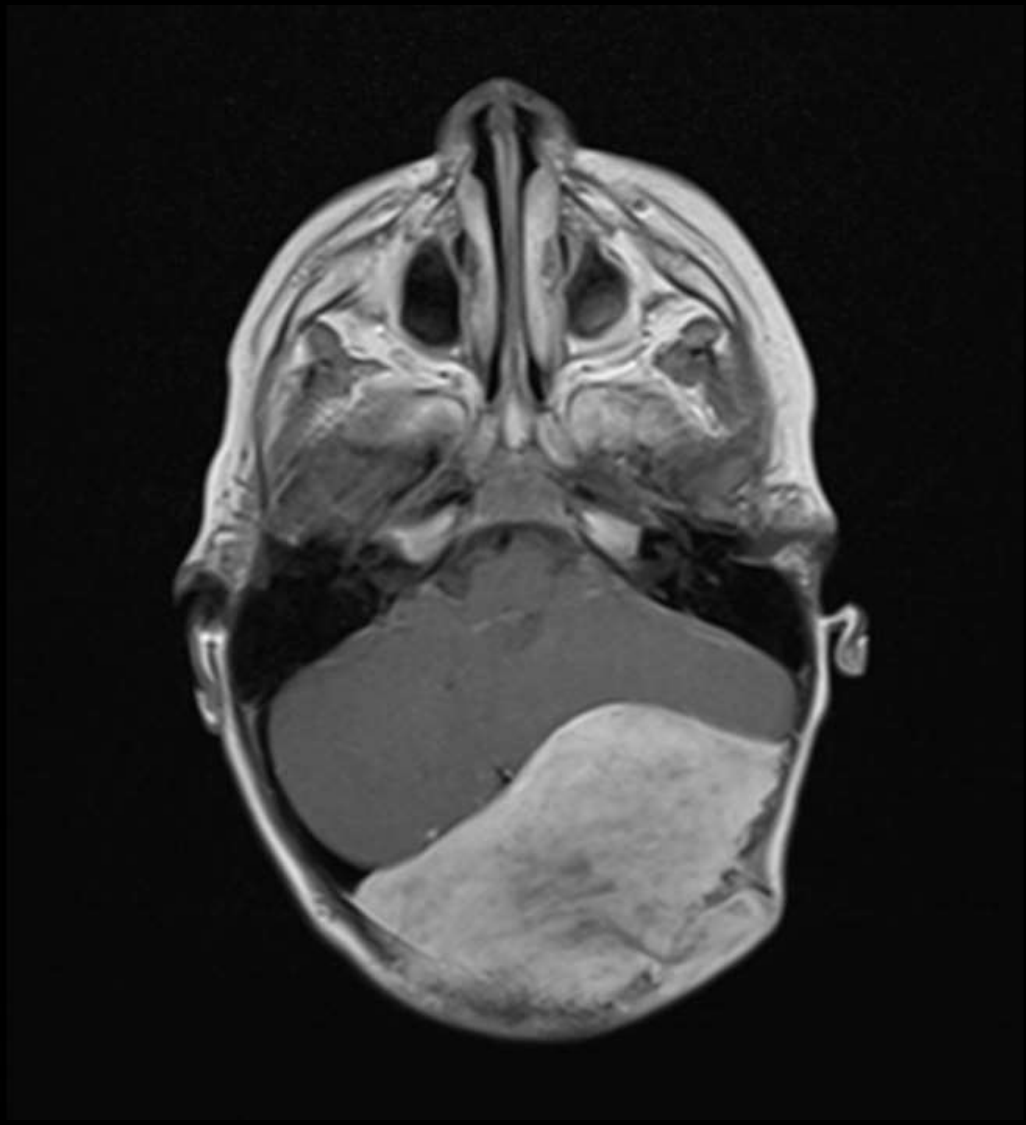
Klinické údaje

- 3-ročný chlapec s bolesťami hlavy
- mäkký nádor v podkoží záhlavia na ľavej strane
- zobrazovacie metódy ukázali obrovskú intrakraniálnu extraaxiálnu léziu v zadnej jame s herniáciou procesu cez defekt kalvy do podkožia záhlavia
- rádiológ suponuje epidermoidnú cystu (CT), prípadne meningeálny SFT/HPC (MR)
- radikálna operácia
- *anamnesticky je u otca geneticky dokázaná FAP (tel.)*

Makro?



Makro CNS procesy je MR

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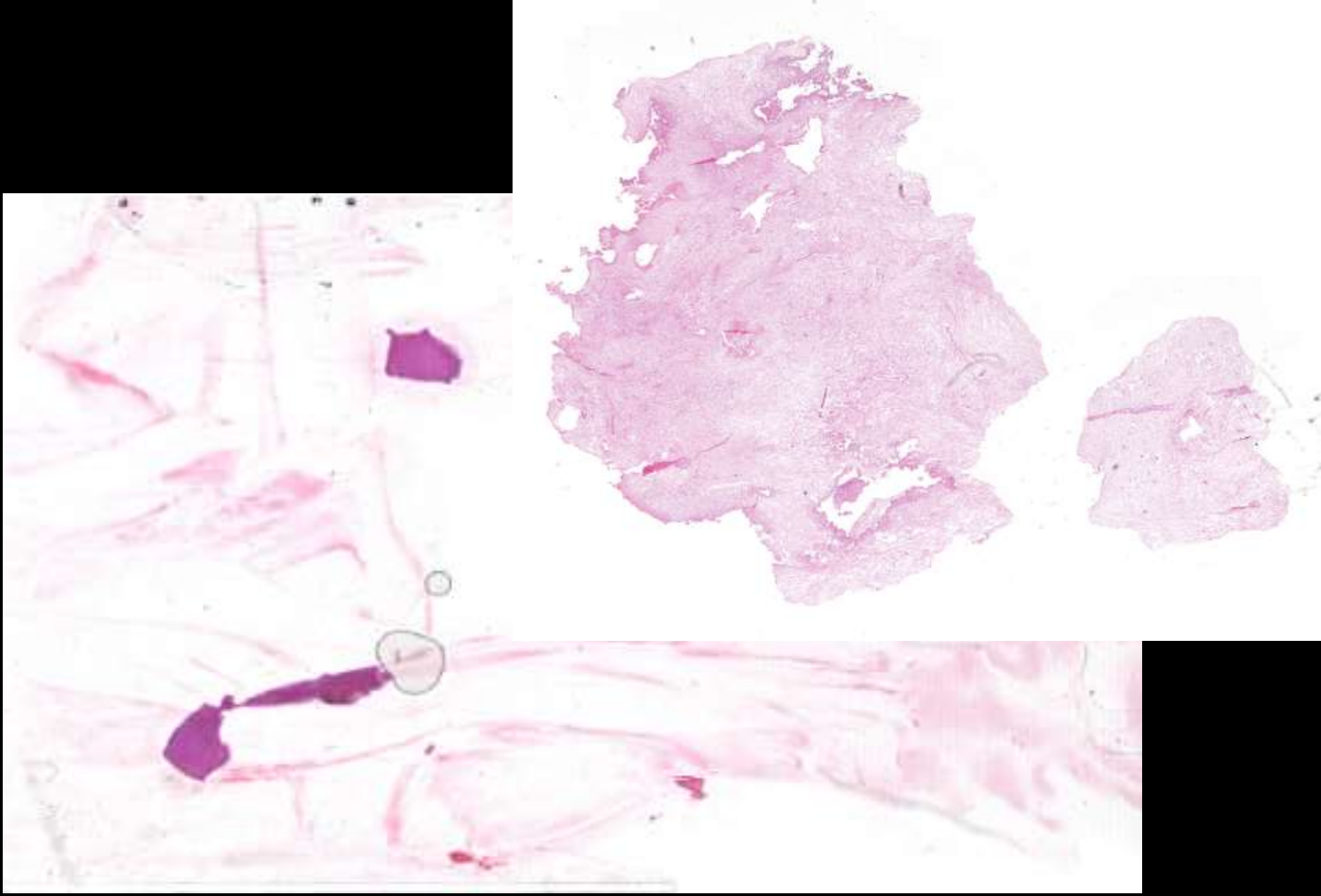
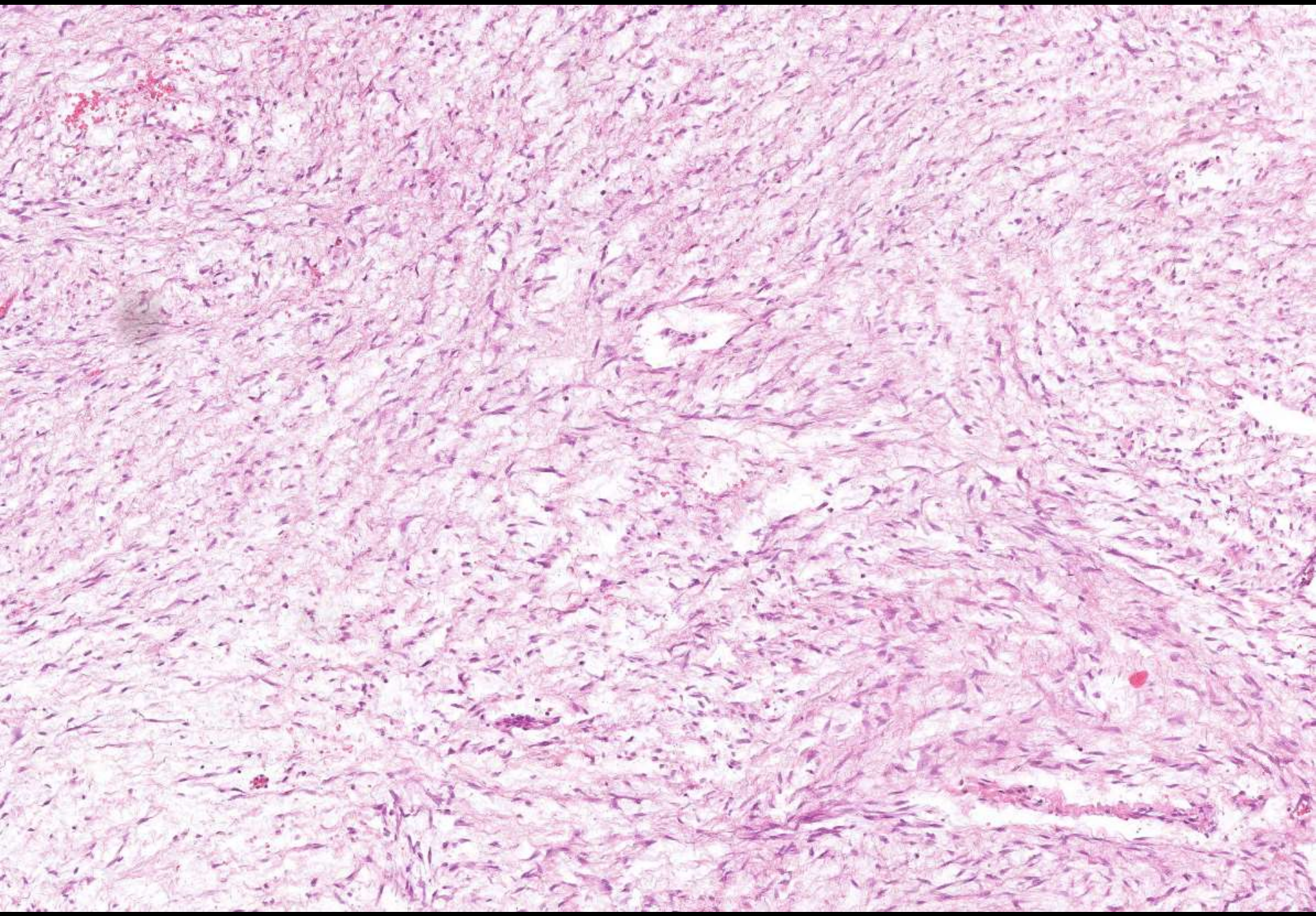
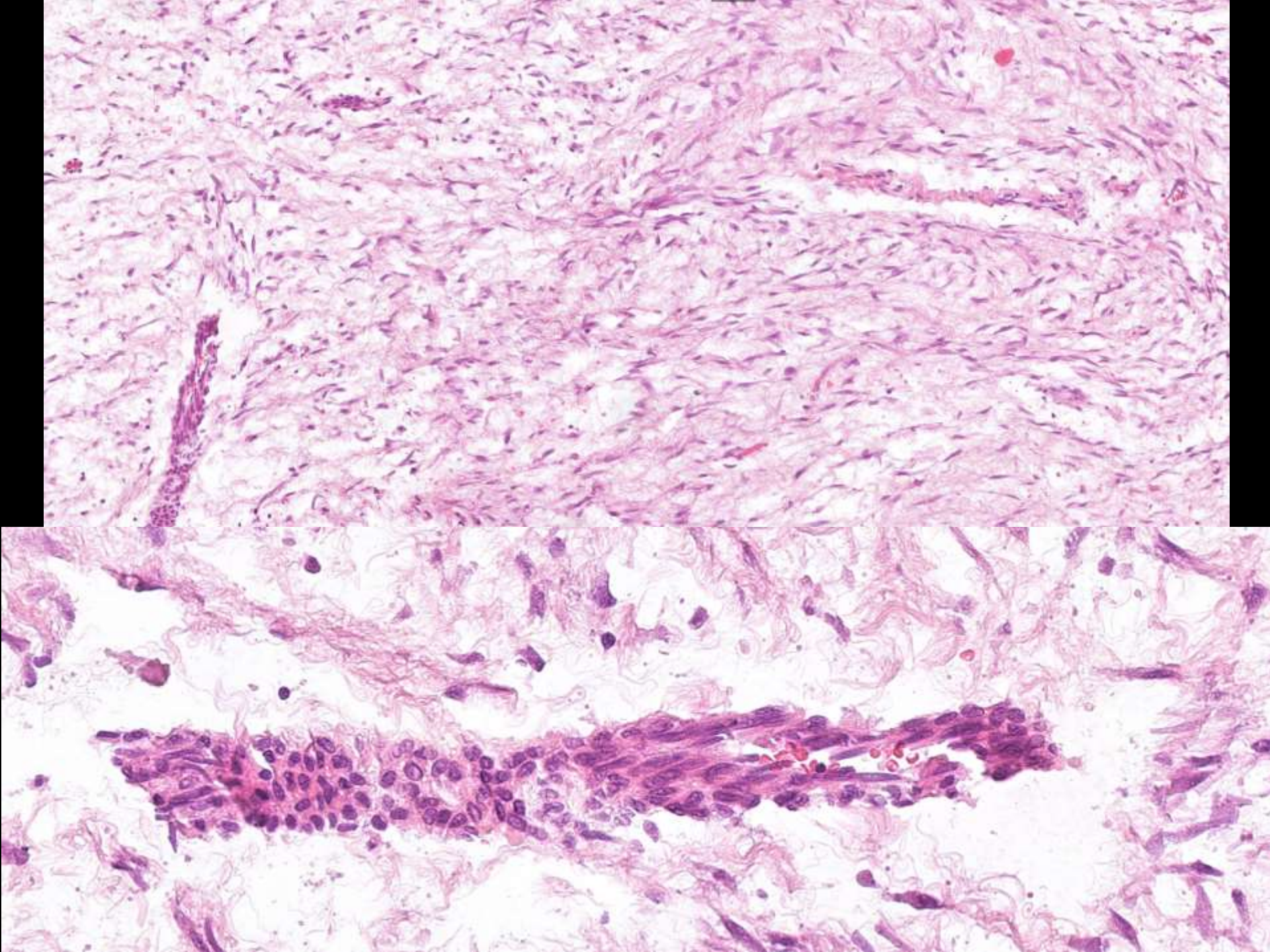
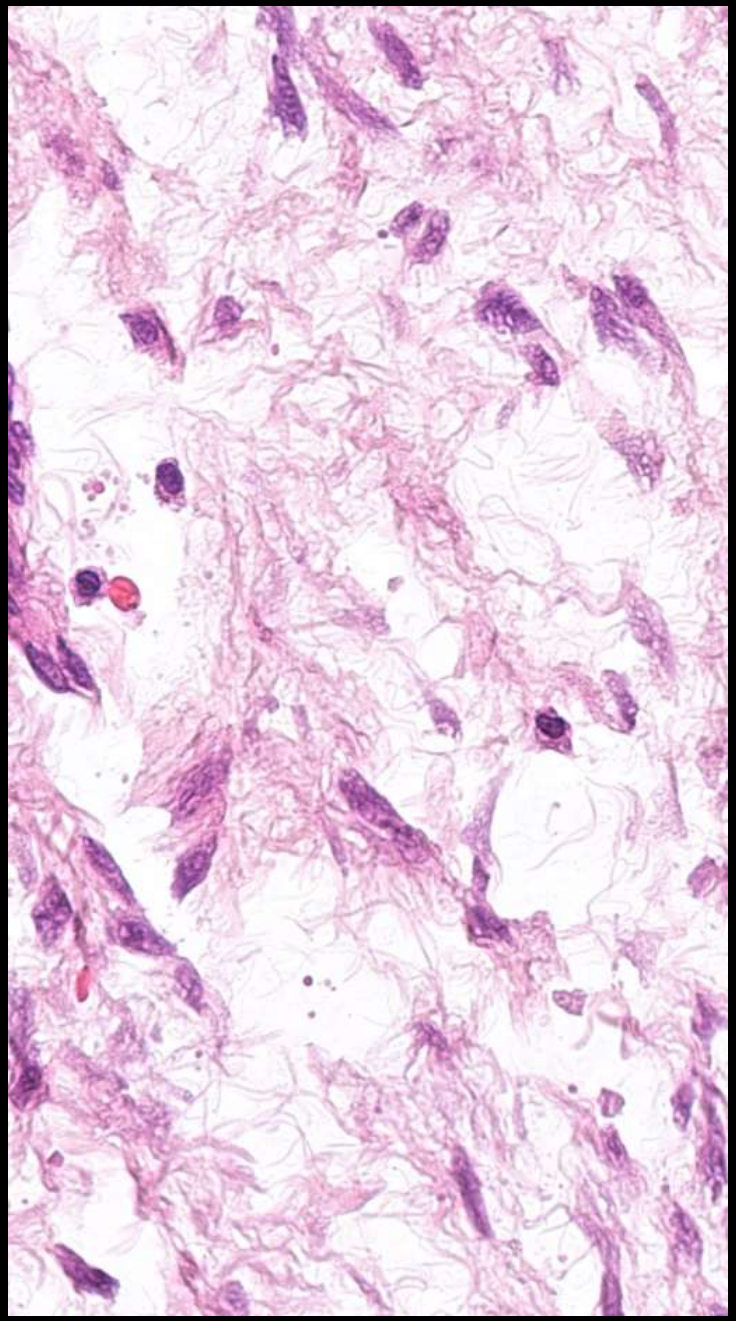
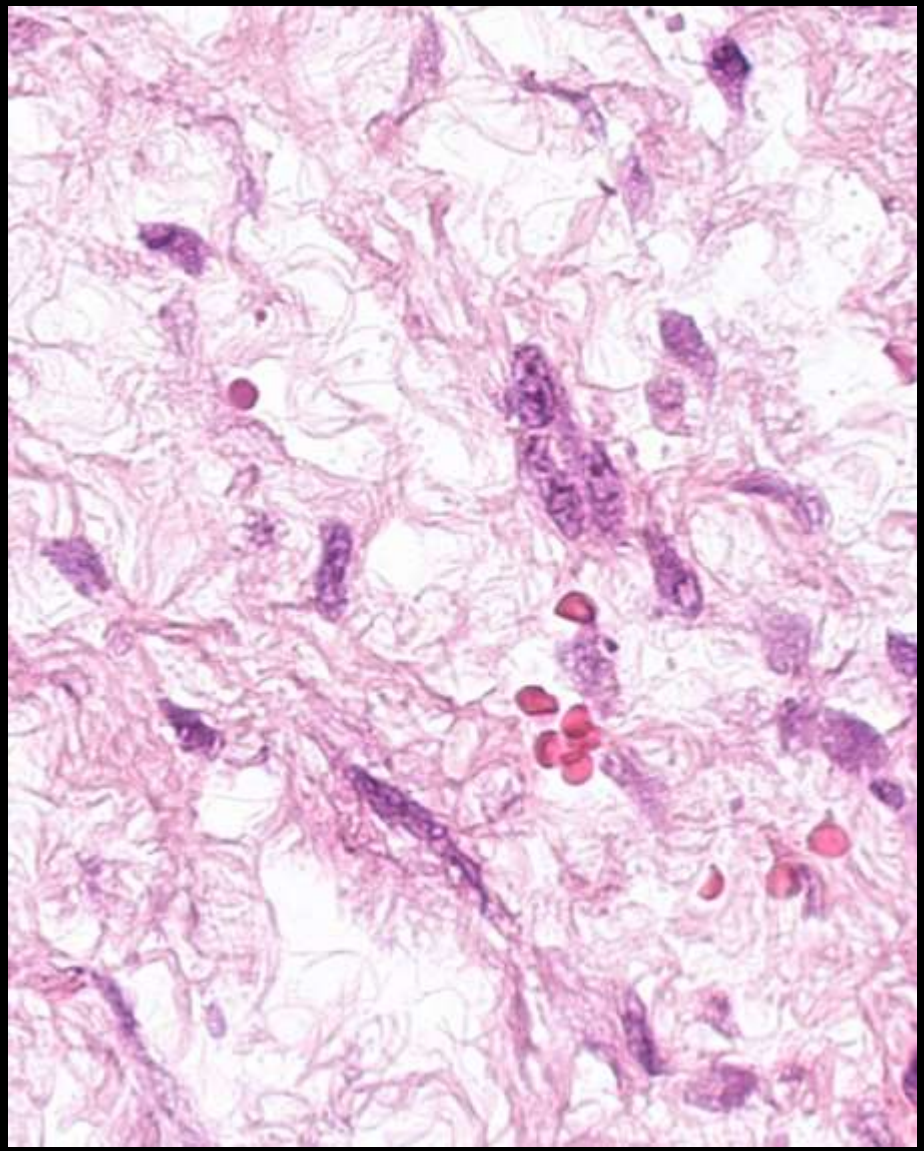


