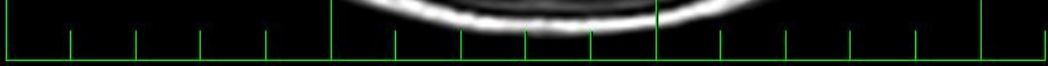
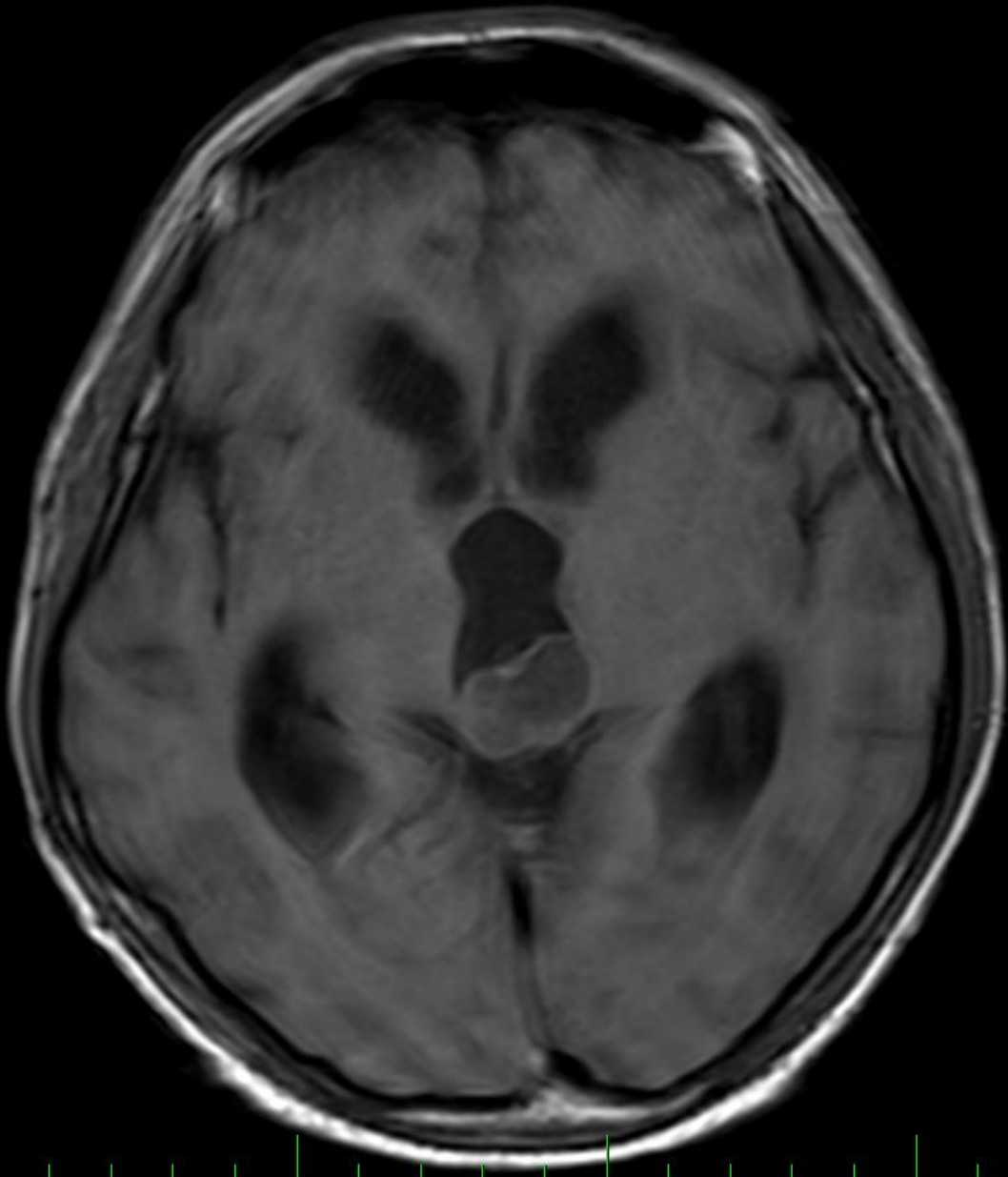


**muž 49 let**

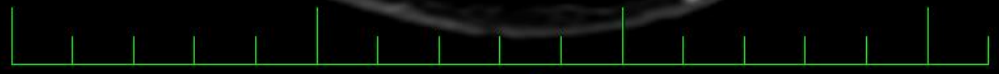
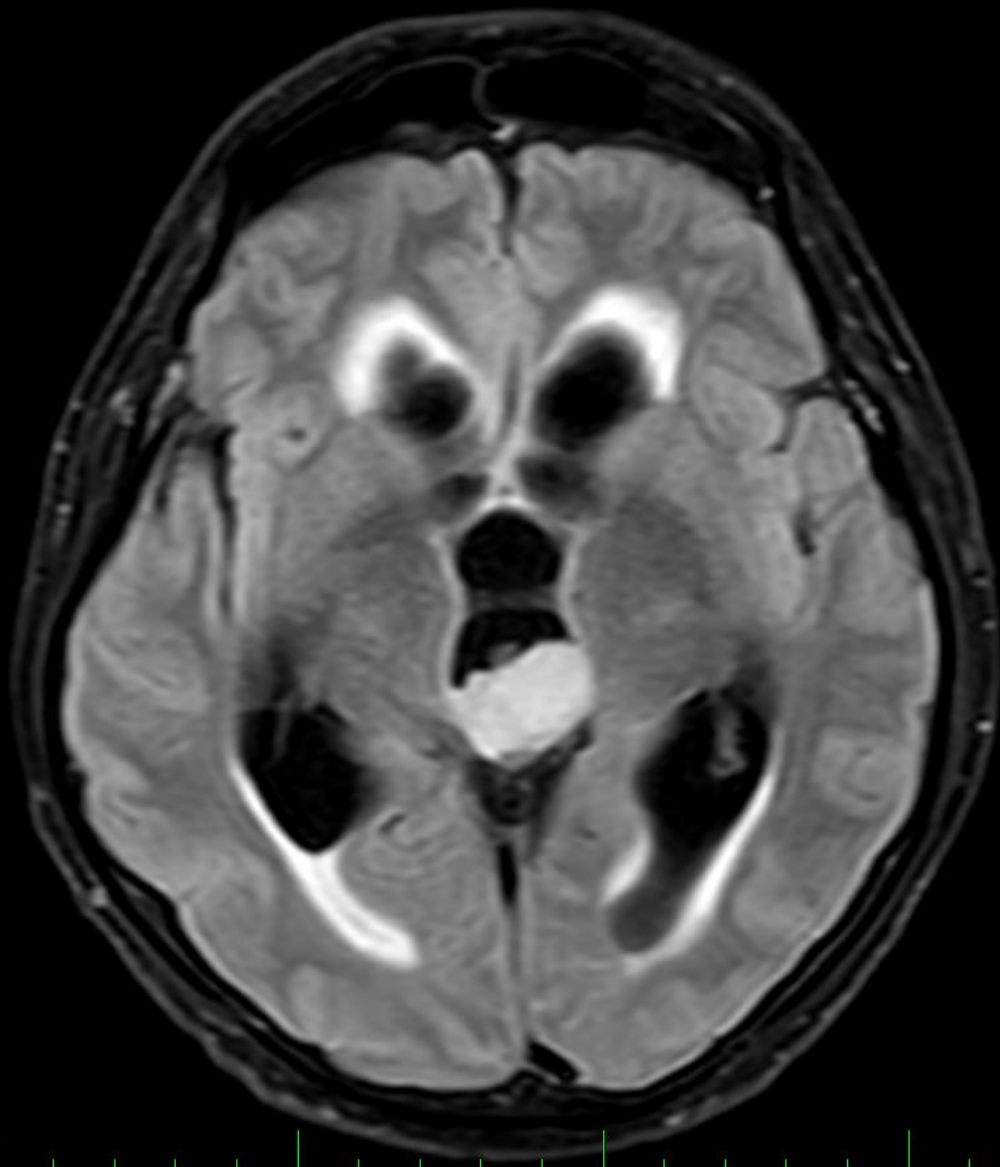
MRI:

- cystický tumor gl. pinealis vel. cca 24x16mm, hypersignalni v T2 i FLAIR, naznacene septovany v T2, postkontrastne se sytici v periferii cysticek.
- supratentorialni hydrocefalus se znamkami dekompenzace,