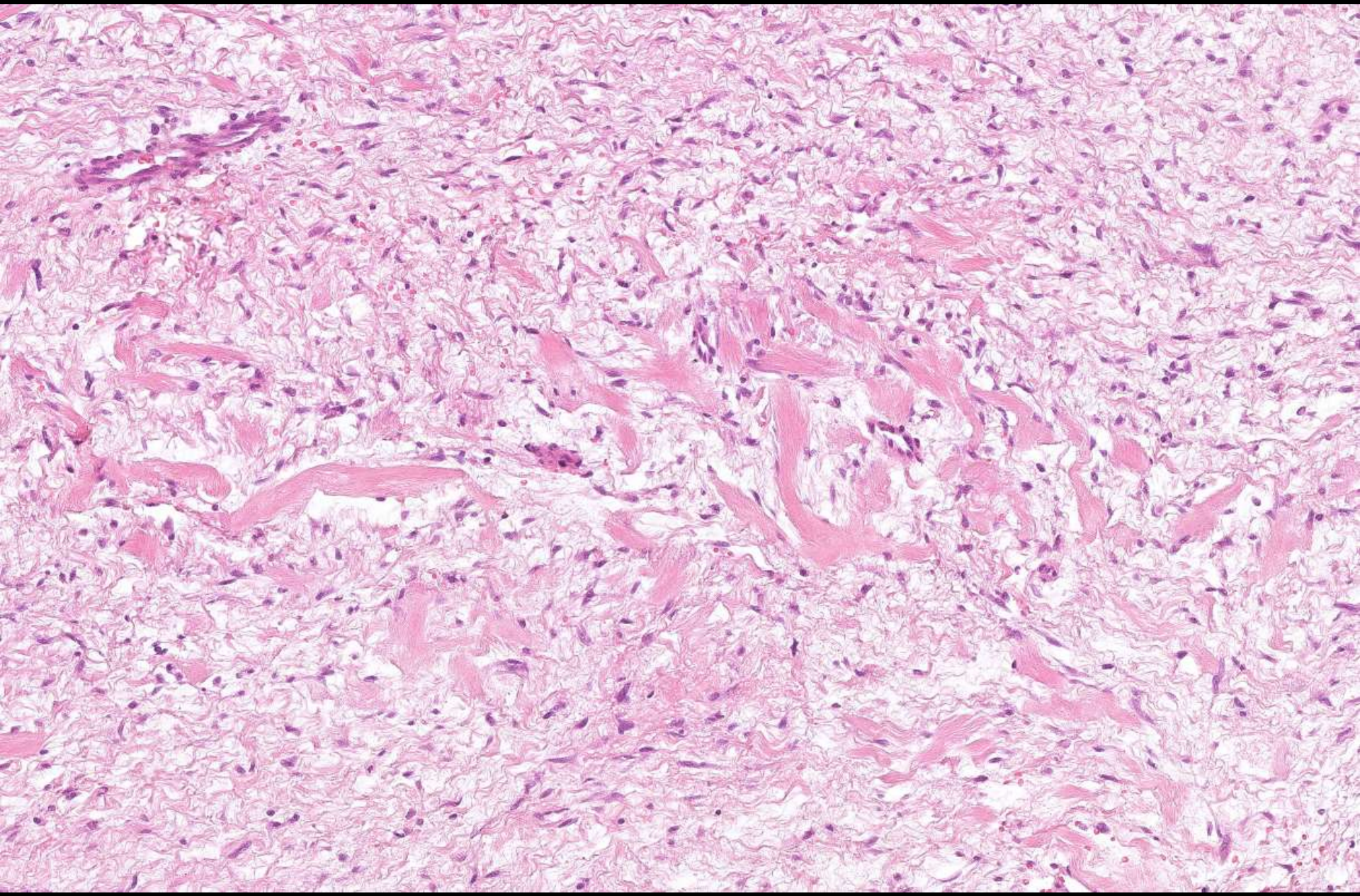
Peroperačná biopsia: Mezenchymálny hypocelulárny tumor, meningióm?

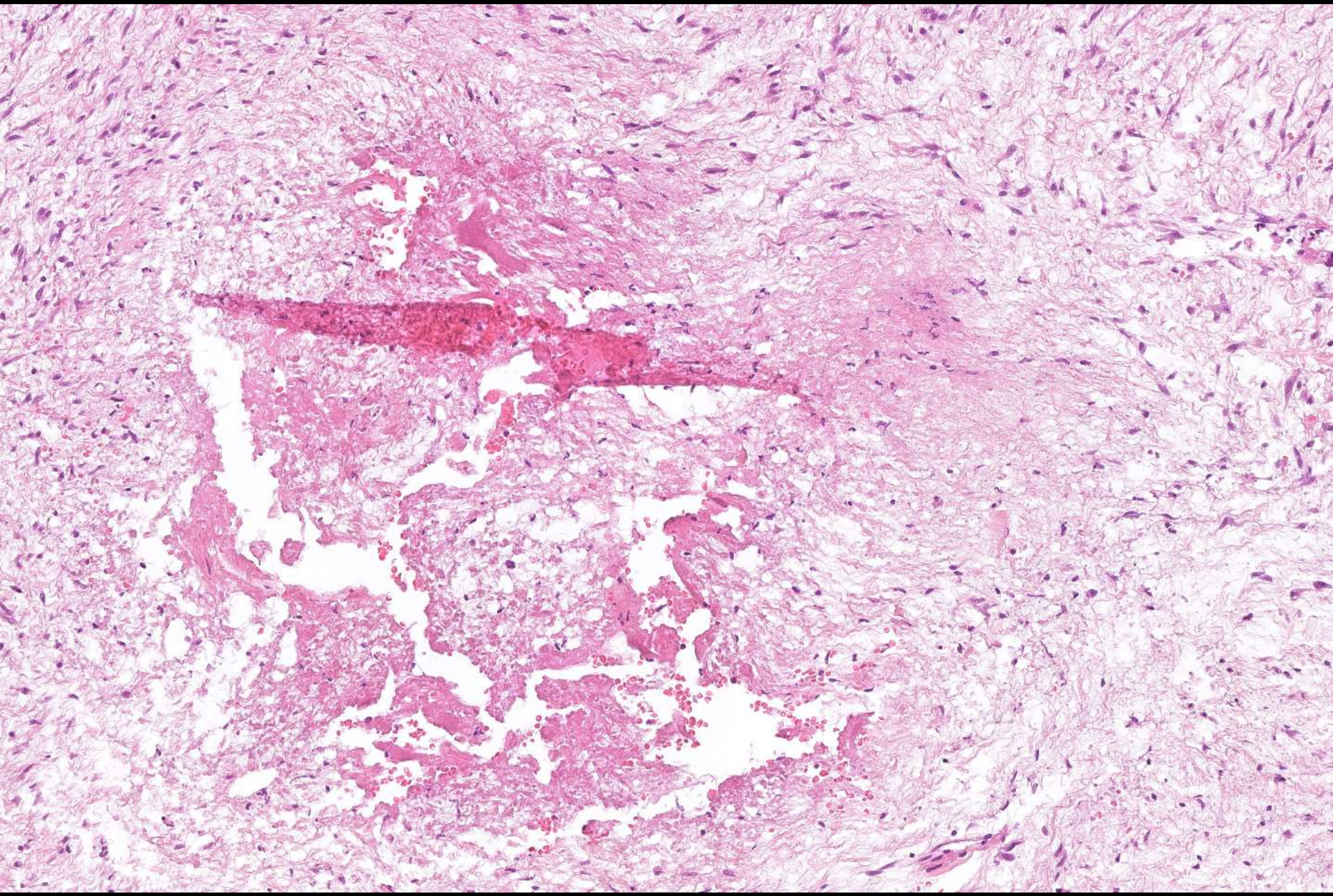


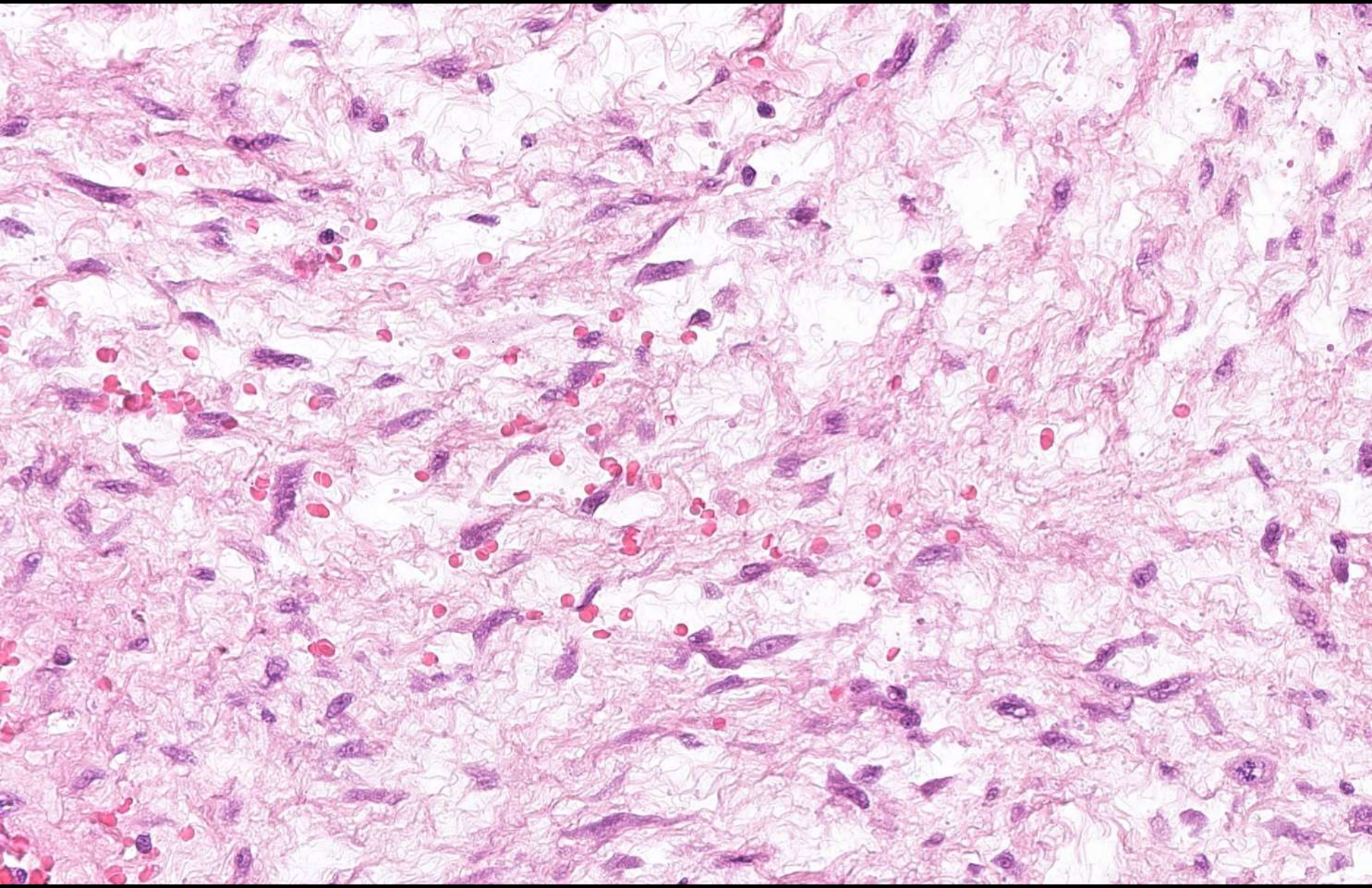


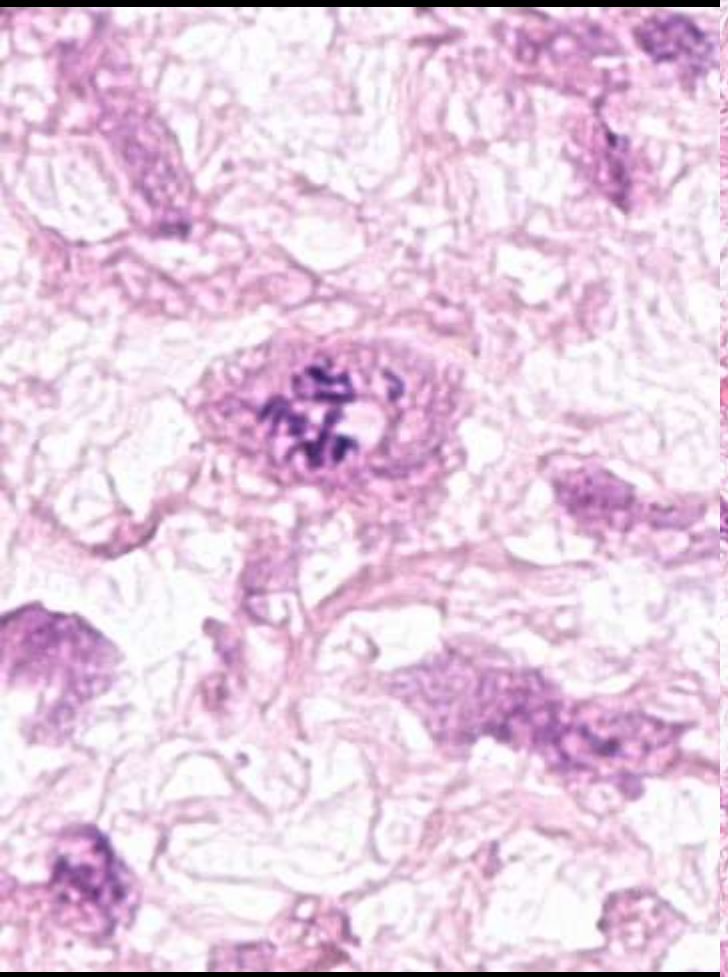


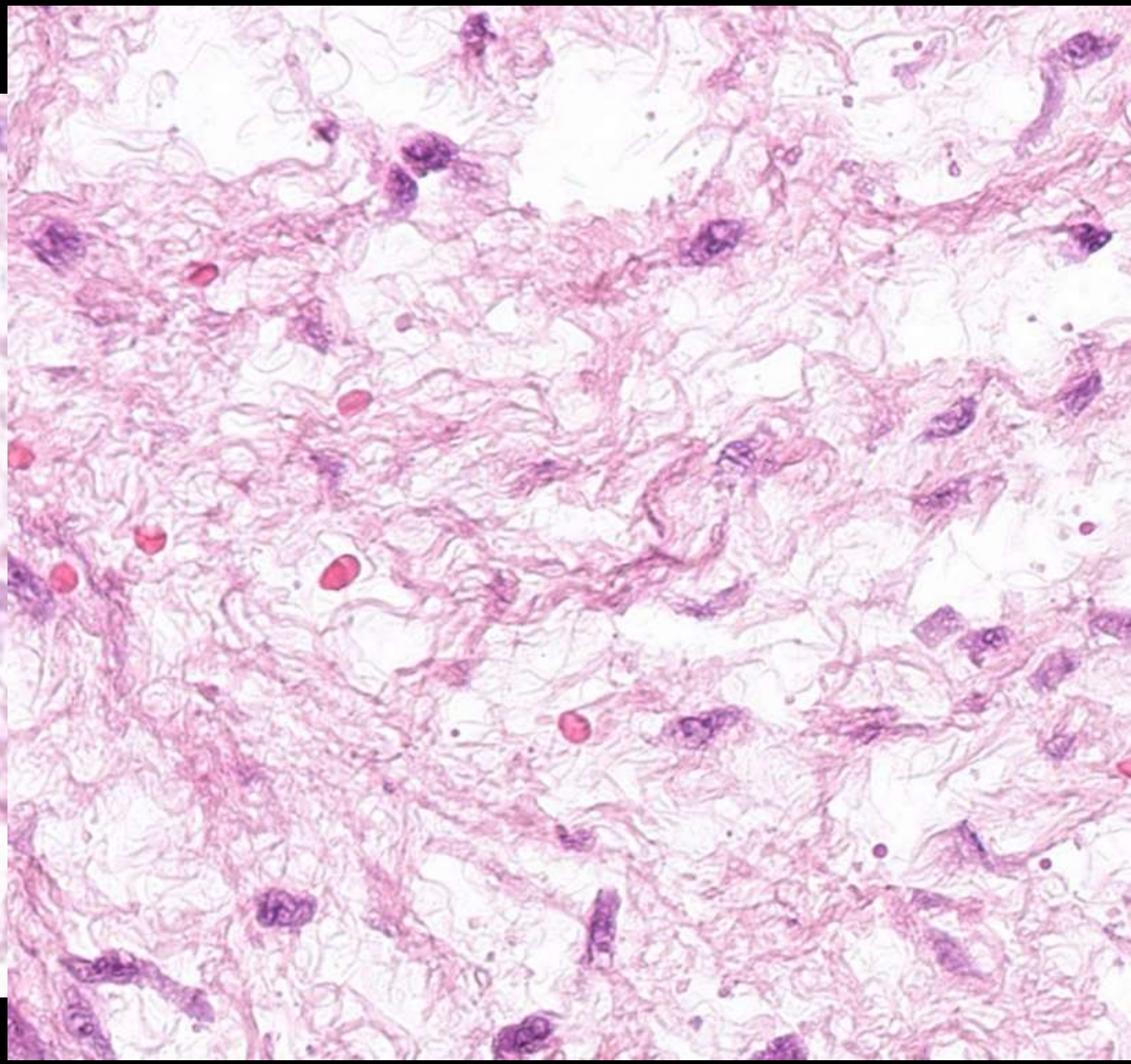


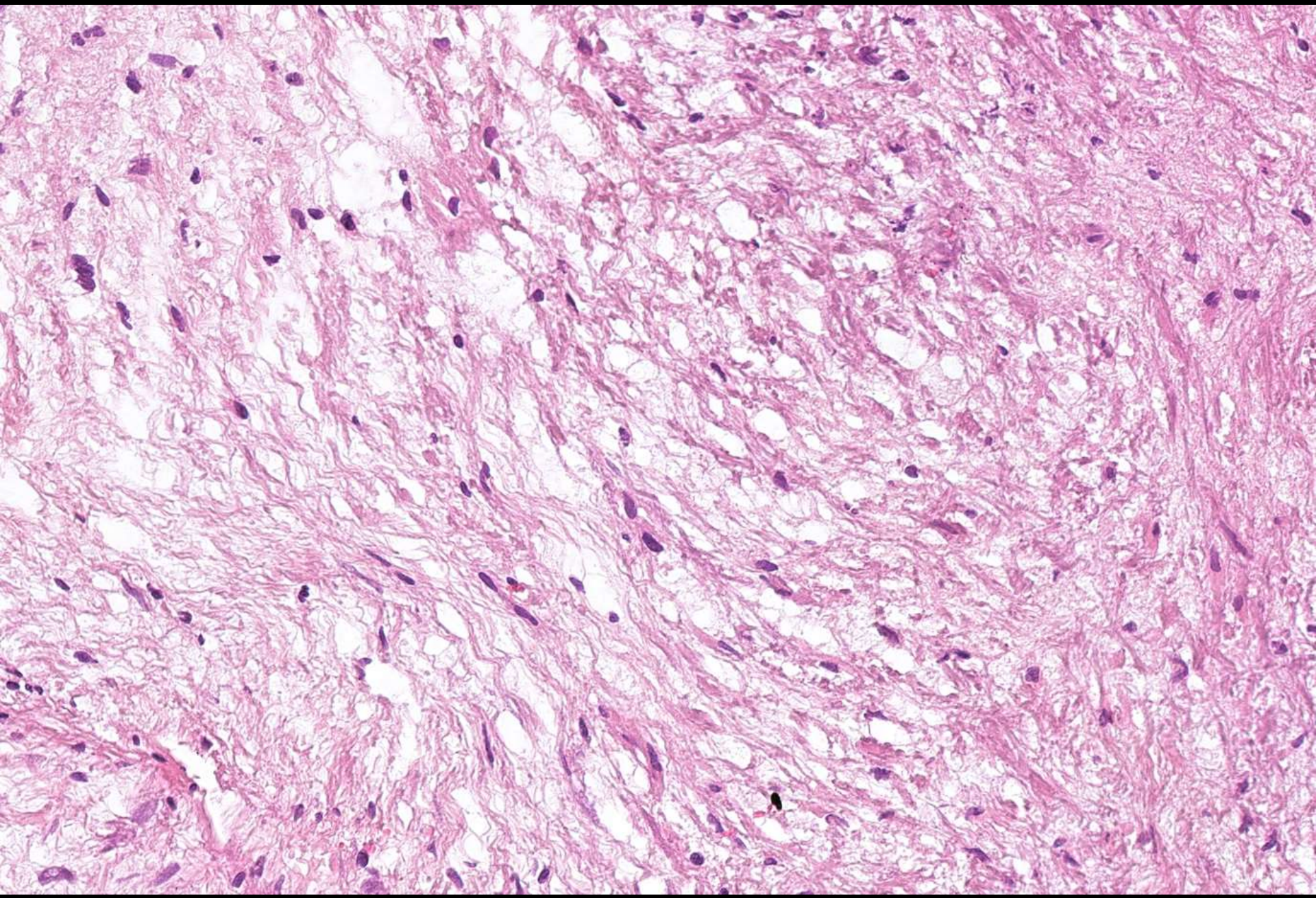


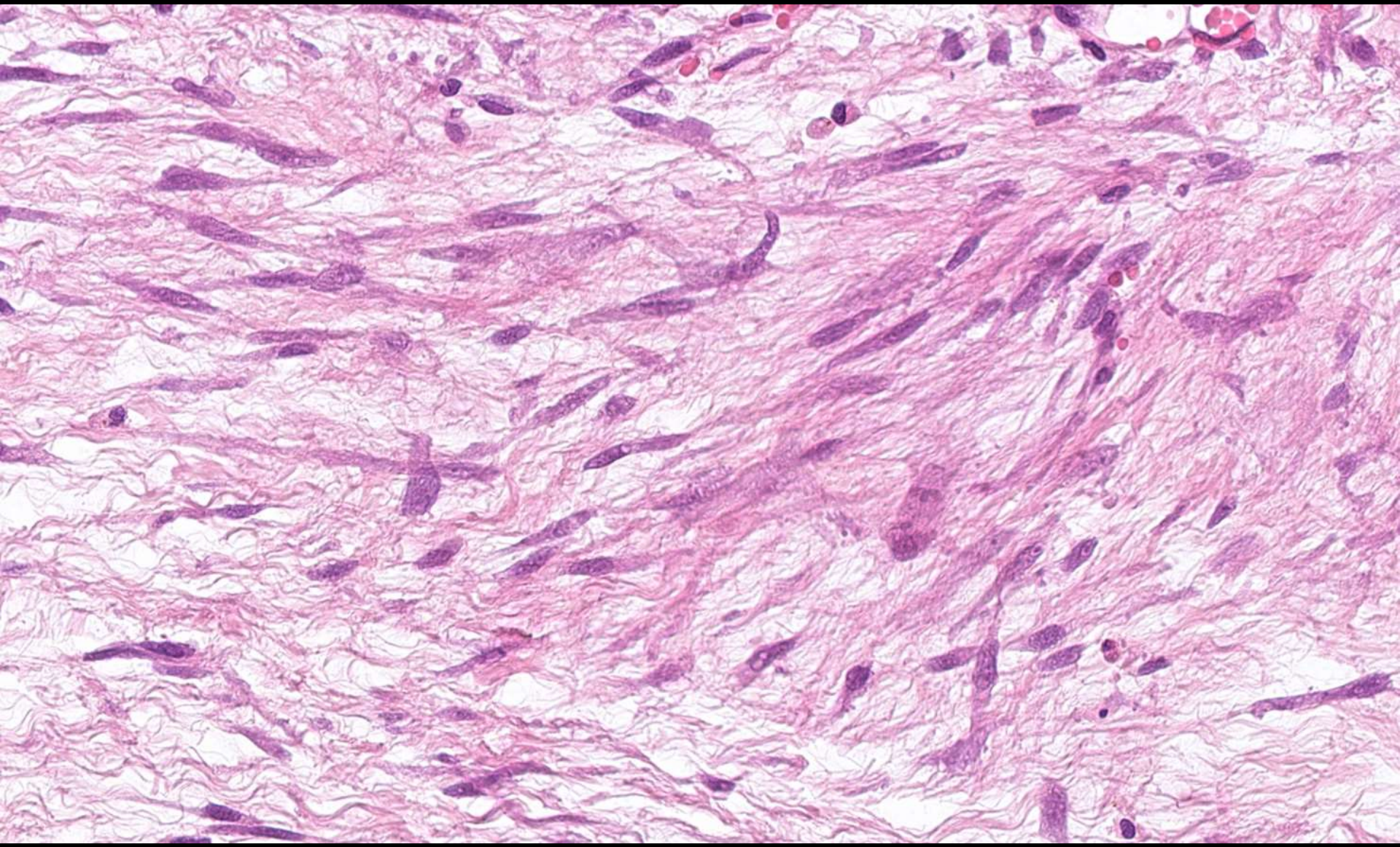


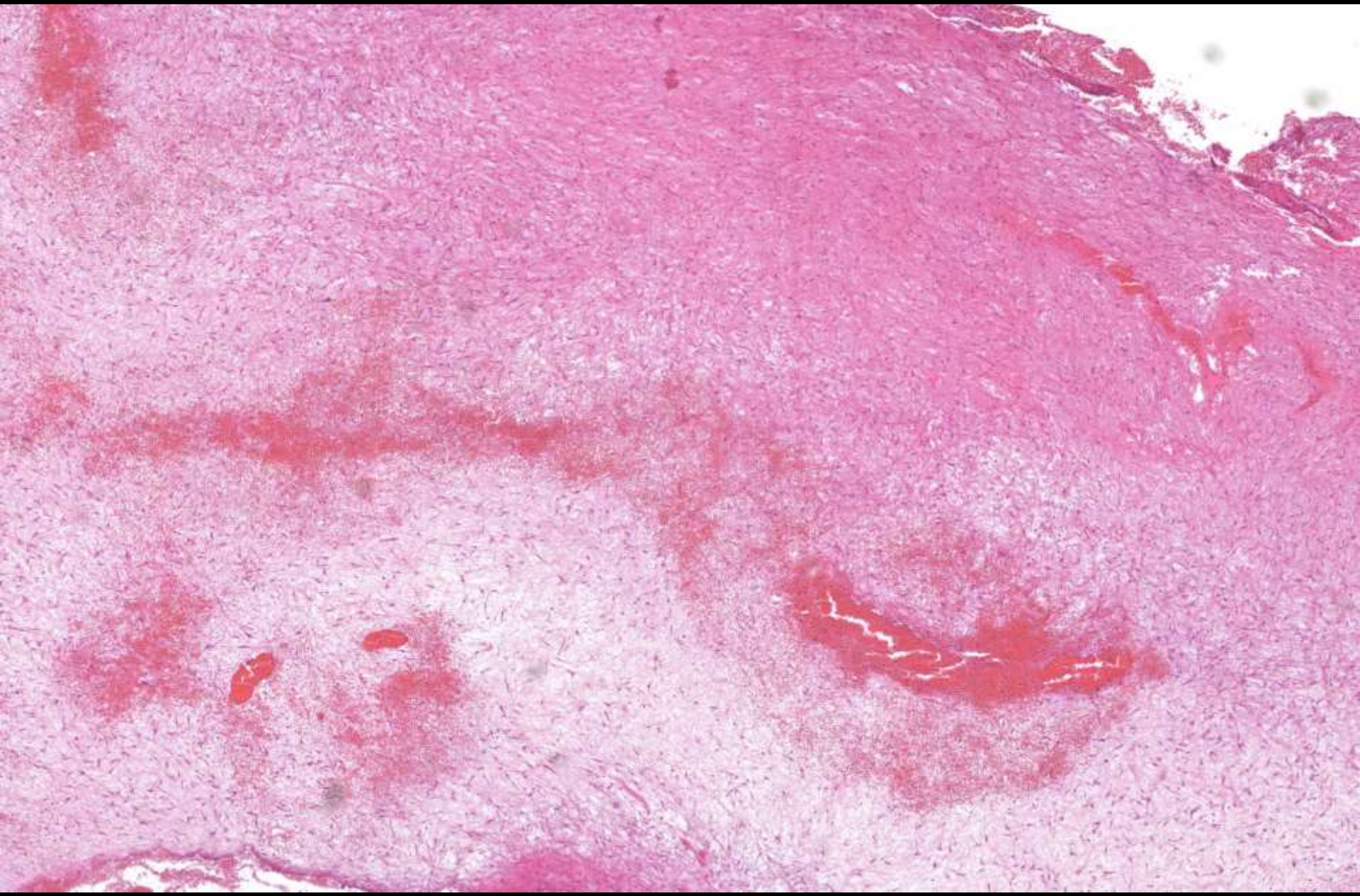