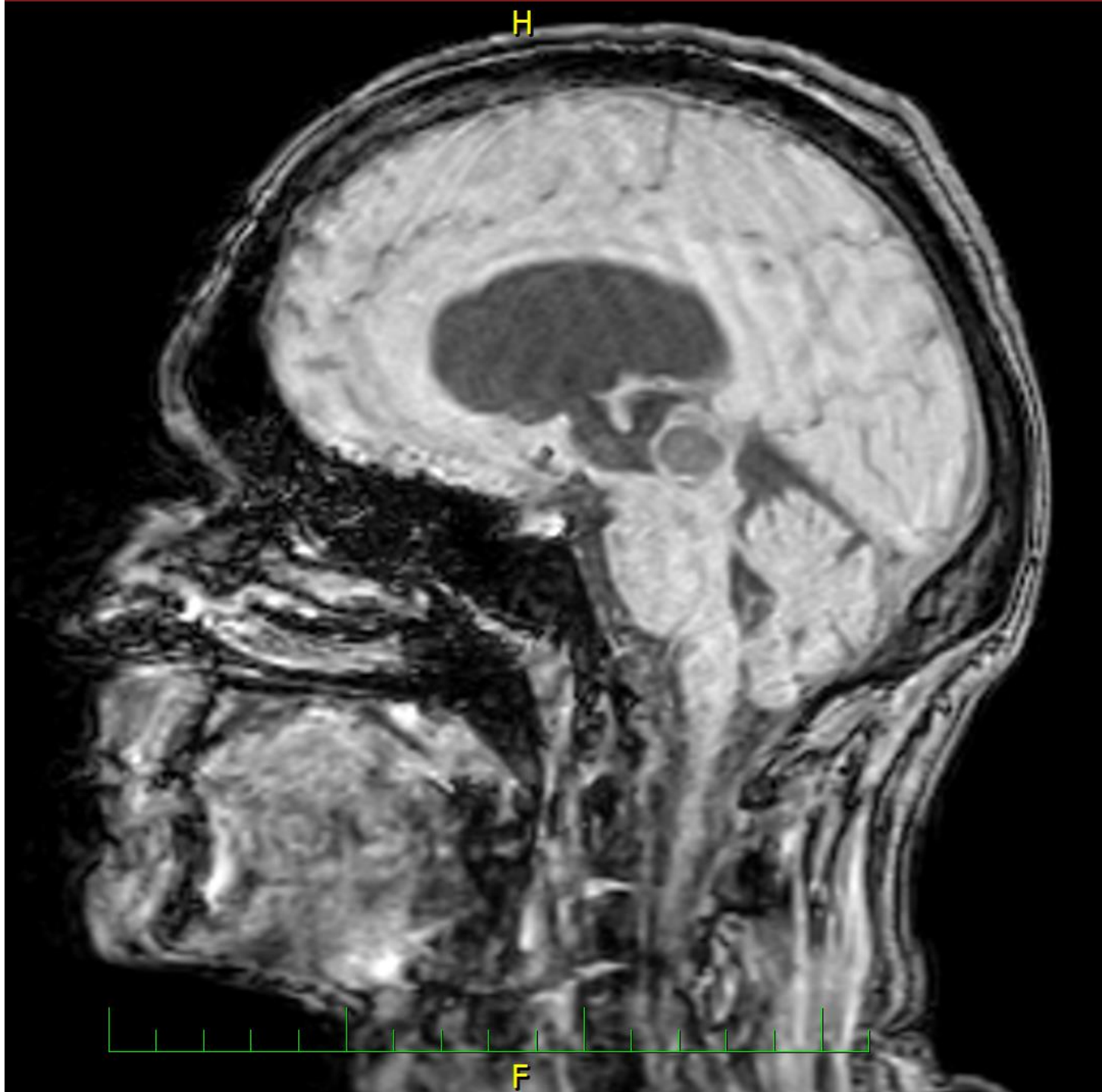


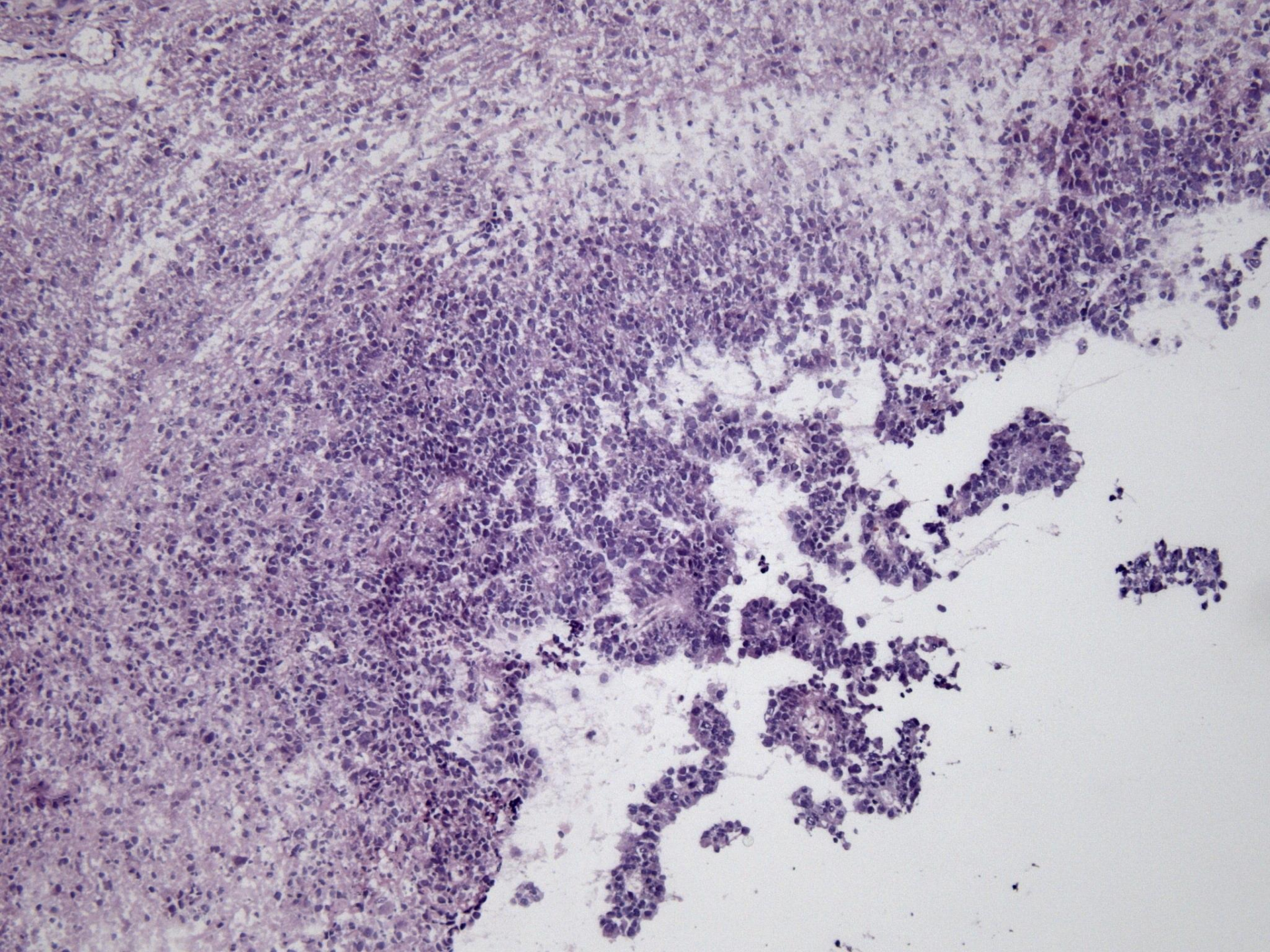
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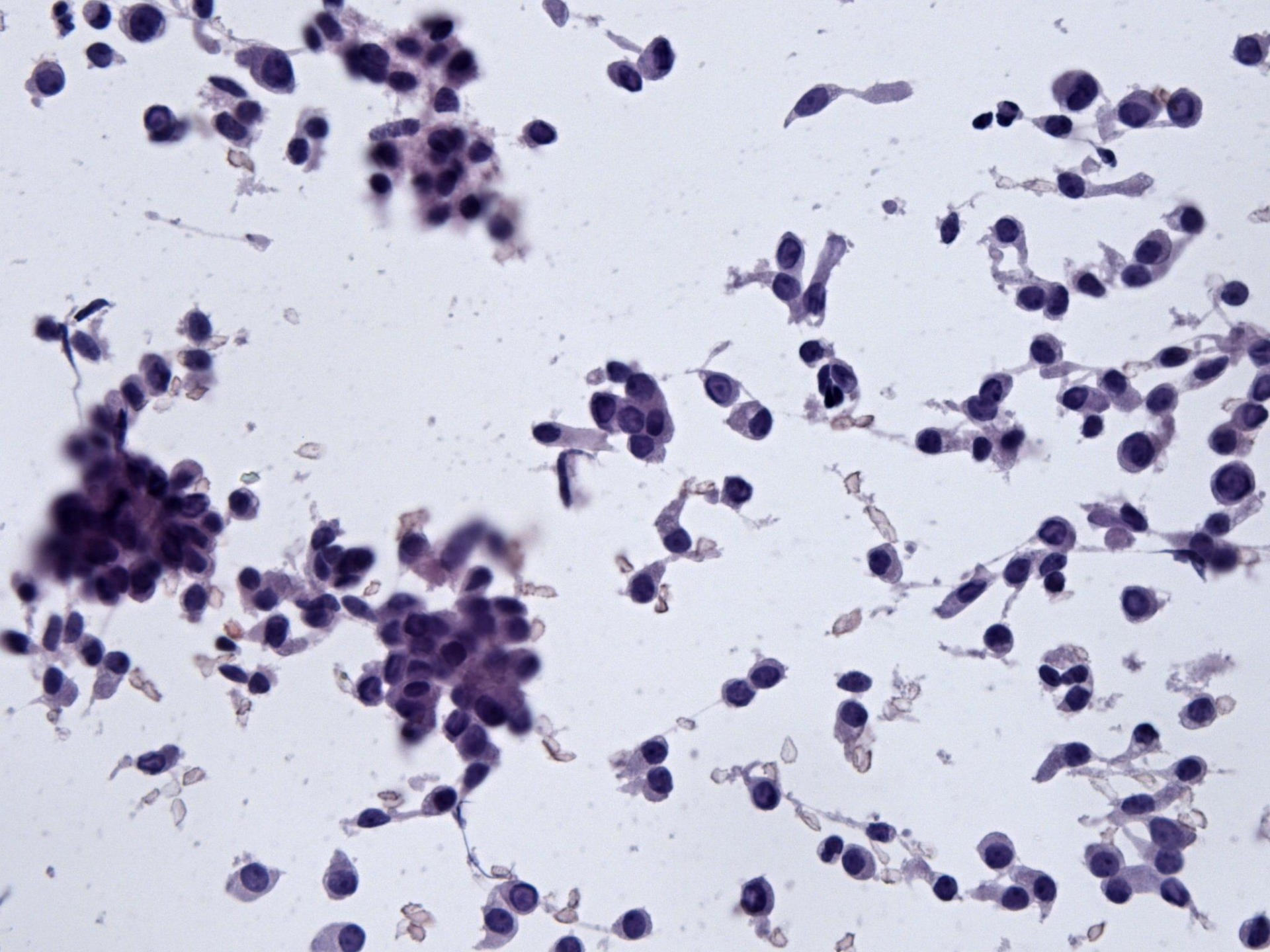
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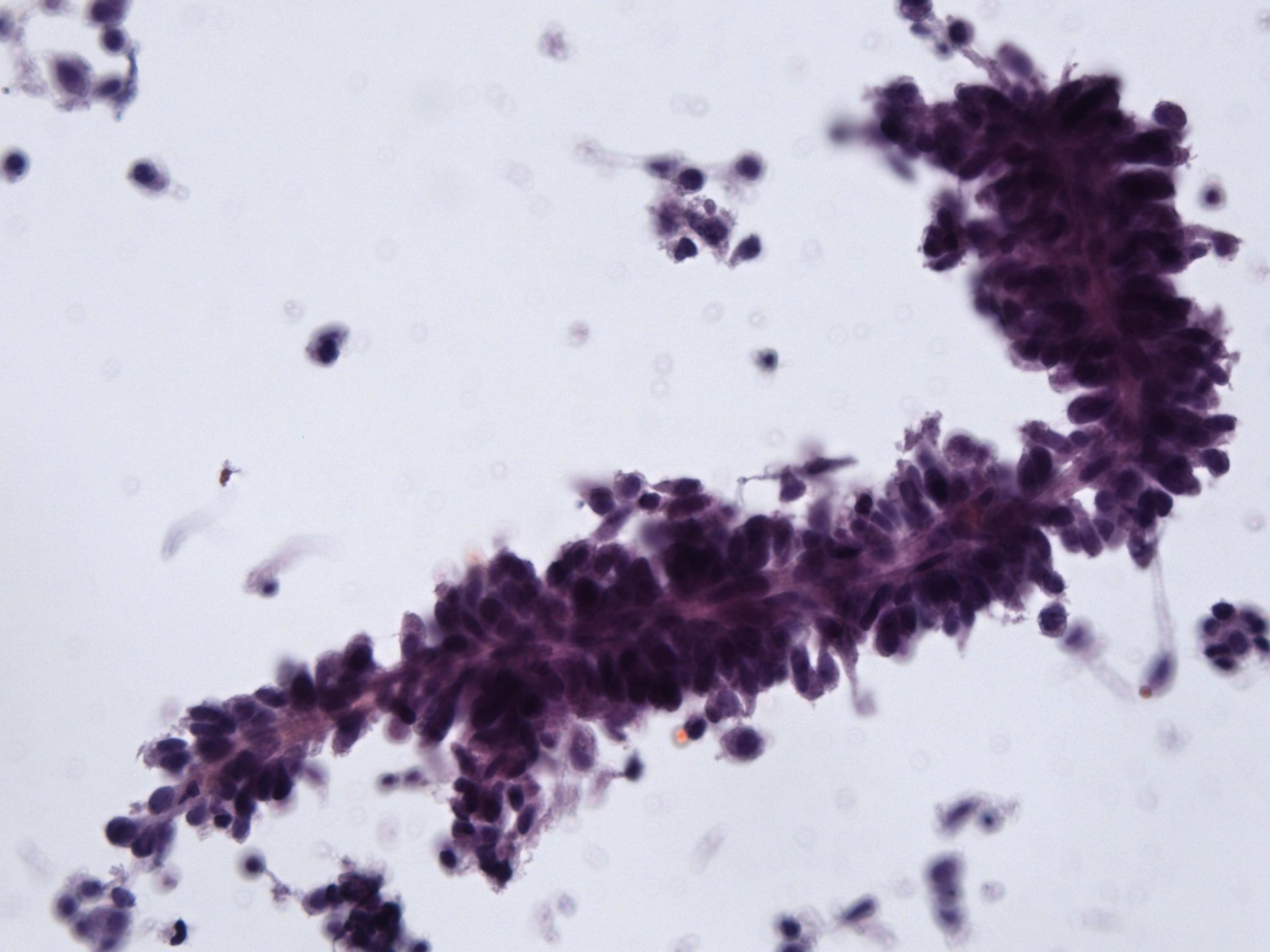