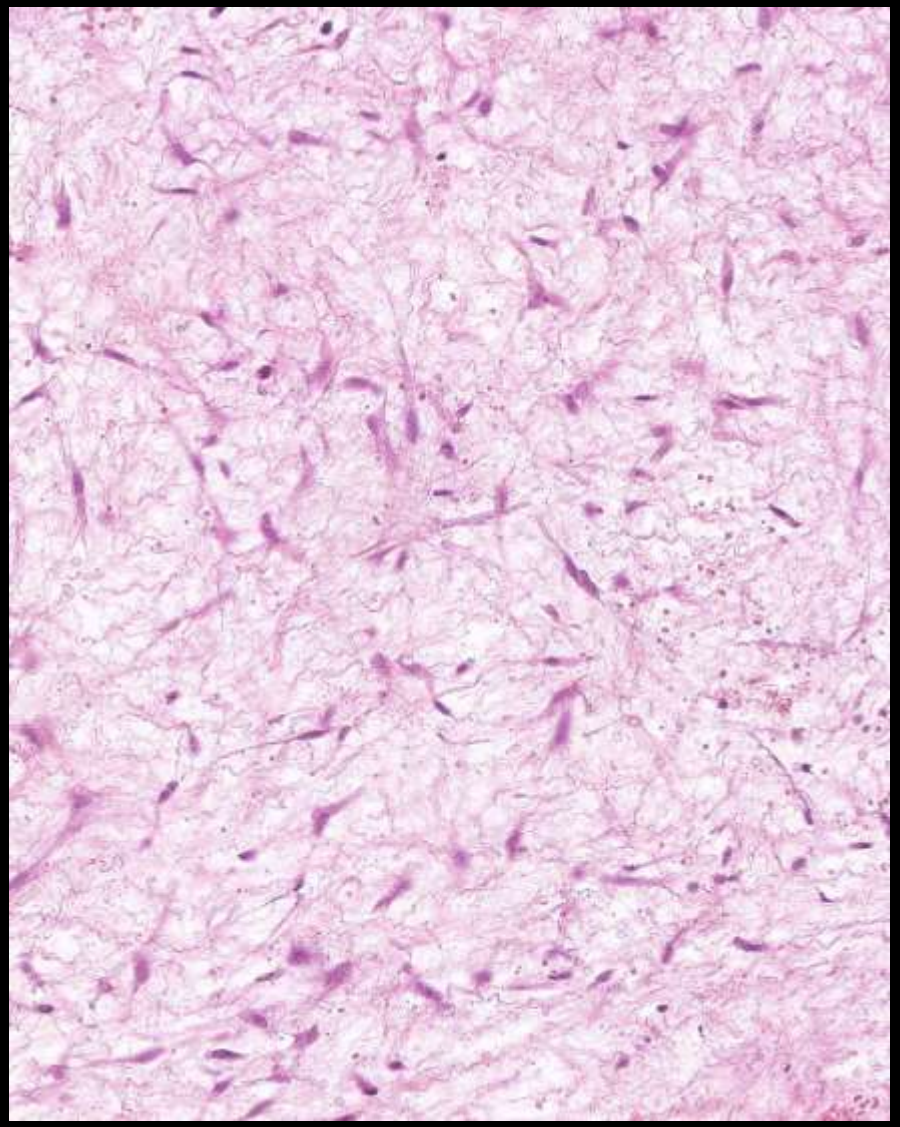


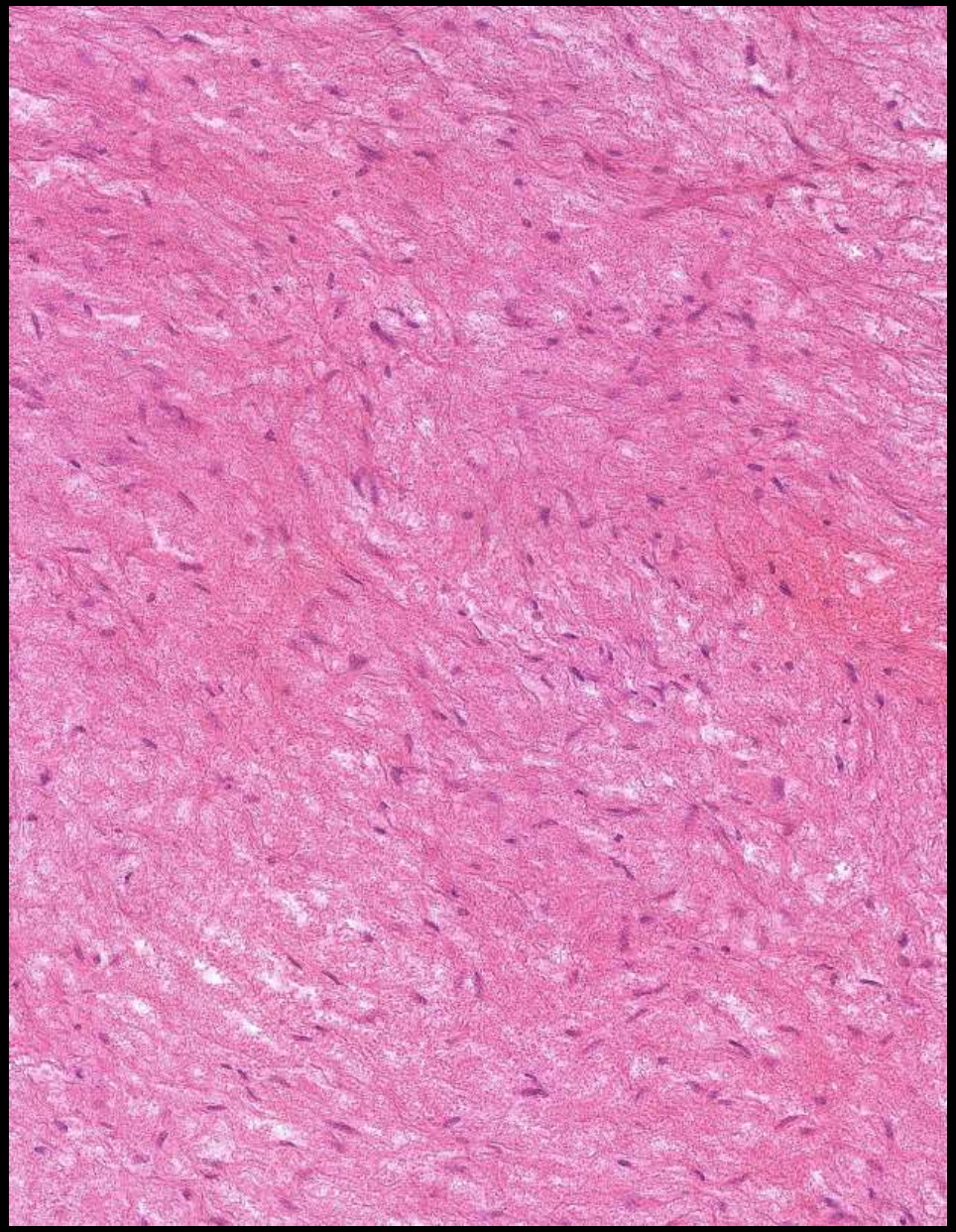


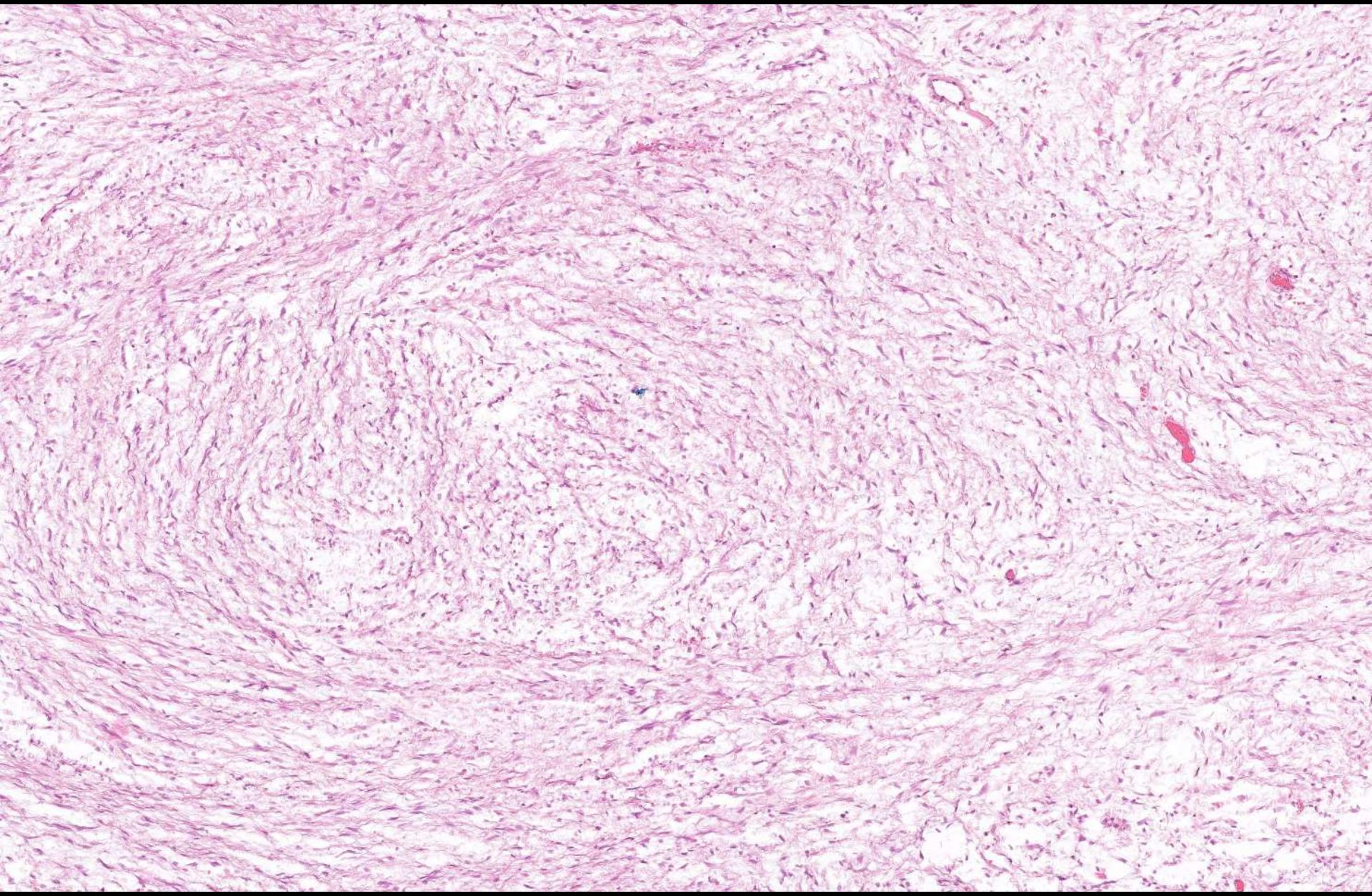


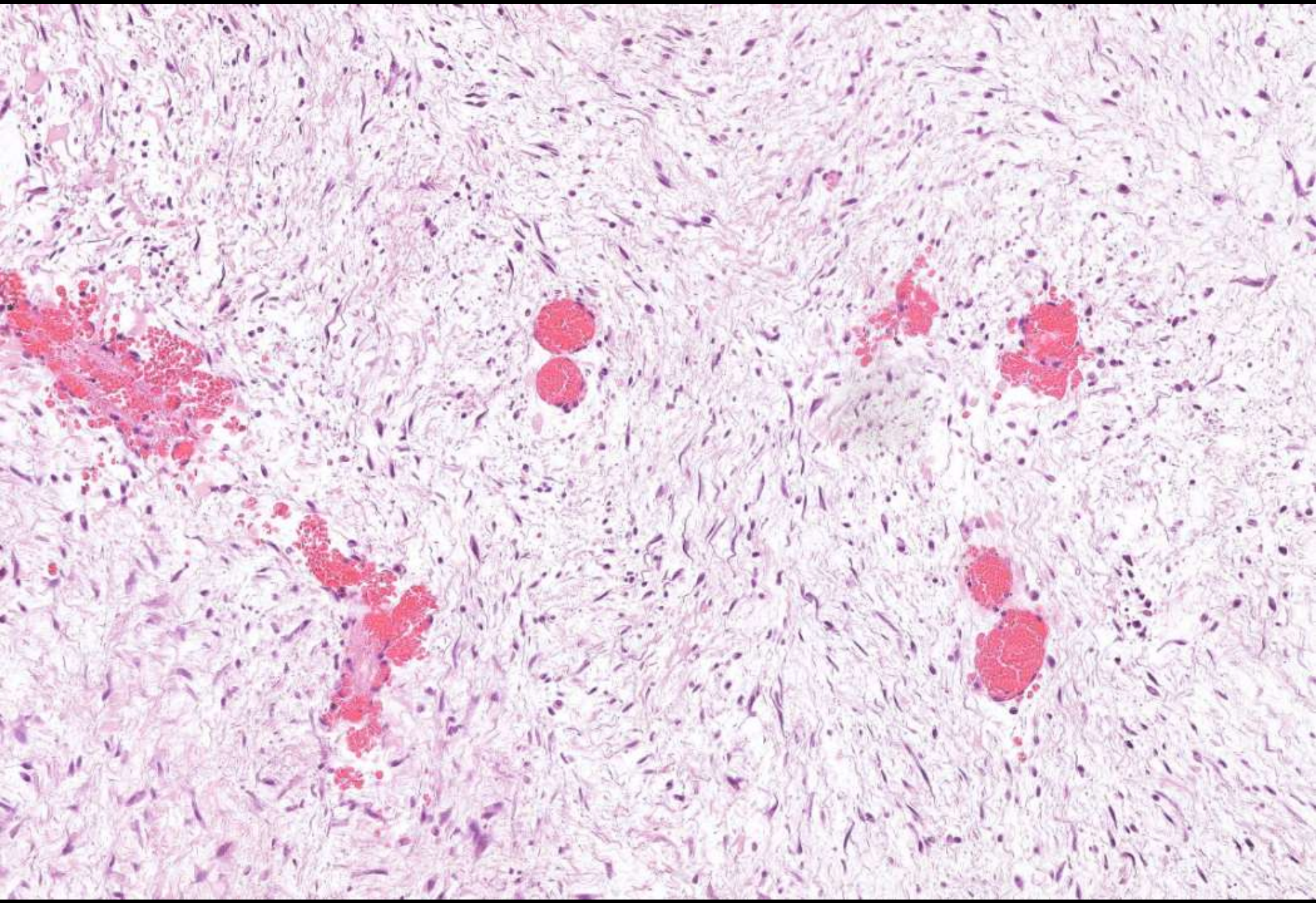


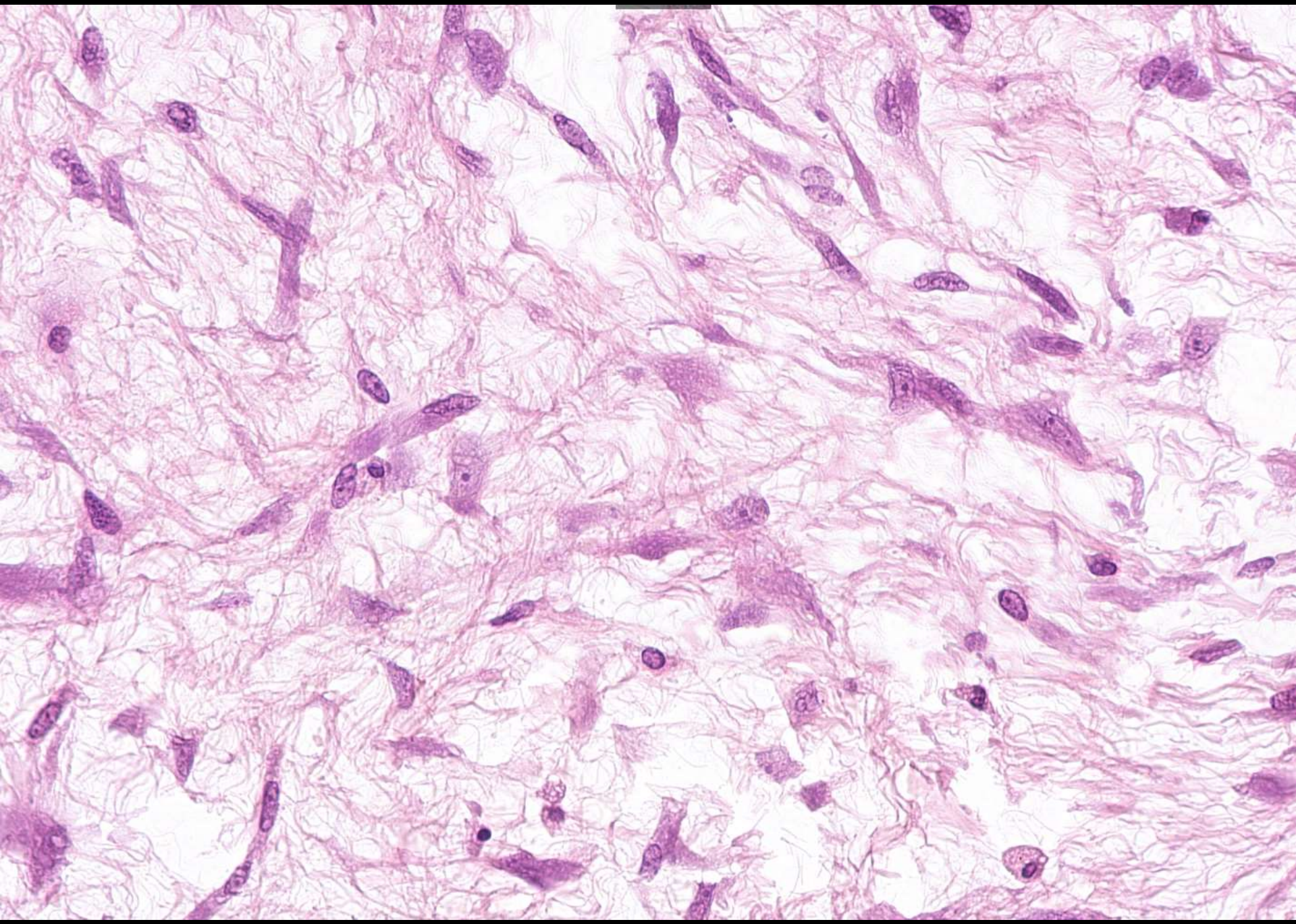


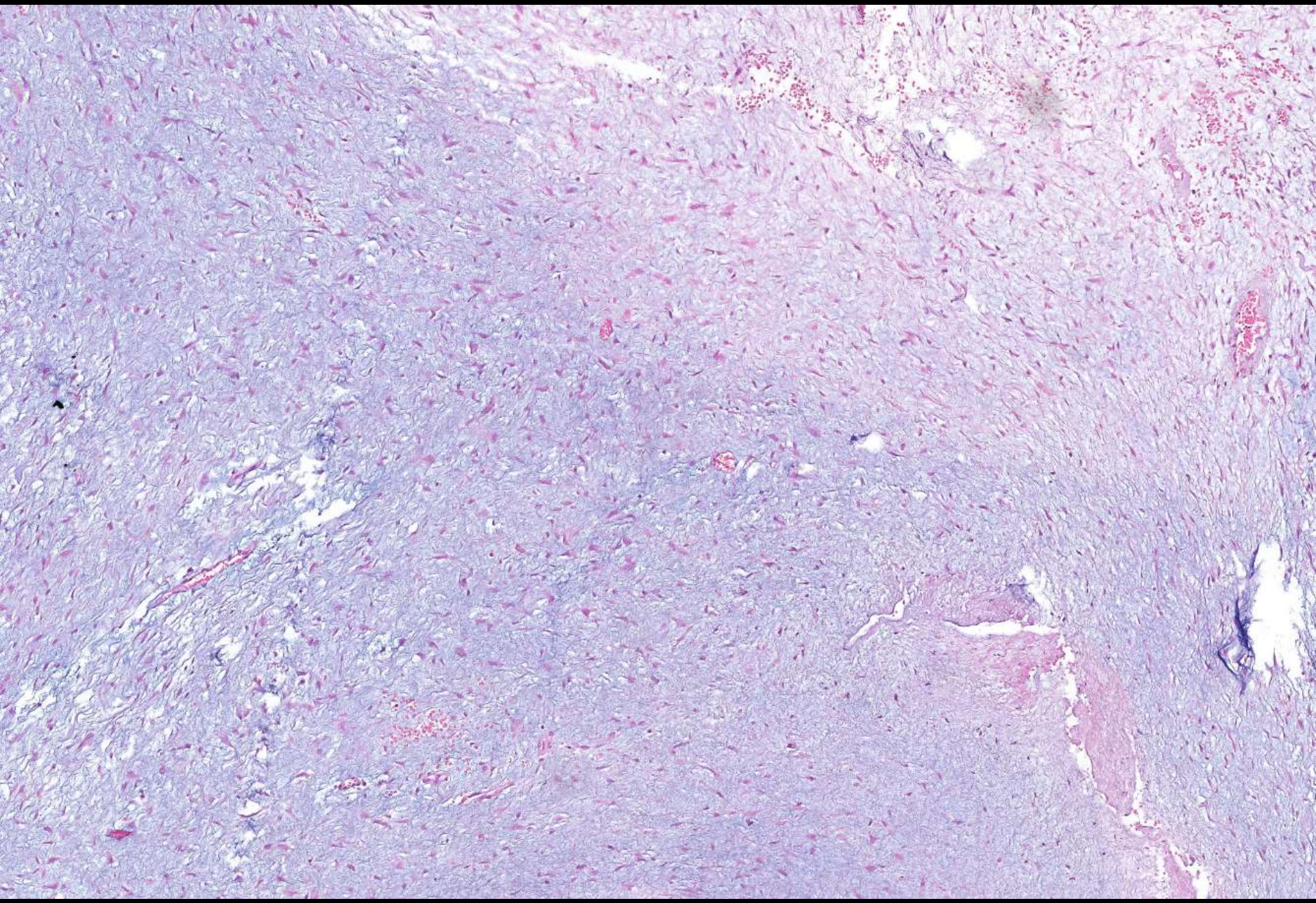


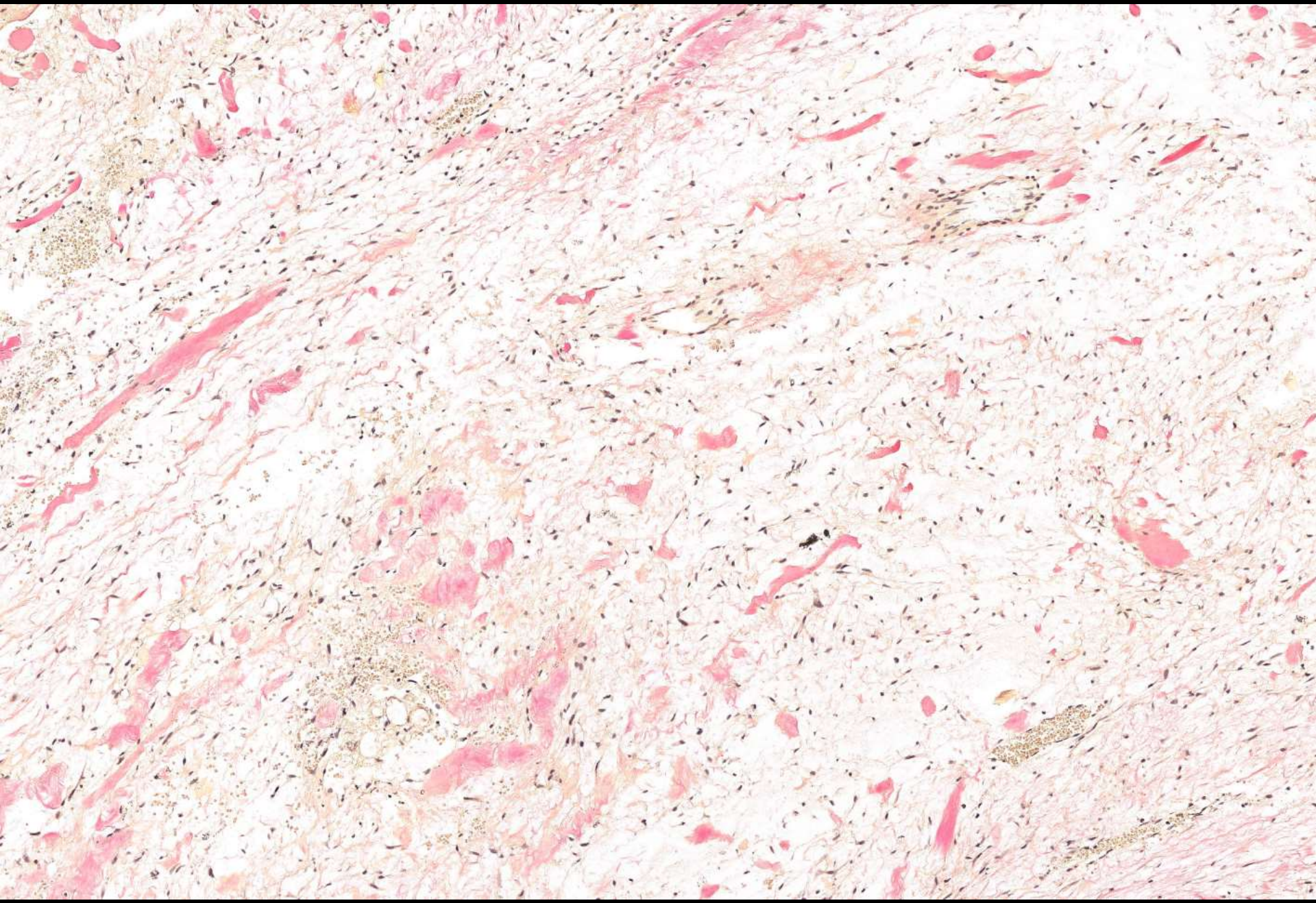












Výstrel na slepo

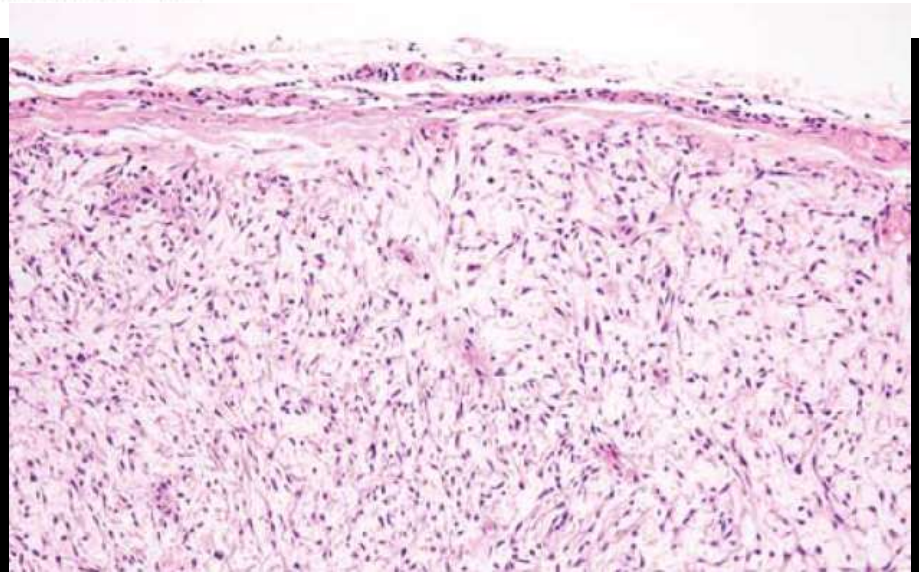
Brain Pathology ISSN 1015–6305

RESEARCH ARTICLE

Intracranial myxoid mesenchymal tumors with *EWSR1–CREB* family gene fusions: myxoid variant of angiomatoid fibrous histiocytoma or novel entity?