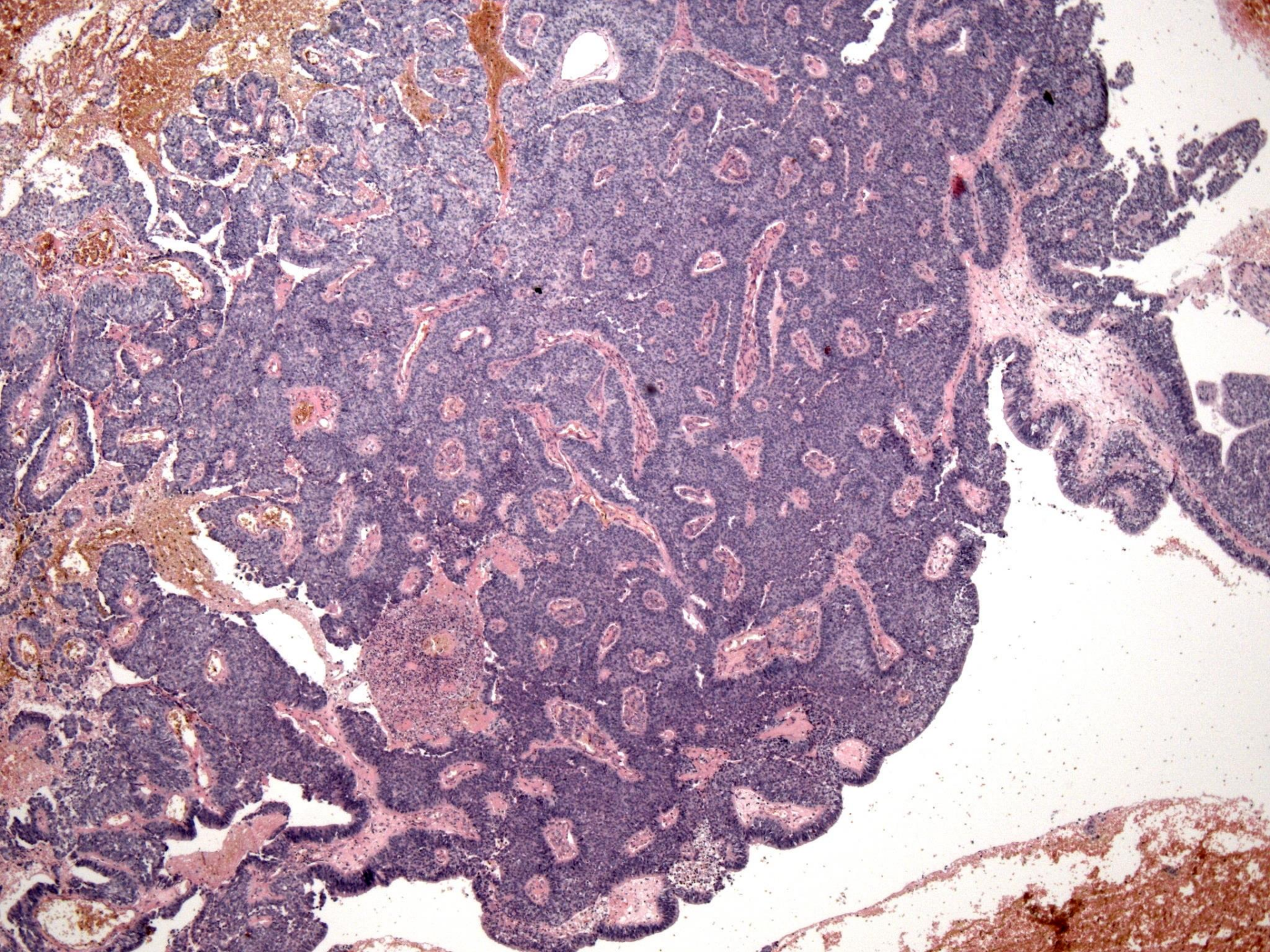
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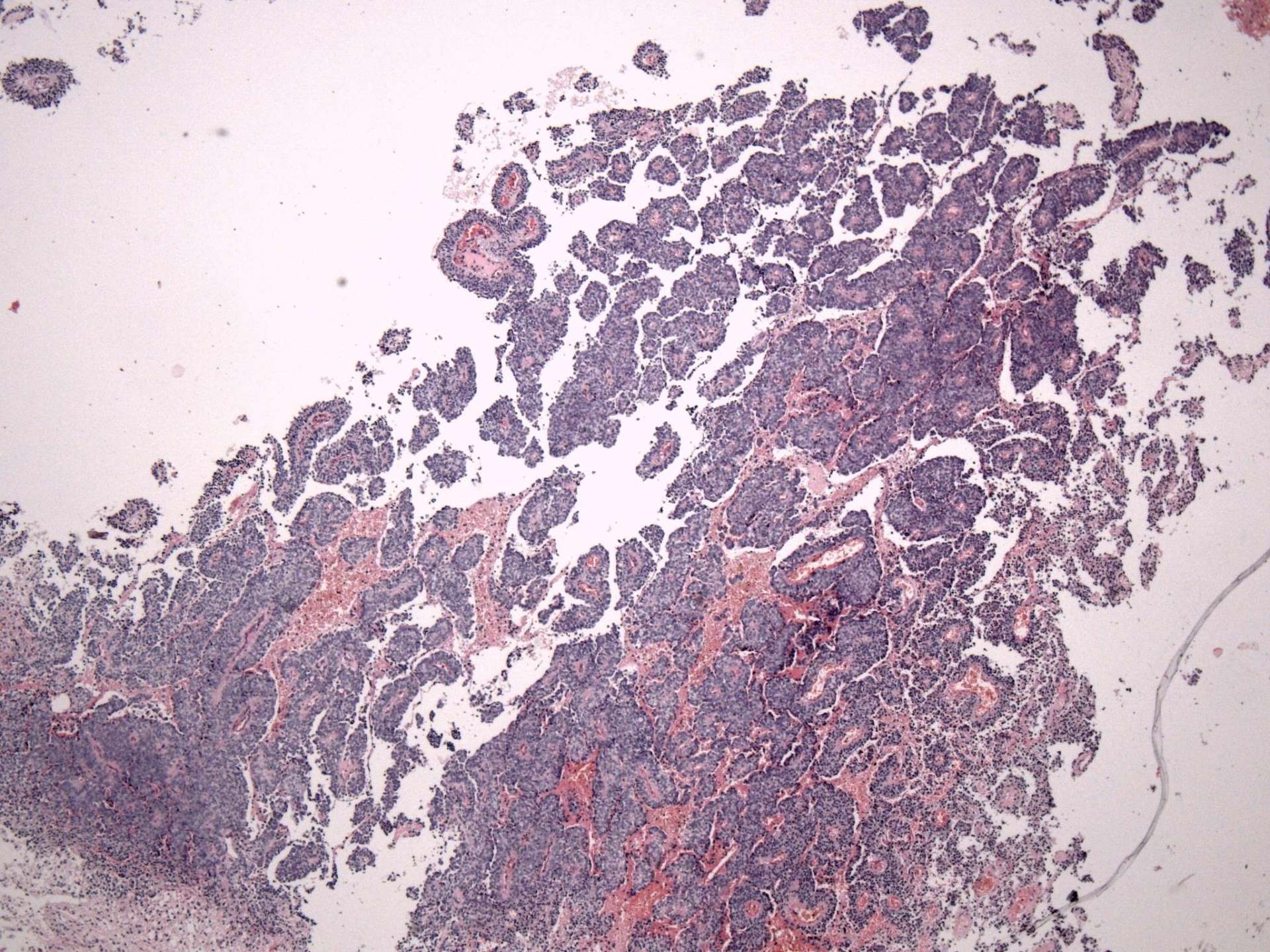