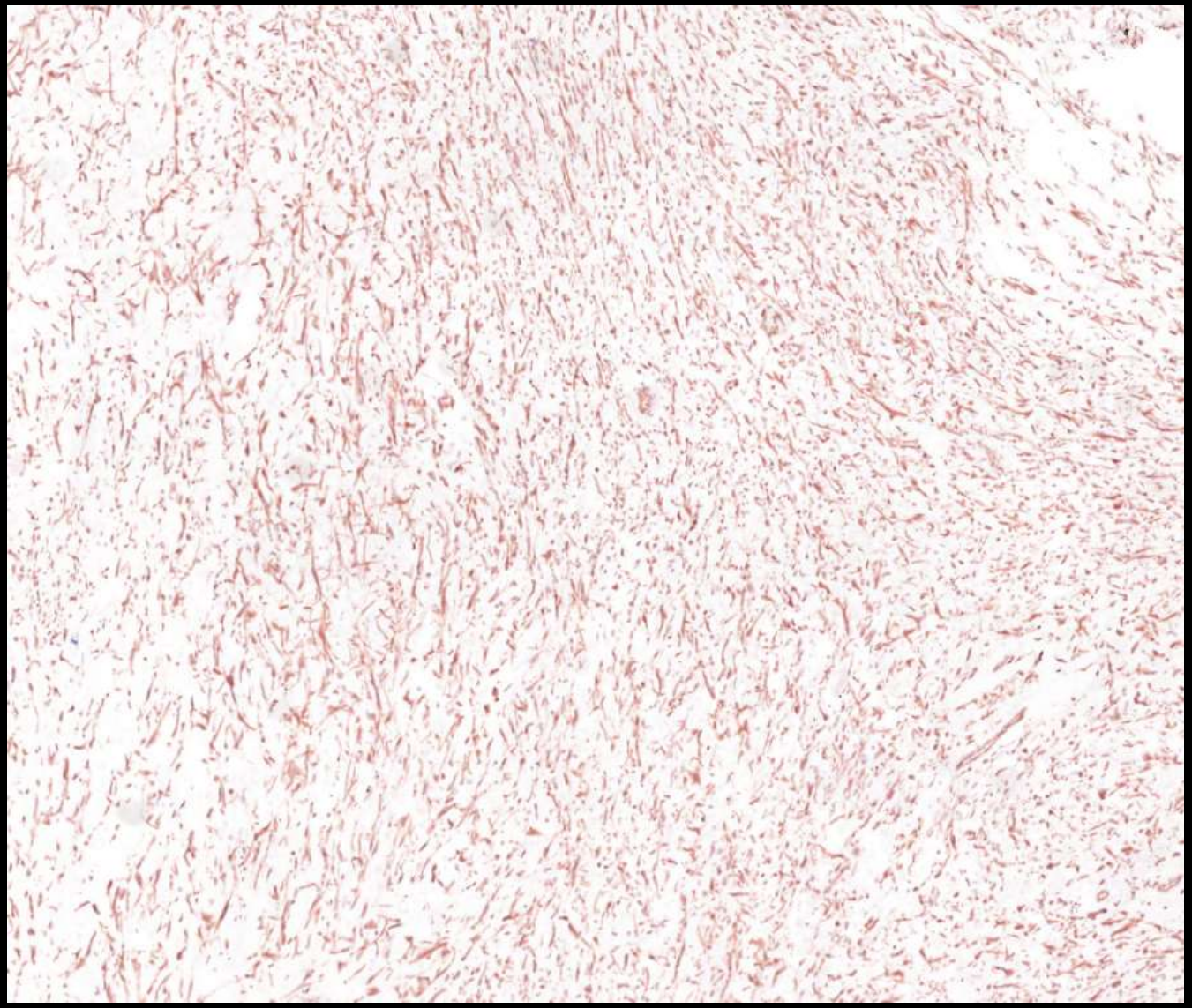
Tejus A. Bale[1], Angelica Oviedo[2], Harry Kozakewich[3], Caterina Giannini[4], Phani K. Davineni[1], Keith Ligon[1,3], Sanda Alexandrescu[3]

[1] Department of Pathology, Brigham and Women Hospital, Boston, MA.
[2] Department of Pathology Laboratory Medicine, IWK Health Center, Halifax, NS.
[3] Department of Pathology, Boston Children's Hospital, Boston, MA.
[4] Department of Pathology, Mayo Clinic, Rochester, MN.

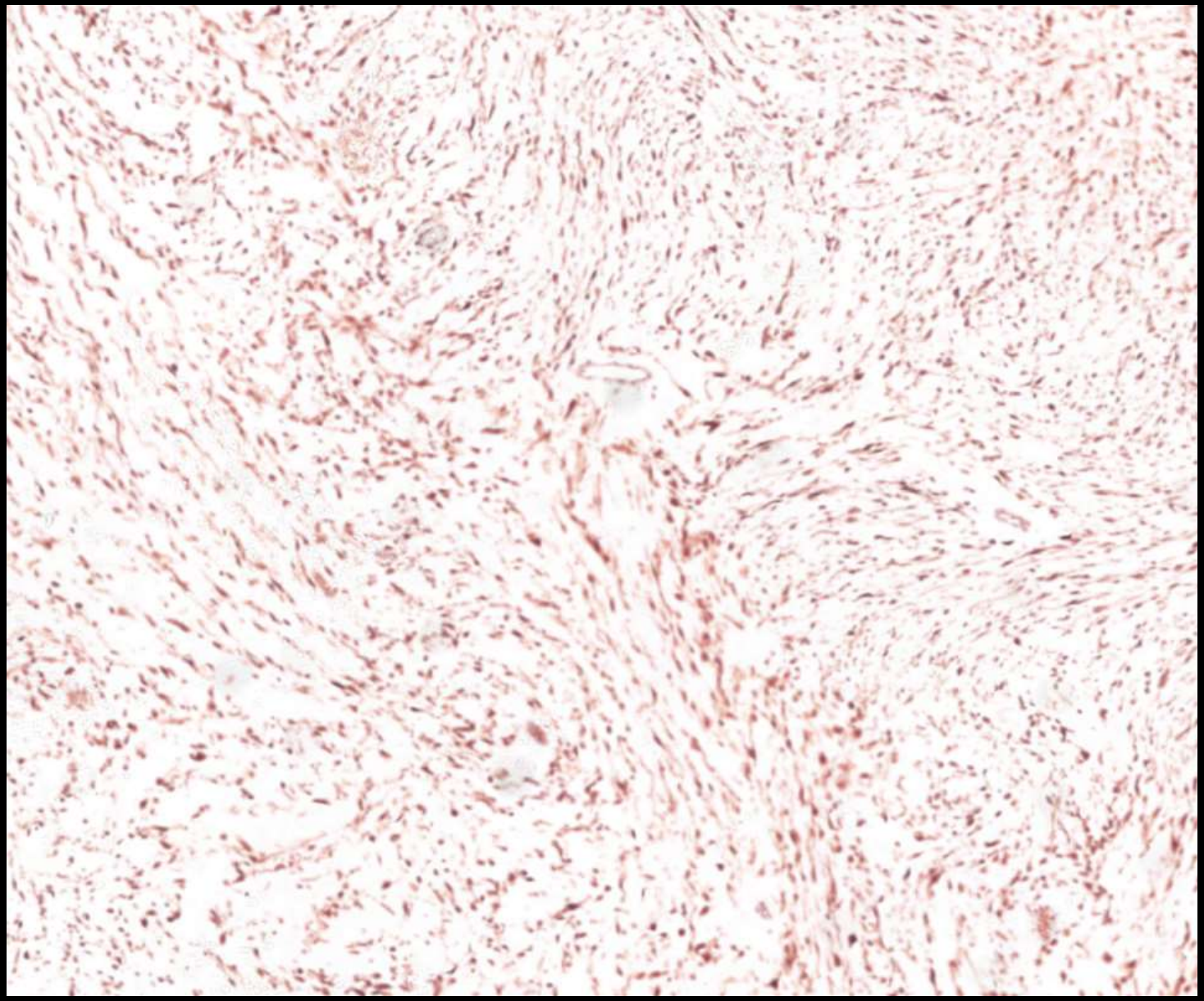


poslané na
EWSR FISH ...