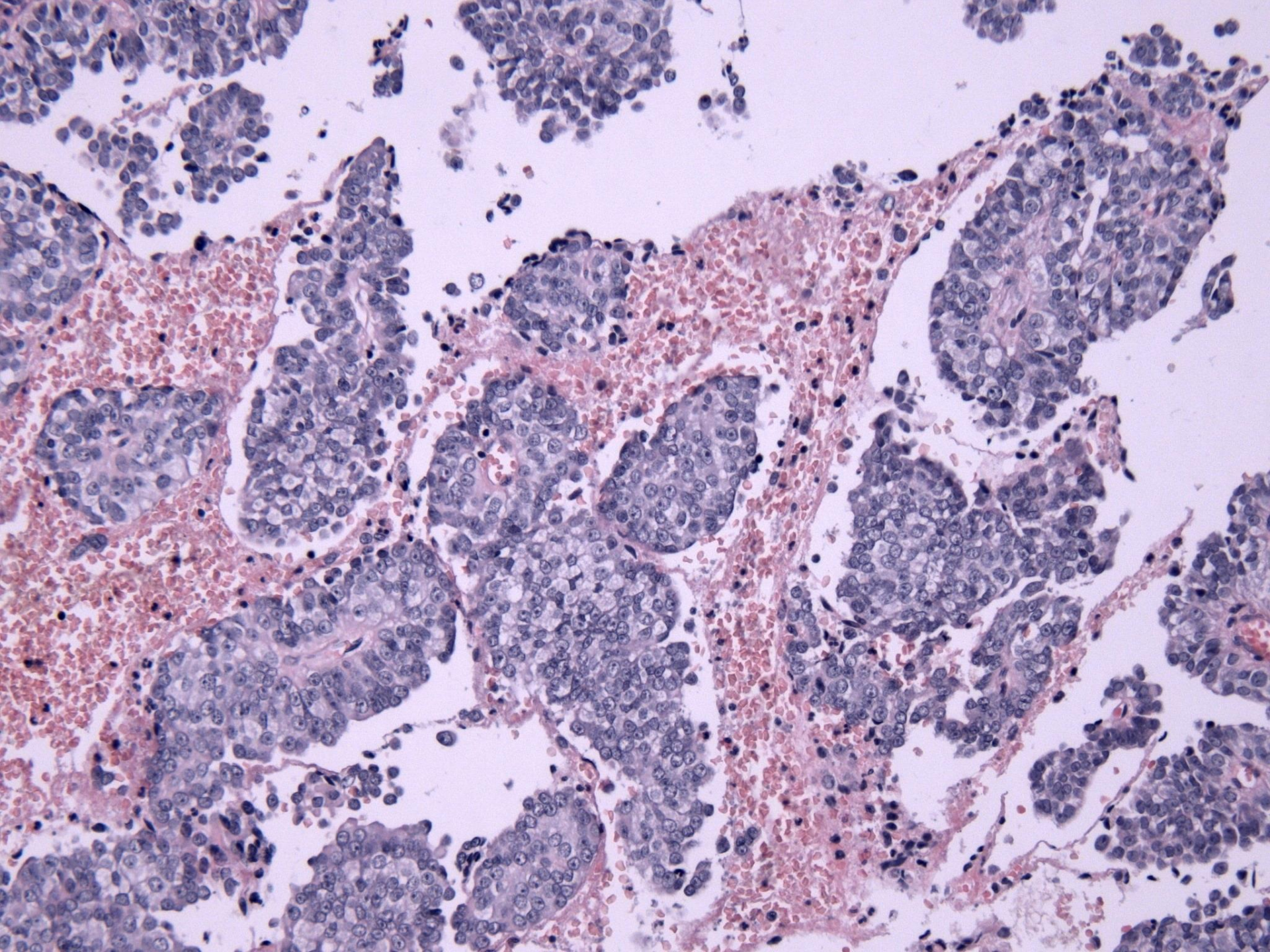


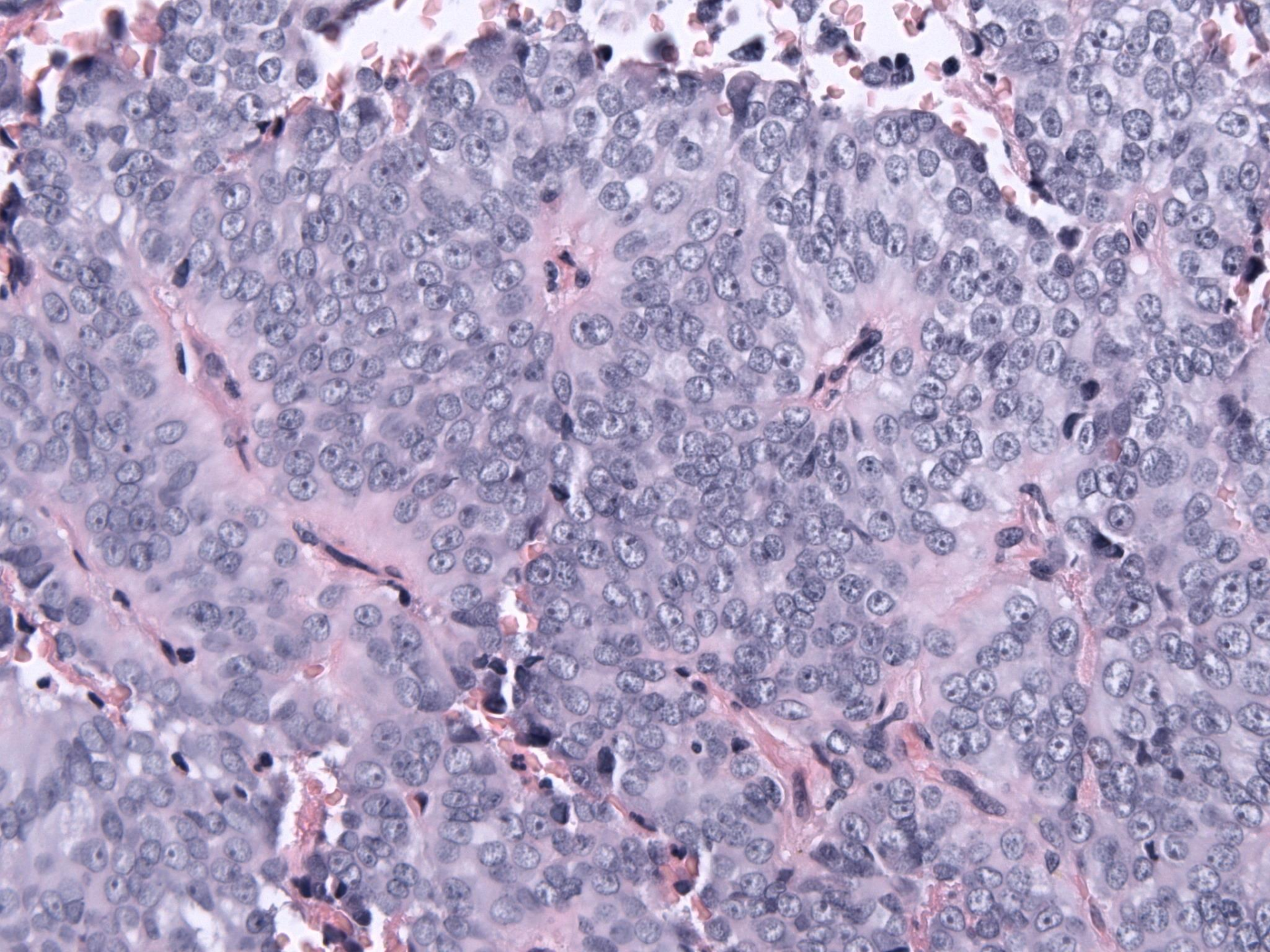


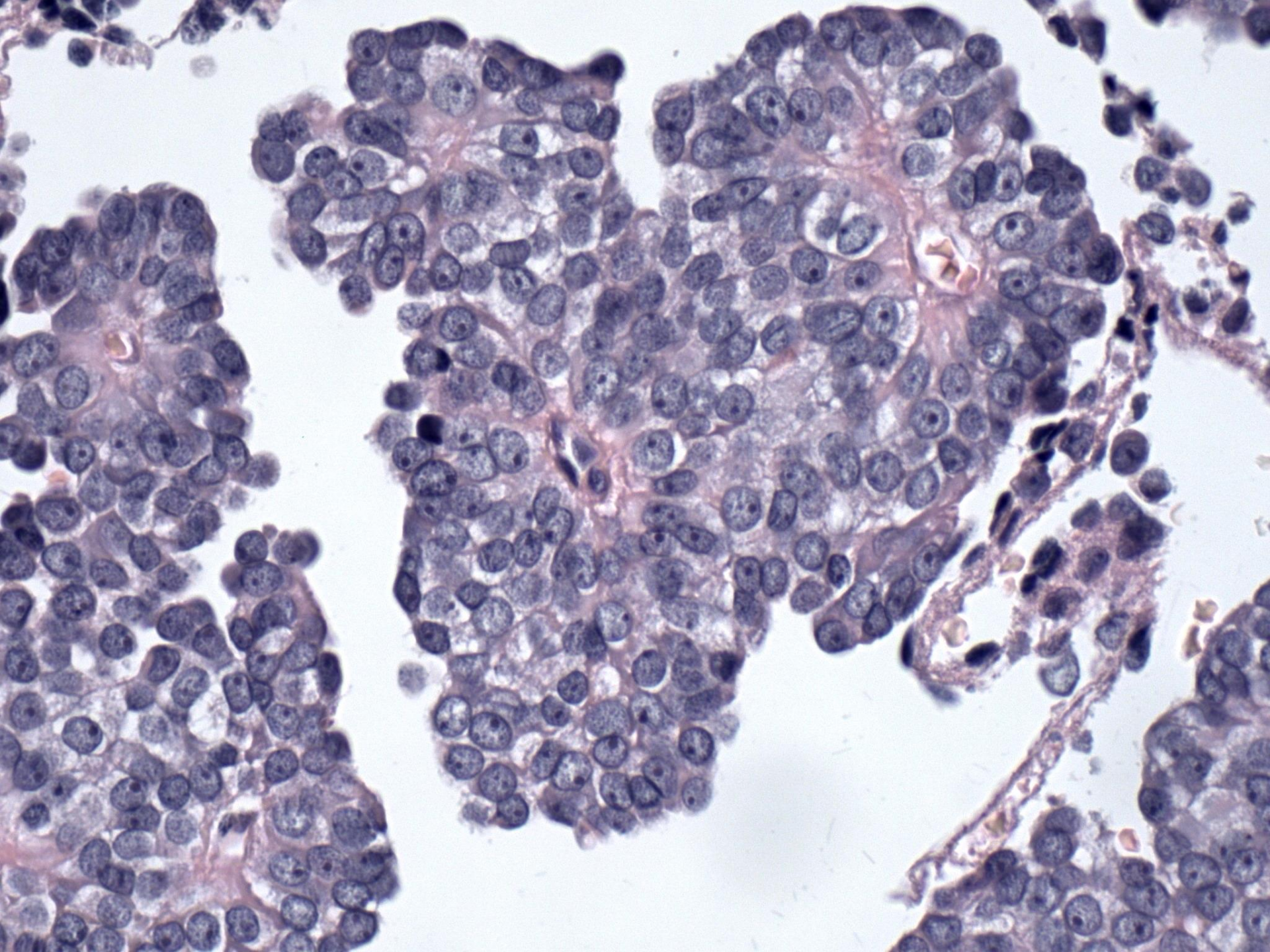


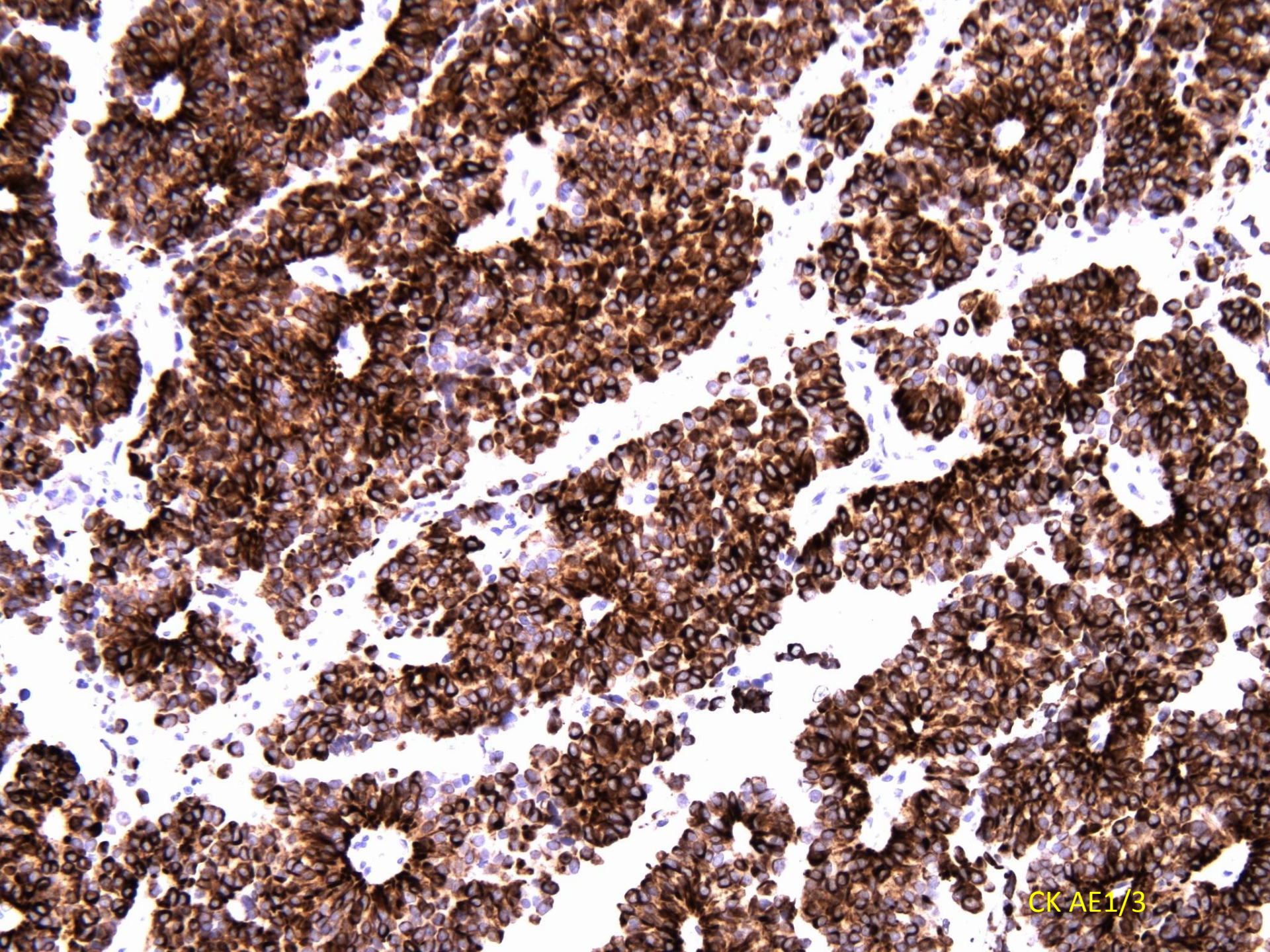