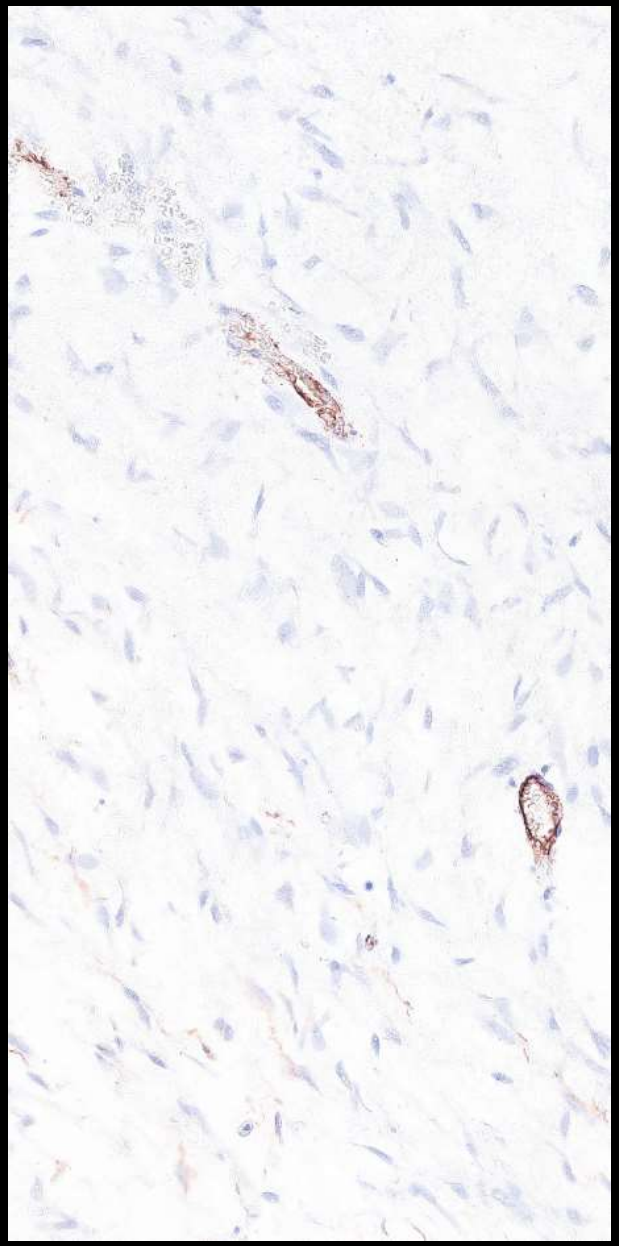
Vimentin



Beta Catenin

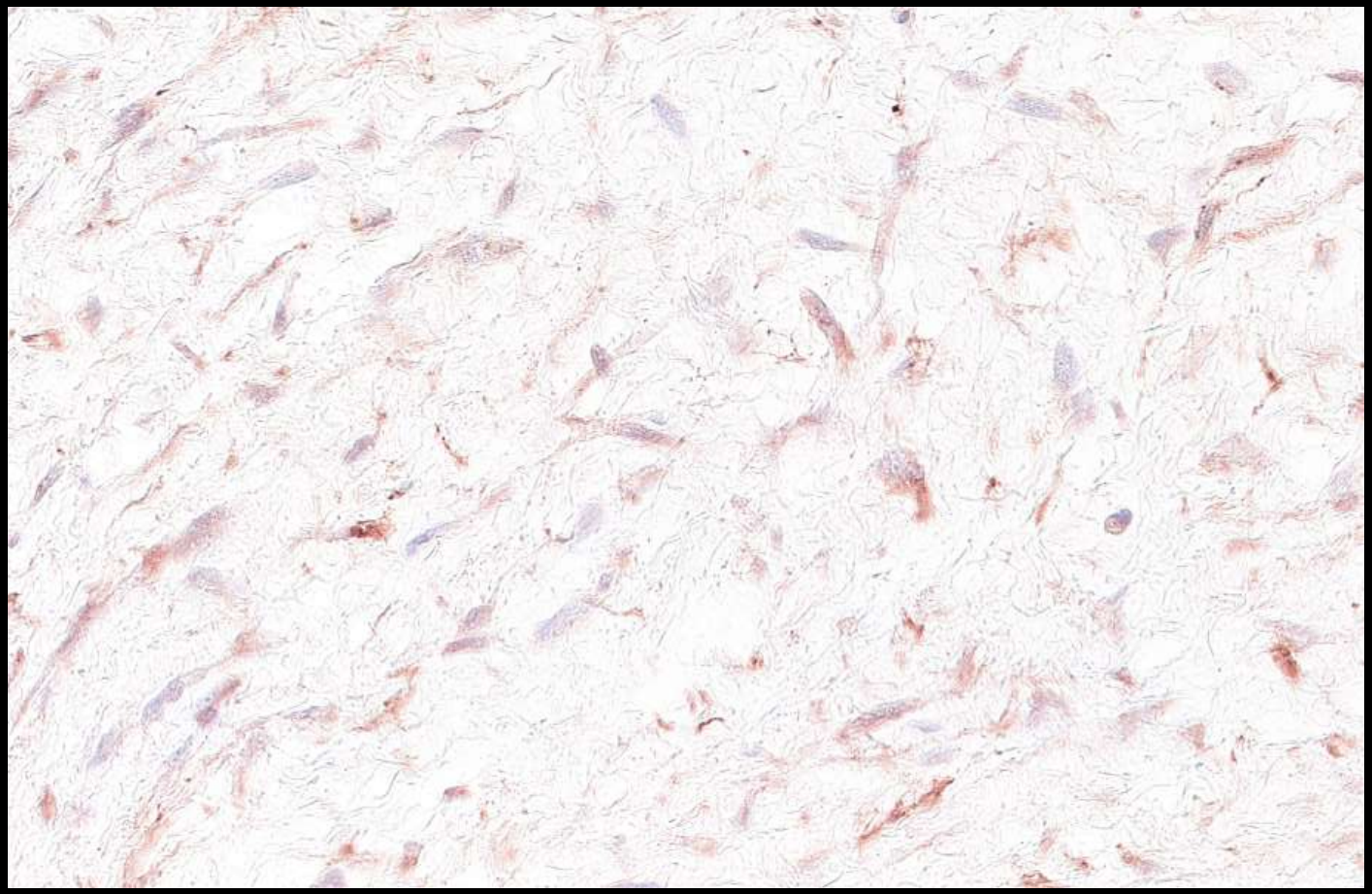


CD34

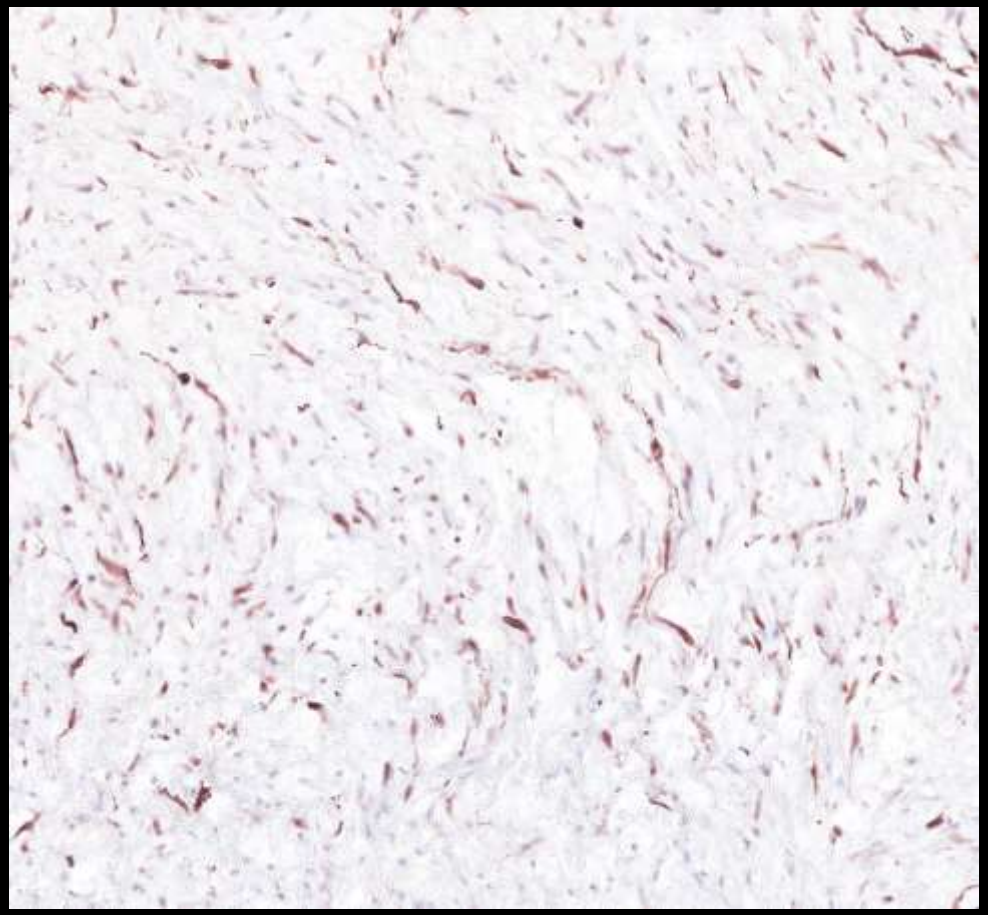


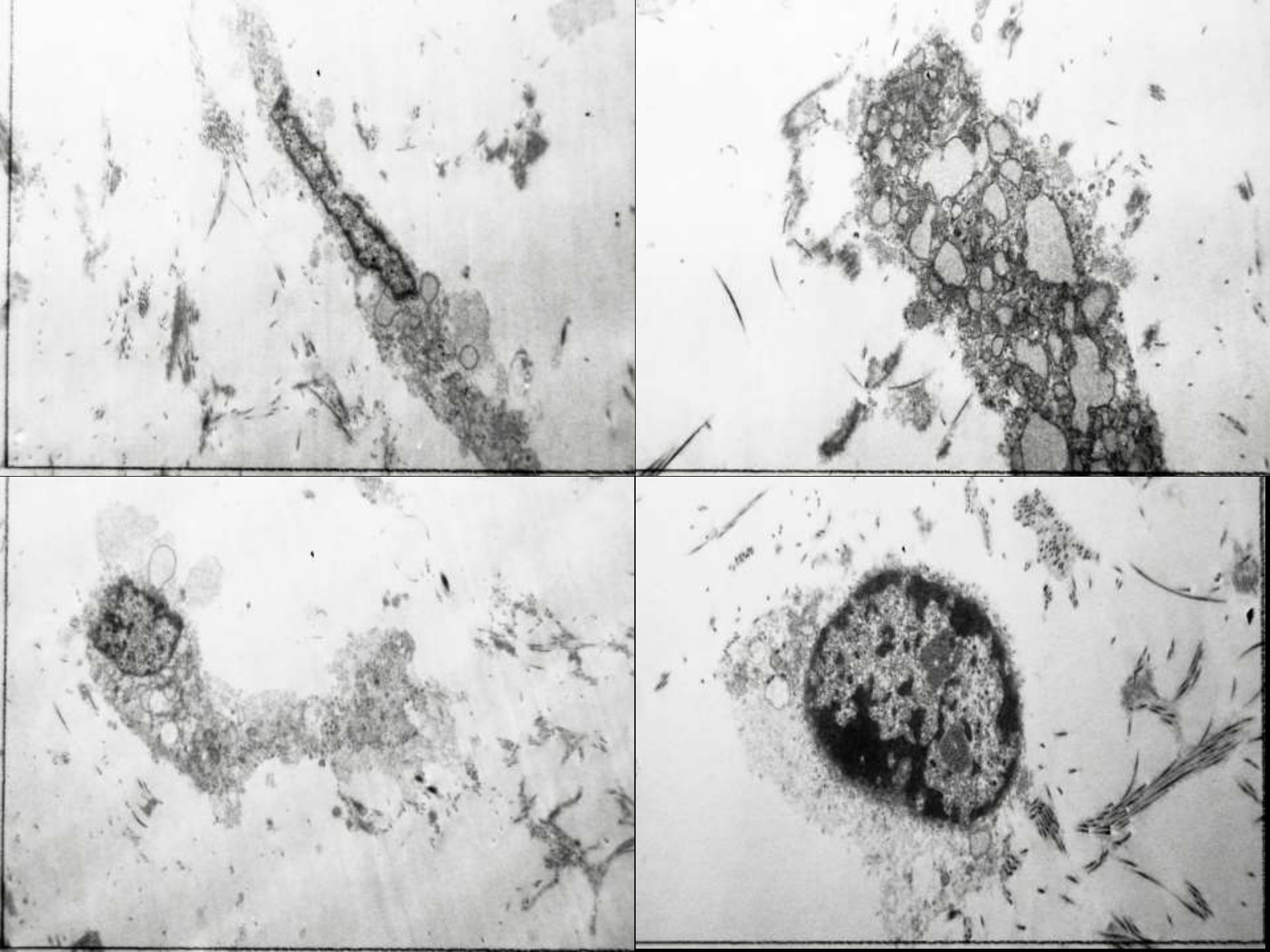
D2-40

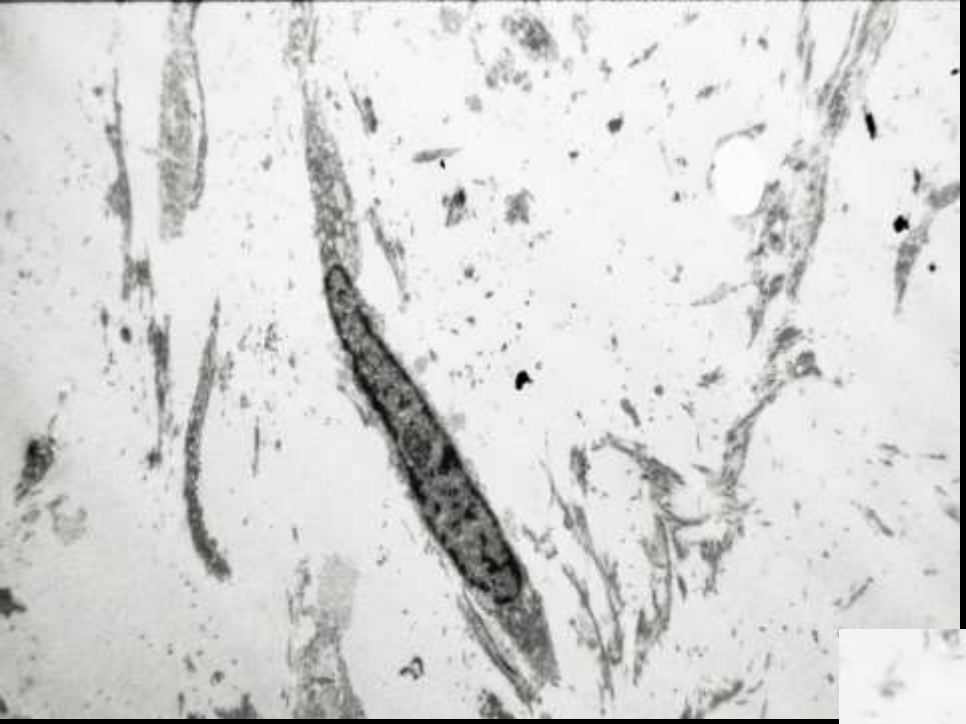
HHF35

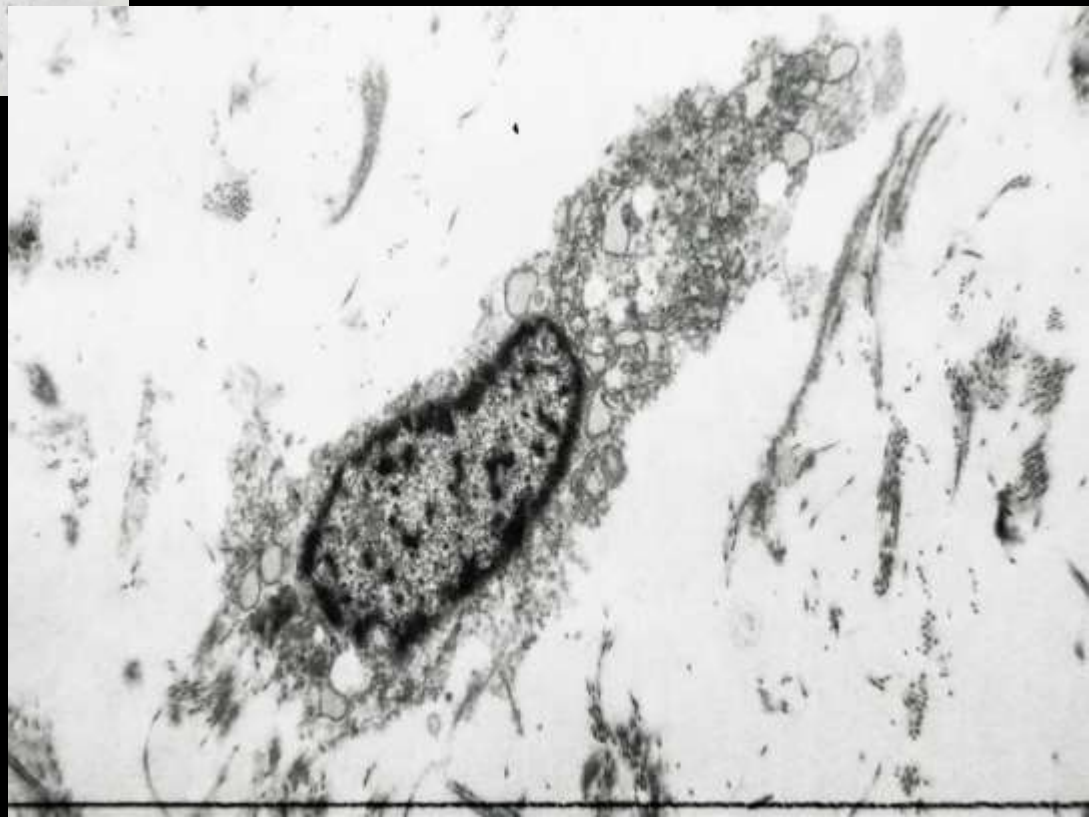


SMA







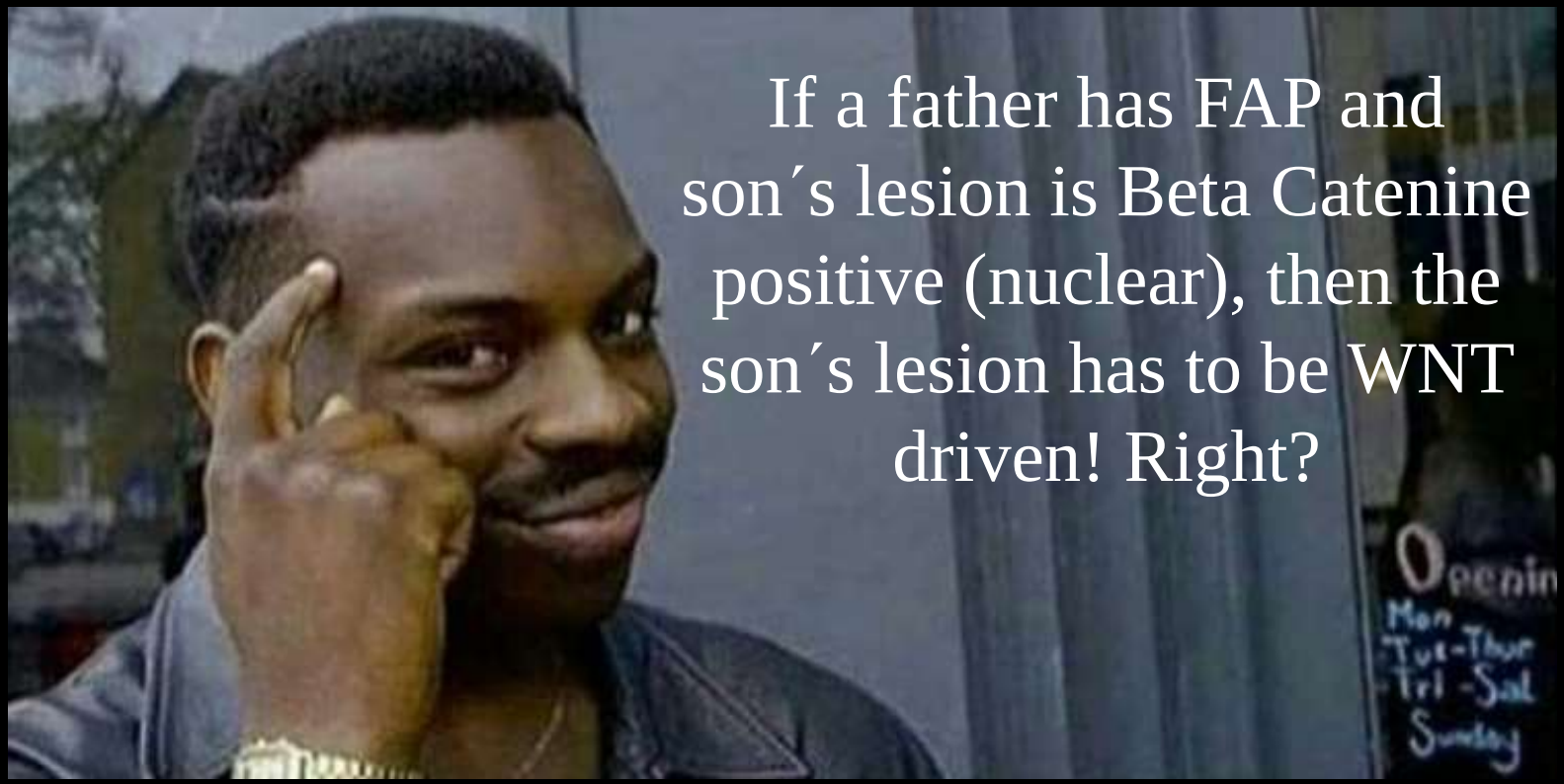


Zhrnutie IHC

- Vimentín pozit.
- zachovaná nukleárna pozitivita INI1
- +/- pozitivita: SMA, HHF35, D2-40
- Ki67 cca 2 %
- negat.: IDH1, Calretinín, Caldesmon, Calponín, CD34, AE1/3, GFAP, Olig2, BCL2, ERG, P53, MyoD1, PR, Dezmín, EMA, CD99, H3K27M, STAT6, Myogenín, TTF1, S100, SOX10, GFAP,

- EWSR bez prestavby
- vlastnú dg. som už mal
- ad Marián Švajdler – súhlasíš?
  - Analýza zlomu genu USP6 metódou FISH – negat.

NGS TruSight Oncology 500



NGS TruSight Oncology 500

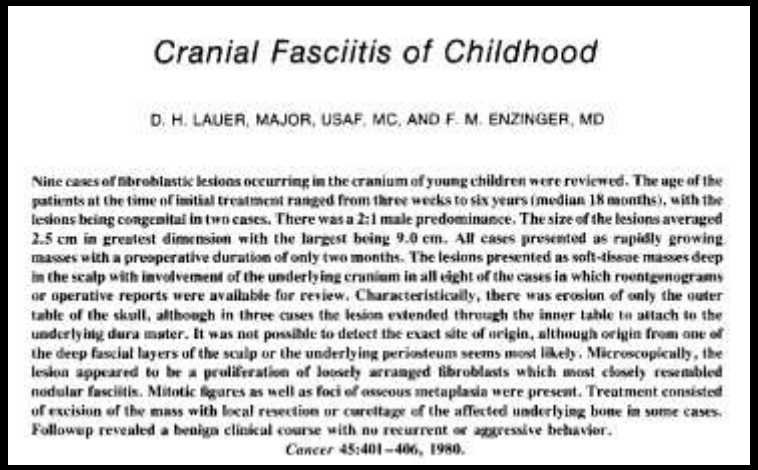
- bez alterácie WNT, ale s alteráciou MAPK – BRAF V600

Naša diagnóza

Kraniálna fascitída

(MŠ: Ide o bližšie neklasifikovateľný mezenchymálny (myo?)fibroblastický tumor s neistým biologickým potenciálom - morfologicky low-grade lézia, ktorá najviac pripomína kraniálnu fascitídu.)

Popísaná Enzingerom ako typ nodulárnej fascitídy



Cranial Fasciitis of Childhood

D. H. LAUER, MAJOR, USAF, MC, AND F. M. ENZINGER, MD

Nine cases of fibroblastic lesions occurring in the cranium of young children were reviewed. The age of the patients at the time of initial treatment ranged from three weeks to six years (median 18 months), with the lesions being congenital in two cases. There was a 2:1 male predominance. The size of the lesions averaged 2.5 cm in greatest dimension with the largest being 9.0 cm. All cases presented as rapidly growing masses with a preoperative duration of only two months. The lesions presented as soft-tissue masses deep in the scalp with involvement of the underlying cranium in all eight of the cases in which roentgenograms or operative reports were available for review. Characteristically, there was erosion of only the outer table of the skull, although in three cases the lesion extended through the inner table to attach to the underlying dura mater. It was not possible to detect the exact site of origin, although origin from one of the deep fascial layers of the scalp or the underlying periosteum seems most likely. Microscopically, the lesion appeared to be a proliferation of loosely arranged fibroblasts which most closely resembled nodular fasciitis. Mitotic figures as well as foci of osseous metaplasia were present. Treatment consisted of excision of the mass with local resection or curettage of the affected underlying bone in some cases. Followup revealed a benign clinical course with no recurrent or aggressive behavior.

Cancer 45:401–406, 1980.

- 9 prípadov, kraniálna pseudosarkomatózna proliferácia u detí
- mäkké tkanivá s defektom kalvy
- len jedna väčšia ako 3,5 cm (9 cm)
- pravdepodobne paraoseálny pôvod
- mitózy
- benígna
- trauma možný etiologický činiteľ

Podtyp nodulárnej fascitídy?

	Kraniálna fascitída	Nodulárna fascitída
lokalita	kránium, temporo-parietálna oblasť	končatiny, trup, chrbát
vek	takmer výlučne do 6, priemer 2 roky	priemer 40 rokov
muži / ženy	2/1	1/1
histológia	uniformne, myxoidné, skor hypocelulárne	variabilná histológia, celularita, zonácia
genetika	USP6 do cca 30%	USP6 viac ako 90%

Kraniálna fascitída - histológia

- dobre ohraničená, neopuzdrená, málo celulárna
- vretenovité, hviezdicovité bunky (fibroblasty, myofibroblasty) na myxoidnom pozadí
- krvácania, stromálna sklerotizácia, tenkostenné cievy
- zápalové bb., makrofágy, občas mnohojadrové bunky

Kraniálna fascitída - histológia

- fokálne môžu byť atypie, mitózy
- občas metaplastická osifikácia
- rast, stacionárna fáza, možná regresia
- bez rekurencií
- excízia je postačujúca Th
- raritná (cca 50 prípadov v literatúre)
- niekedy po traume, 12-19 % (vrátane pôrodnej), kraniotómii, RAT pre mozgový TU


J Neurosurg 106:1080–1085, 2007

Postoperative cranial fasciitis

Report of two cases and review of the literature

Lori E. Summers, M.D.,[1] Luisa Florez, M.D.,[2] John M. Berberian, M.D.,[2] Meena Bhattacharjee, M.D.,[4] and John W. Walsh, M.D., Ph.D.[3]

[1]Department of Neurological Surgery, University of Florida, Gainesville, Florida; Departments of [2]Pathology and [3]Neurological Surgery, Tulane University Medical Center, New Orleans, Louisiana; and [4]Department of Pathology, Texas Children's Hospital, Houston, Texas


Neuropathology 2014; 34, 291–294

Case Report

Parasagittal cranial fasciitis following infratemporal fossa rhabdomyosarcoma

Eyas M. Hattab,[1,2] Lauren E. Dvorscak,[1] Joel C. Boaz,[1] Annette C. Douglas[1] and Thomas M. Ulbright[1]

Departments of [1]Pathology and Laboratory Medicine, [2]Neurological Surgery, [3]Radiology, Indiana University School of Medicine, Indianapolis, Indiana, [4]Department of Pathology, University of New Mexico School of Medicine, Albuquerque, New Mexico, USA


Case Reports > Neurosurgery. 2004 May;54(5):1263-6; discussion 1266-7.

doi: 10.1227/01.neu.0000119604.10923.63.

Parasagittal cranial fasciitis after irradiation of a cerebellar medulloblastoma: case report

Pierluigi Longatti [1], Elisabetta Marton, Laura Bonaldi, Enrico Orvieto

Affiliations + expand

doi:10.1111/neup.12302

DOI: 10.1227/01.neu.0000119604.10923.63

Môže byť čisto intrakraniálna

Pediatric and Developmental Pathology 15, 146–150, 2012
DOI: 10.2350/11-02-0990-CR.1

Cranial Fasciitis Presenting as an Intracranial Mass in a 10-Year-Old Girl

Anirudha Halder,[1] Clarence S. Greene,[2] Douglas C. Rivard,[3] and Lei Shao[4]*
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[2]Children's Hospital, New Orleans, LA 70118, USA
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Received March 7, 2011; accepted November 14, 2011; published online November 23, 2011.

Pediatric Neurosurgery

Case Report

Pediatr Neurosurg 2008;44:148–152
DOI: 10.1159/000113119

Received: March 22, 2006
Accepted after revision: November 29, 2006
Published online: January 24, 2008

Cranial Fasciitis Presenting with Intracranial Mass: A Case Report

Naoya Takeda[a] Katsuzo Fujita[a] Shigenori Katayama[a] Nobuyuki Akutsu[a]
Kimio Hashimoto[b] Eiji Kohmura[c]

Departments of [a]Neurosurgery and [b]Pathology, Nishi-Kobe Medical Center, [c]Department of Neurosurgery, Kobe University Graduate School of Medicine, Kobe, Japan

Acta Neuropathol (2006) 111: 286–288
DOI 10.1007/s00401-005-0020-1

LETTER TO THE EDITOR

Manuela Agozzino · Alessandra Cavallero
Frediano Inzani · Ilaria Acchiardi · Davide Locatelli
Paola Scagnelli · Clara Malattia · Eloisa Arbustini

Cranial fasciitis with exclusive intracranial extension in an 8-year-old girl

J Neurosurg 83:744–747, 1995

Exclusively intracranial cranial fasciitis in a child

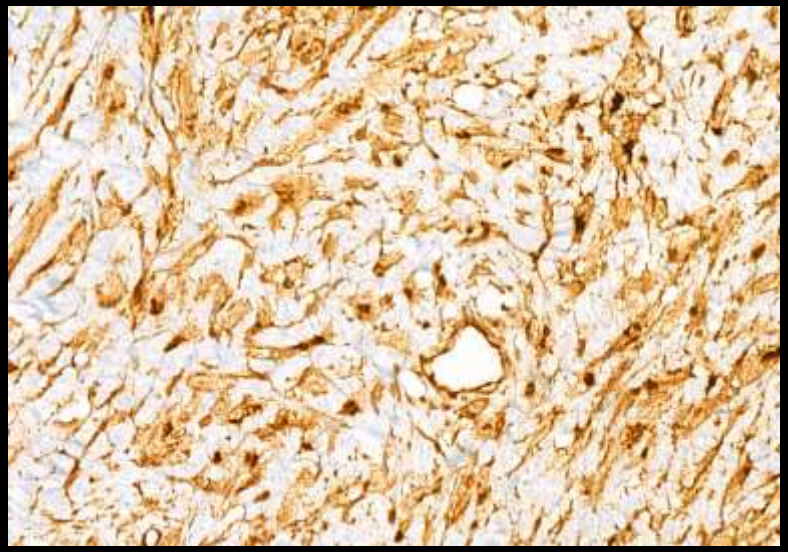
Case report

Axel Pagenstecher, M.D., Beate Emmerich, M.D., Vera van Velthoven, M.D., Rudolf Korinthenberg, M.D., and Benedikt Volk, M.D.

Departments of Neuropathology, Neuropediatrics, and Neurosurgery, University of Freiburg, Freiburg, Germany

Je nukleárny Beta Catenín špecifický pre dezmoid fibromatózu?

- SFT, Osteosarkóm, Malígny Phyllodes, Liposarkóm, MFH / nediferencovaný sarkóm, Synoviálny Sarkóm, MPNS, ...



Nukleárna pozit. stačí fokálna, ale vnútorná negat. kontrola musí byť negat.
Len cca 75% dezmoid fibromatóz má nukleárny Beta Catenín.

Beta Catenín

- USP6 fúzia aktivuje Wnt dráhu
- USP6 lézie často majú fokálne nukleárne pozit. Beta Catenín

Genetika, patogenéza

- USP 6 (**COL1A1-USP6**, COL3A1-USP6, SERPINH1-USP6, SPARC-USP6, MYH9-USP6), cca v 30% v kraniálnej, 92% v nodulárnej
- COL1A1-CAMTA1
- USP6-CTNNB1
- Wnt / Beta Catenín
- *EWSR-CREB ?*
- *BRAF ?*

Modern Pathology
https://doi.org/10.1038/s41379-019-0422-6

USCAP

ARTICLE

Recurrent and novel *USP6* fusions in cranial fasciitis identified by targeted RNA sequencing

Vera A. Paulson[1,2] · Ivan A. Stojanov[3] · Jay K. Wasman[4] · Tamara Restrepo[1] · Samantha Cano[1] · Joanna Plunkitt[1] · Sekhar Duraisamy[1] · Marian H. Harris[1] · Deborah J. Chute[5] · Alyaa Al-Ibraheemi[1] · Alanna J. Church[1]

Received: 22 August 2019 / Revised: 30 October 2019 / Accepted: 31 October 2019
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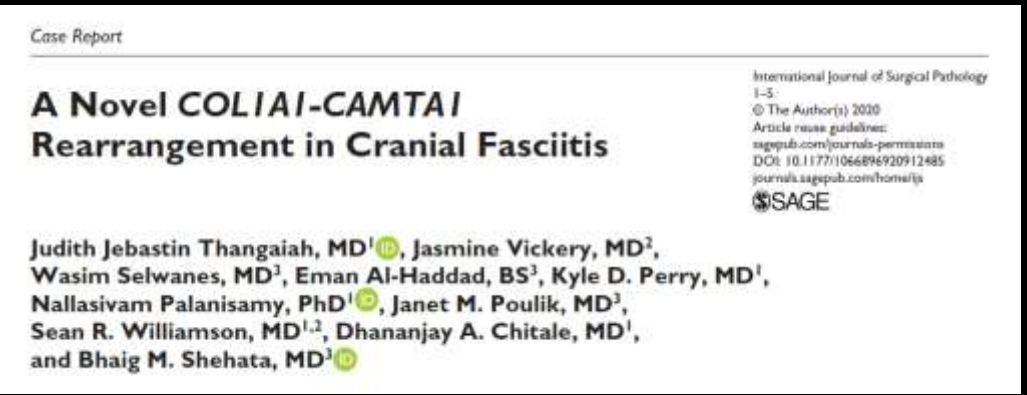

Case Report

A Novel *COL1A1-CAMTA1* Rearrangement in Cranial Fasciitis

International Journal of Surgical Pathology
1–3
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A subset of cranial fasciitis is associated with dysregulation of the Wnt/β-catenin pathway

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BRAF fúzie a bodové mutácie

- PTC, melanóm, glióm
- v mezenchymálnych tumoroch raritné: GIST, glomus tumor, MIFS, metanefrický stromálny tumor a v raritných „infantile sarcoma like“ tumoroch

Kraniálna fascitída, závery

- reaktívna / nádorová (?) fibroblastická – myofibroblastická proliferácia
- v minulosti ako kraniálna varianta nodulárnej fascitídy (len časť)
- postihuje hlavu často s kostným defektom a intrakraniálnou extenziou
- u malých detí
- benígna, bez rekurencie, po inkompletnej resekcii môže regredovať
- hlavný význam – **nezameniť za sarkóm**

Heterogénna skupina

- „normálny genóm“ (reaktívna, potraumatická)
- USP6 (kraniálna nodulárna fascitída)
- COL1A1-CAMTA1
  (blízka nodulárnej fascitíde?)
- Wnt / Beta Catenín
  (myxoidná dezmoid fibromatóza?)
- EWSR-CREB intracranial myxoid mesenchymal tumor (myxoidný intrakraniálny angiomatoidný fibrózny histiocytóm?)
- ***BRAF V600 (ešte nikto!)***

ďakujem za pozornosť