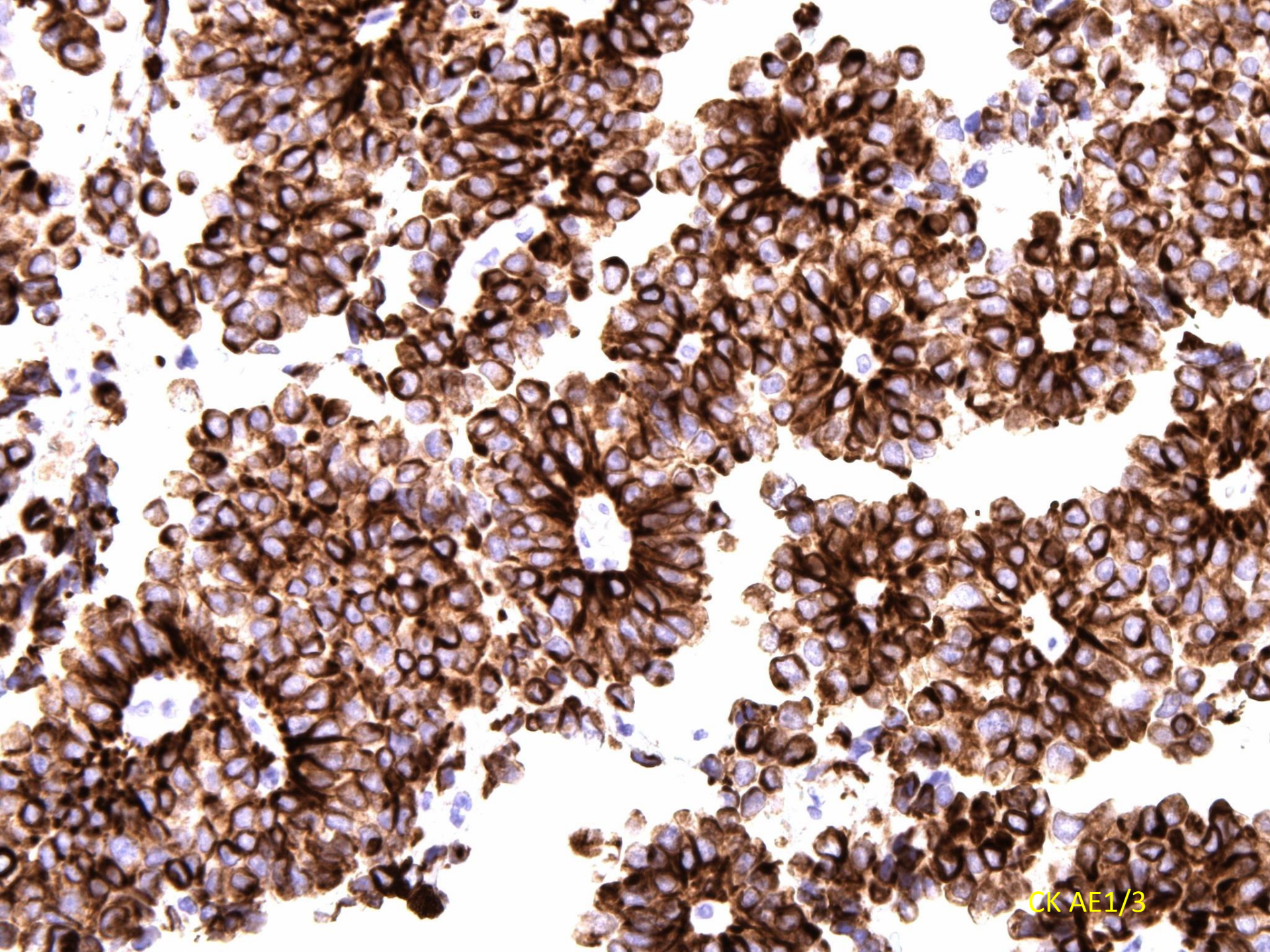




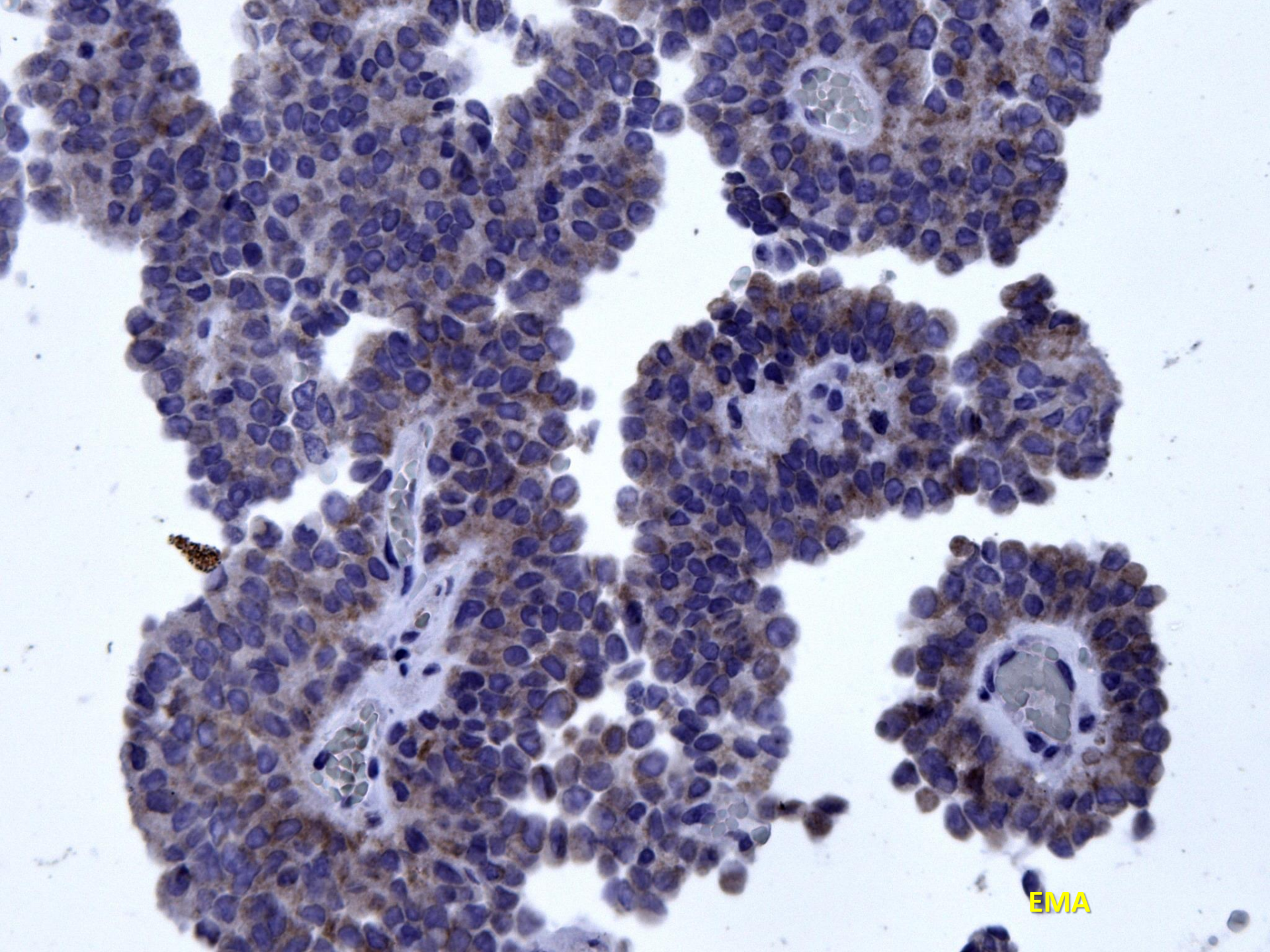




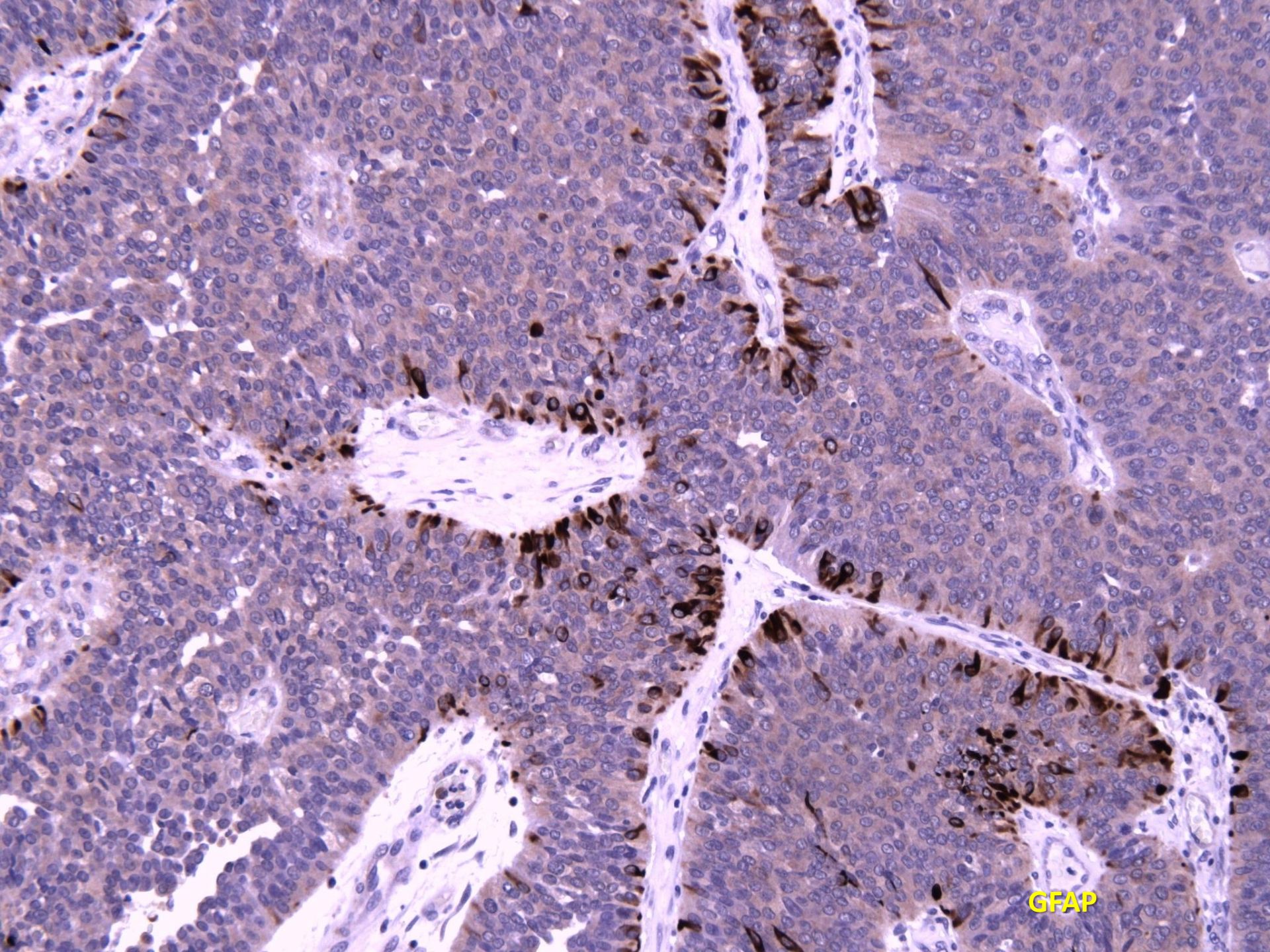
CK AE1/3



CK AE1/3

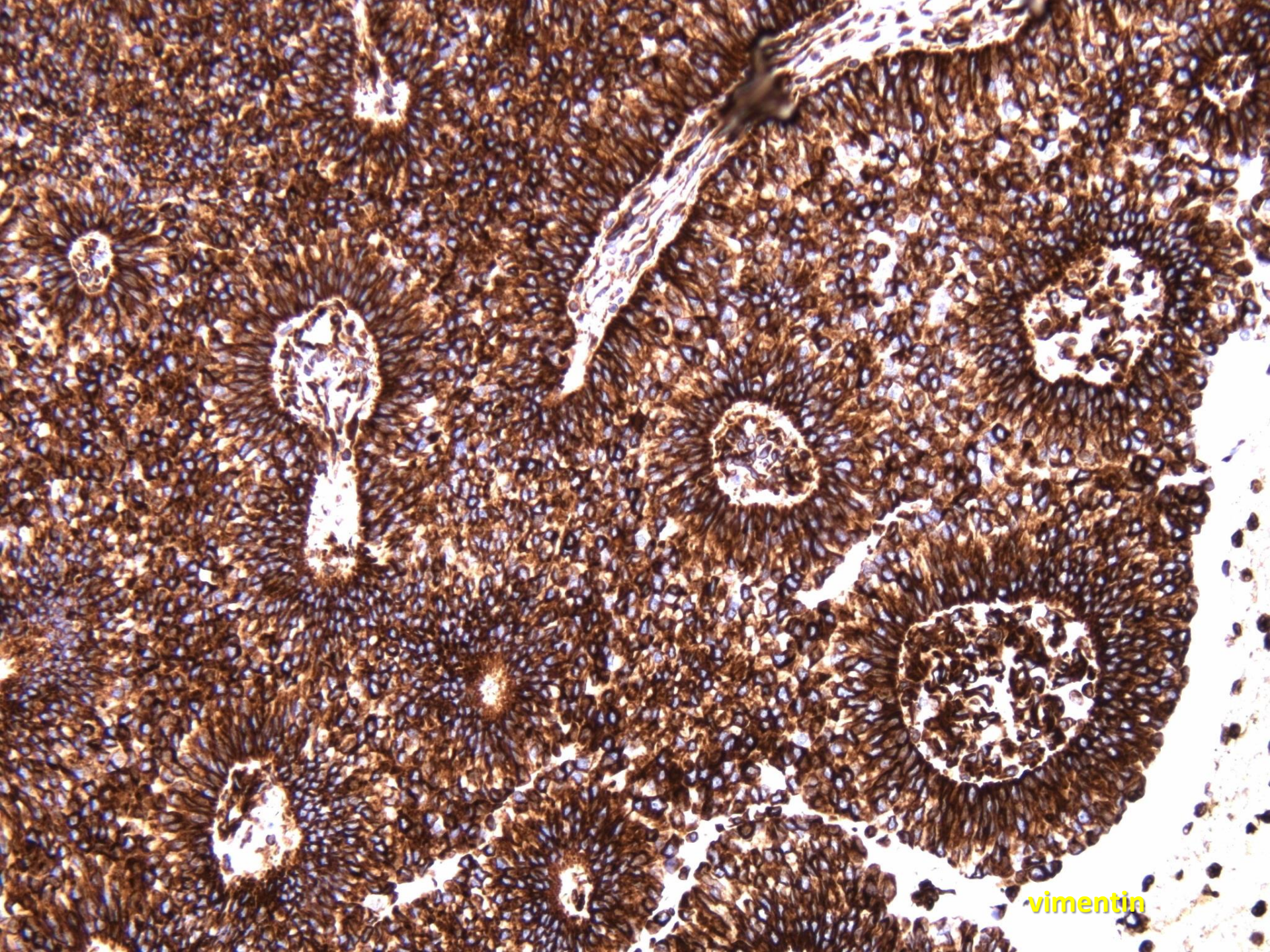


EMA

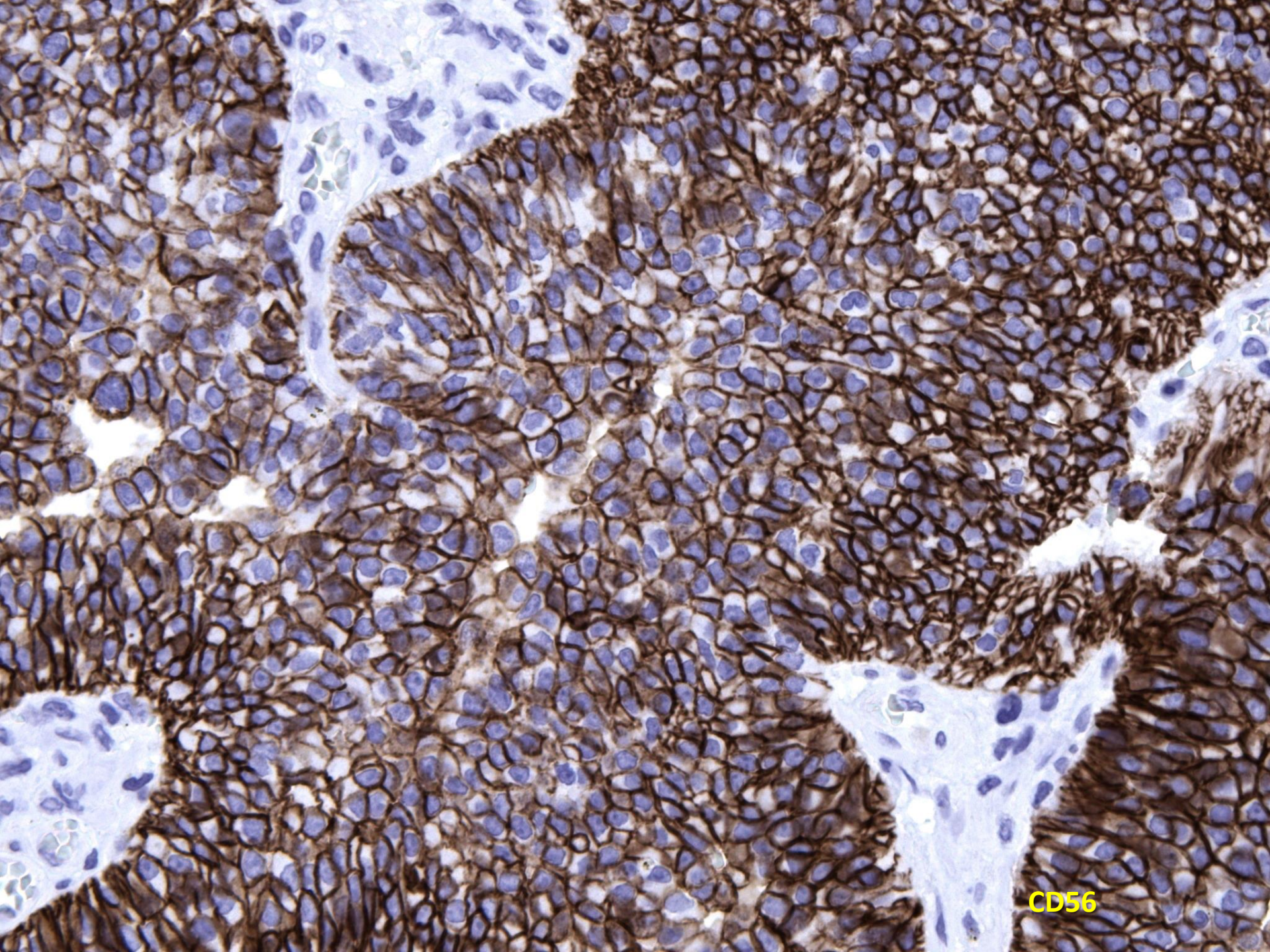


GFAP

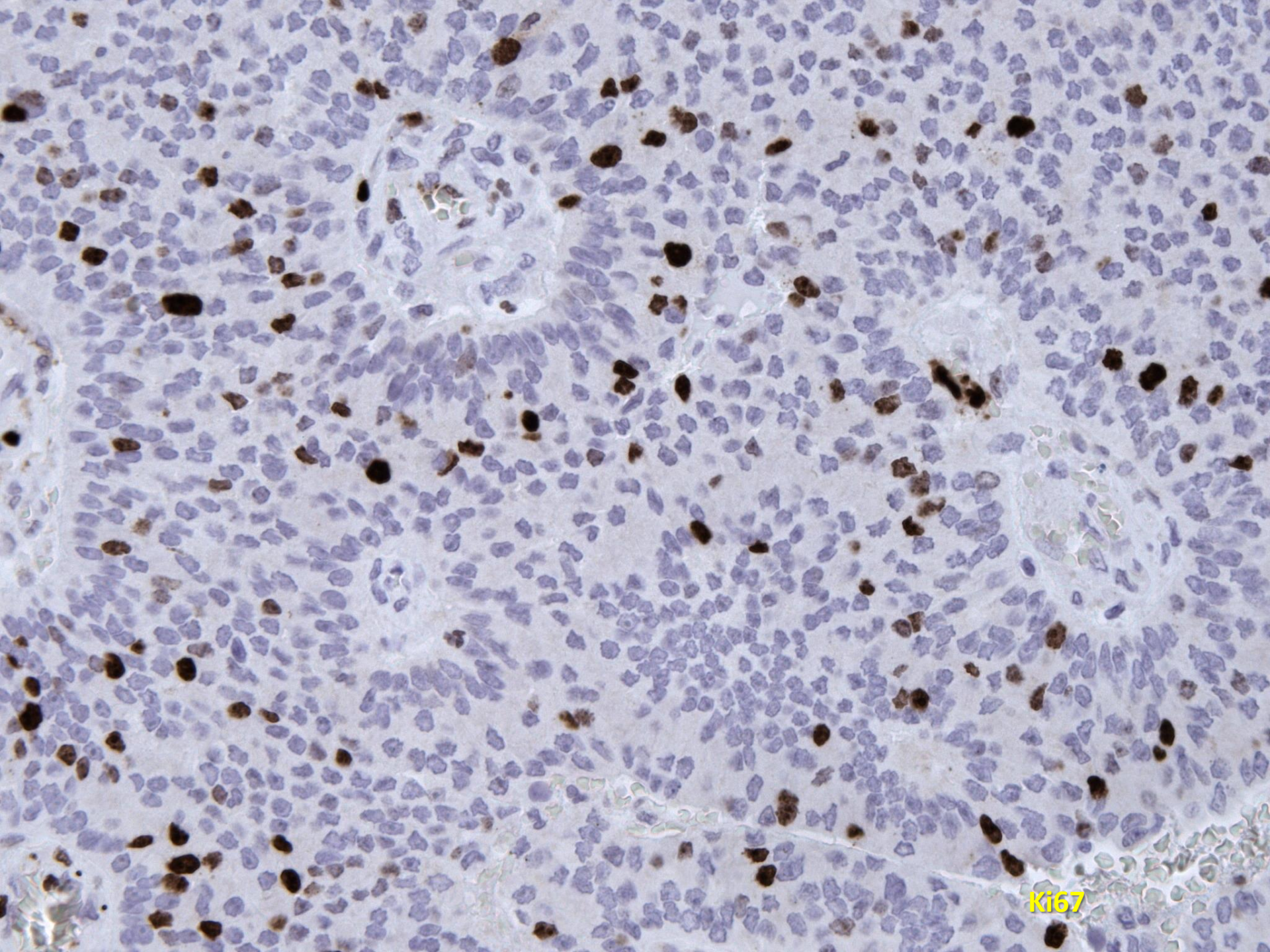




vimentin



CD56



Ki67

Diagnóza:

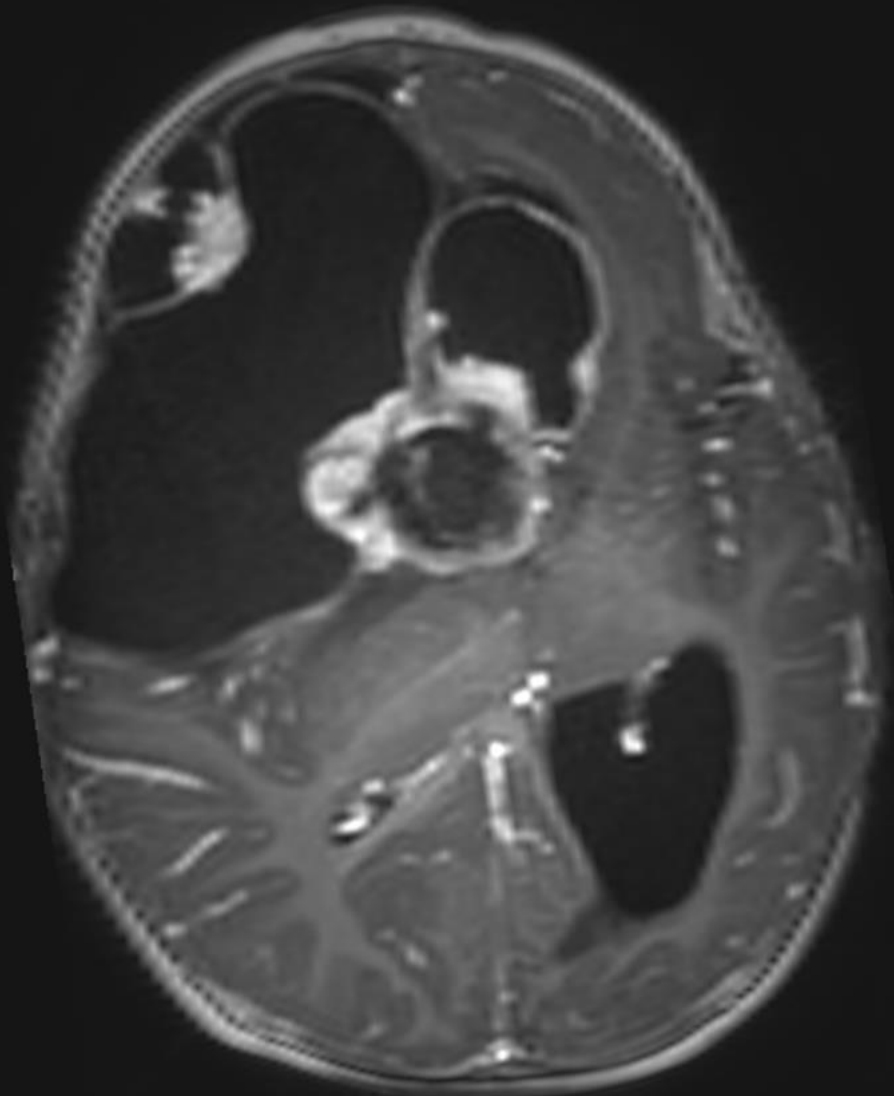
## **PAPILÁRNÍ NÁDOR PINEÁLNÍ OBLASTI**

**Ve WHO klasifikace není přesně stanovený systém gradingu pro tento nádor (grade II-III), vzhledem k atypiím a zvýšené proliferační aktivitě však nelze vyloučit recidivující průběh a nádor bychom tedy hodnotili spíše jako grade III.**

**13 M, chlapec**

Objemná expanze hypotalamu solidně cystická  
dx. s objemnou kalcifikací

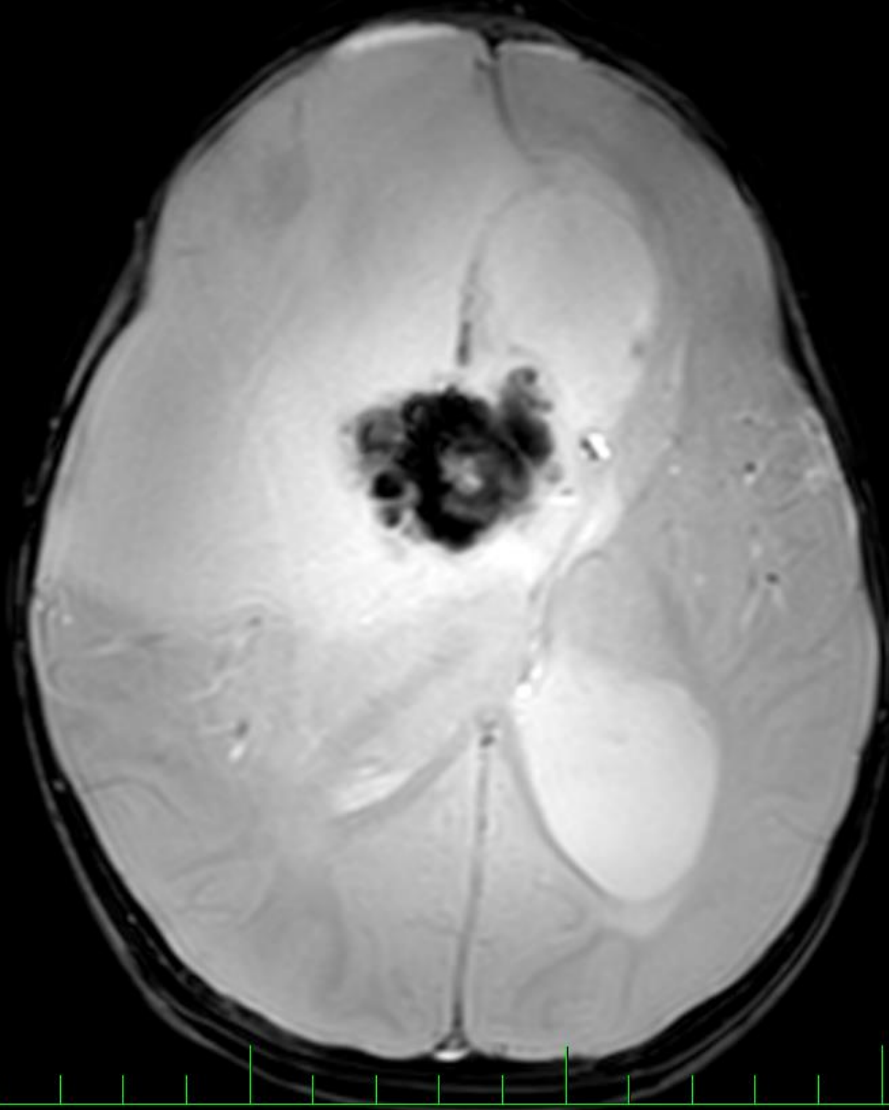
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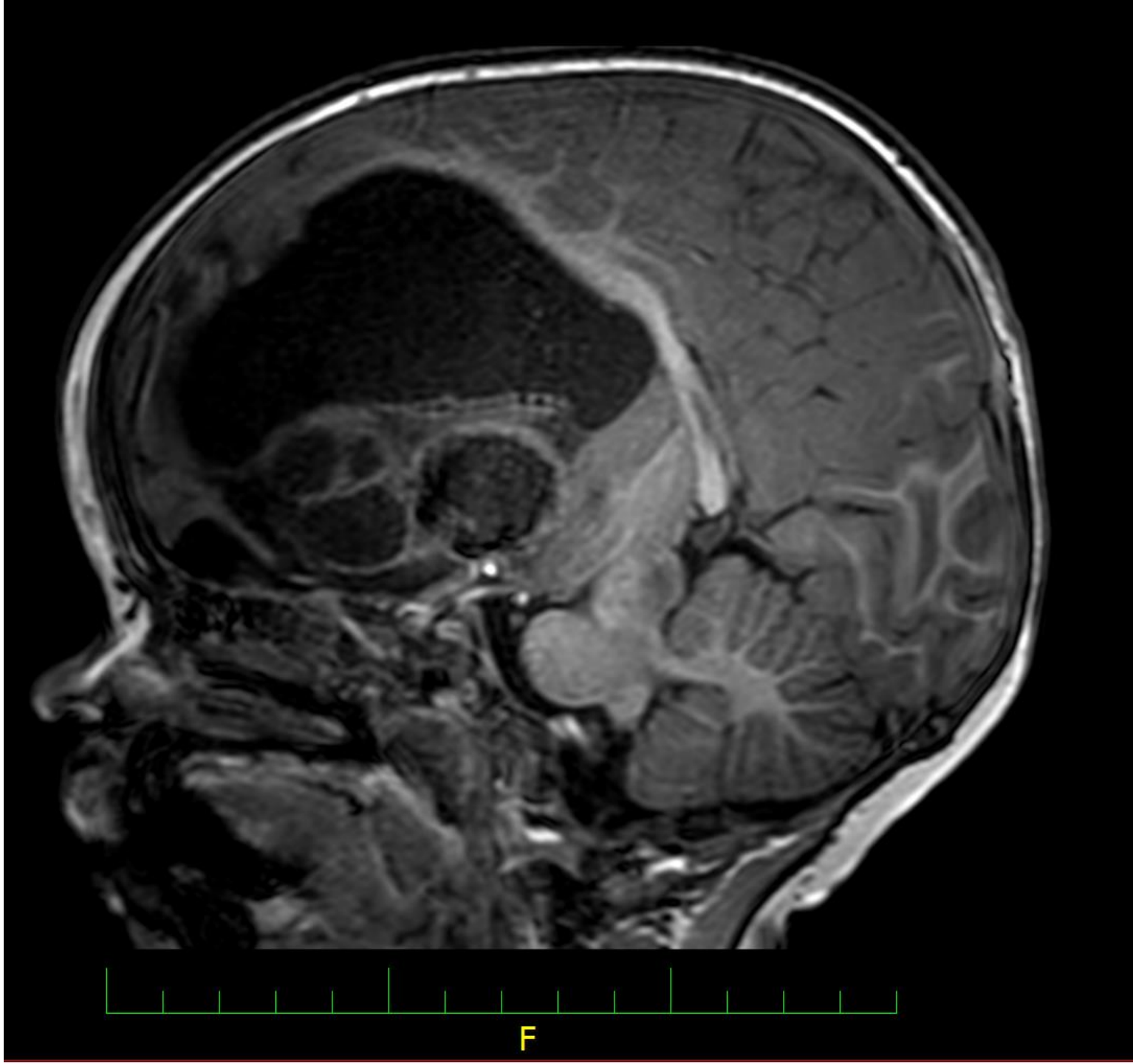
R F

P

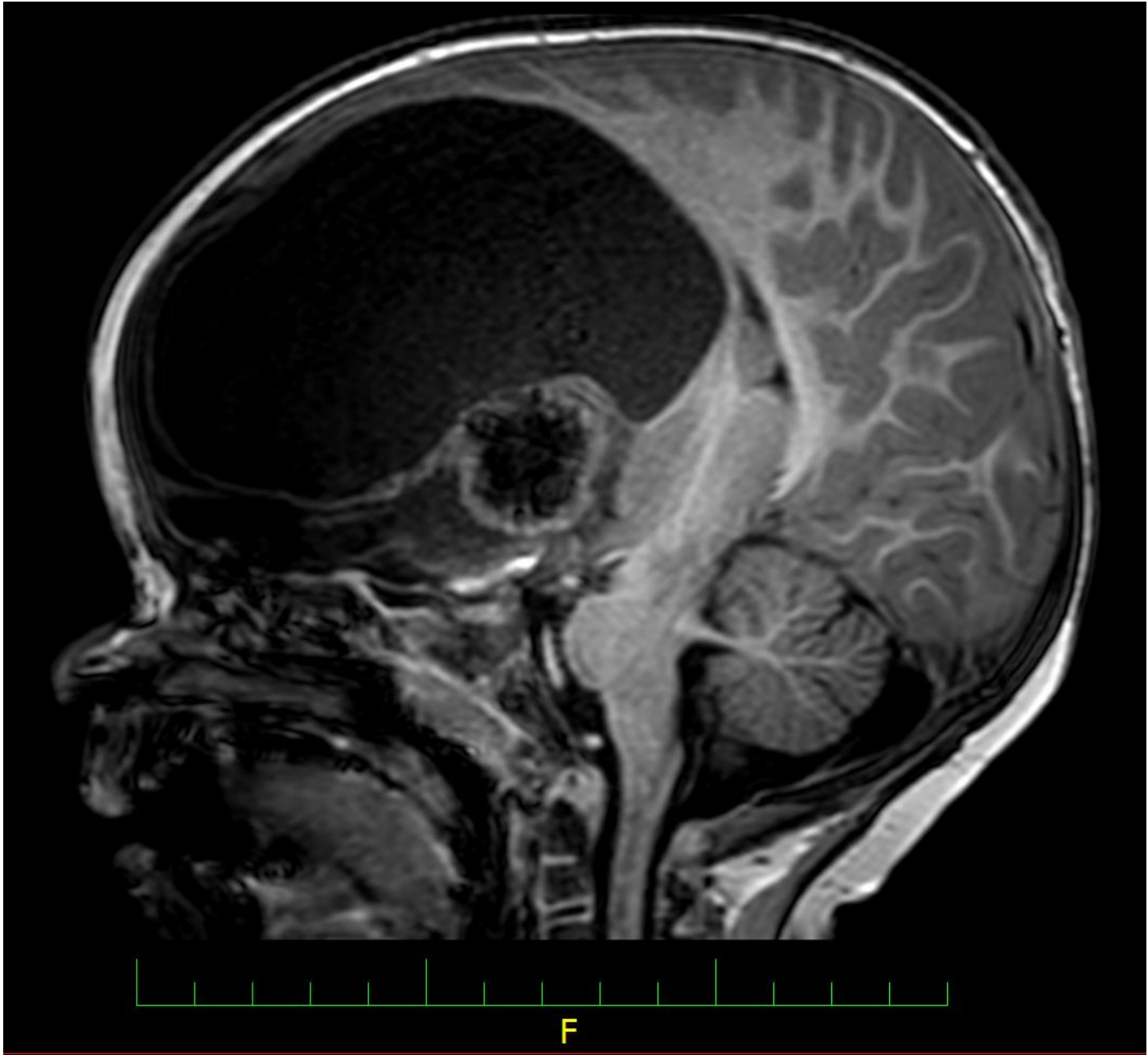
A



P

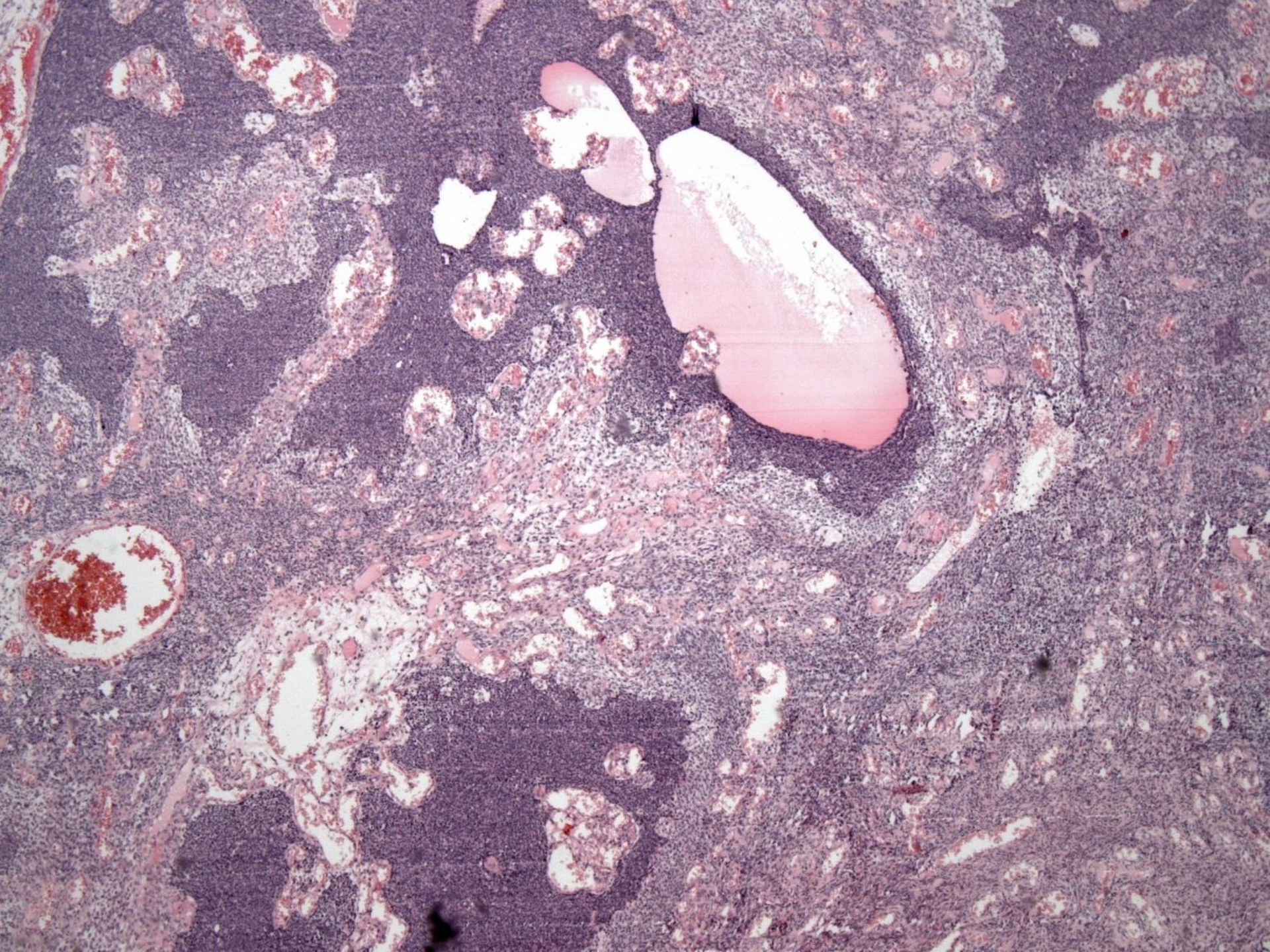


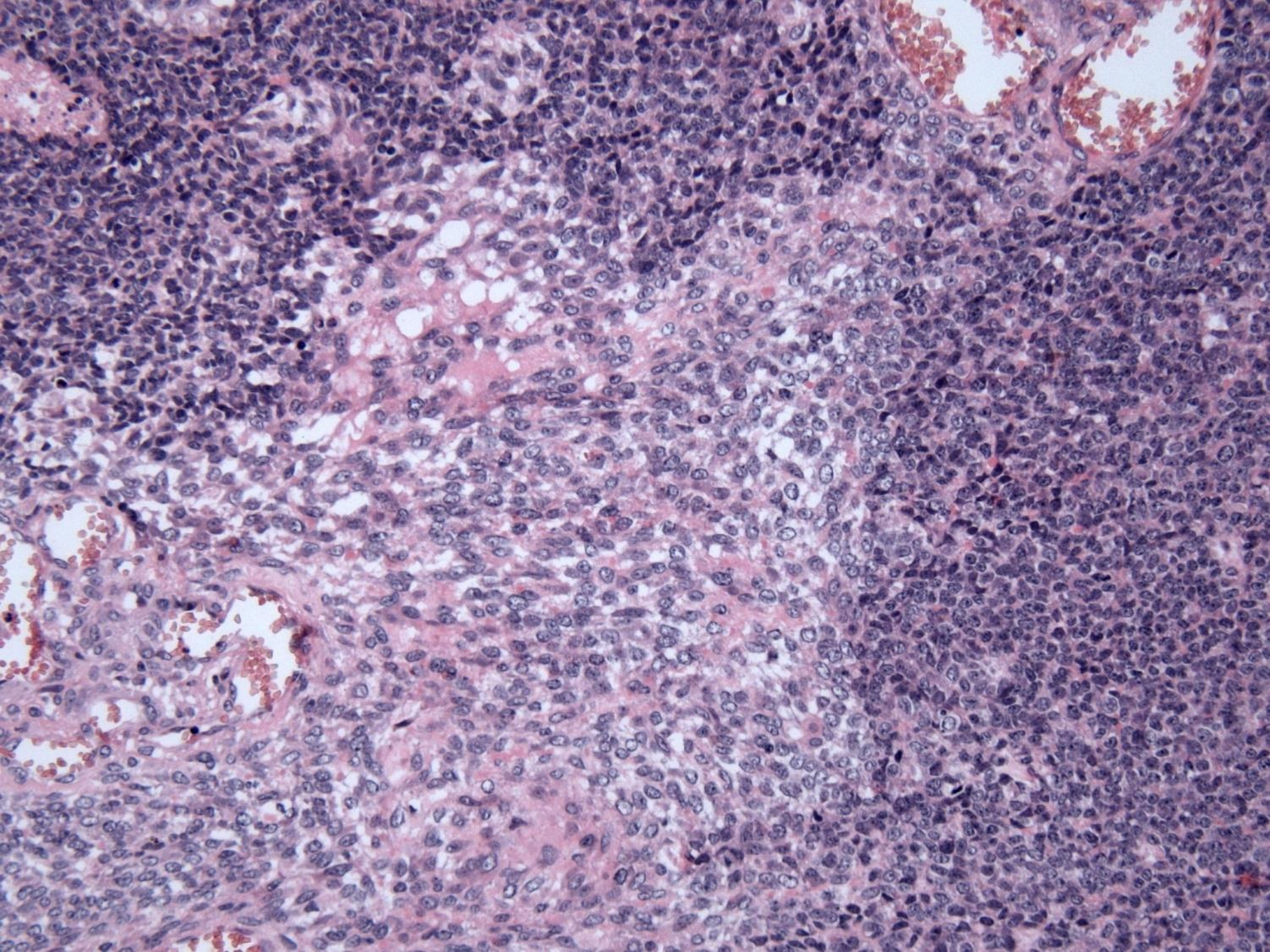


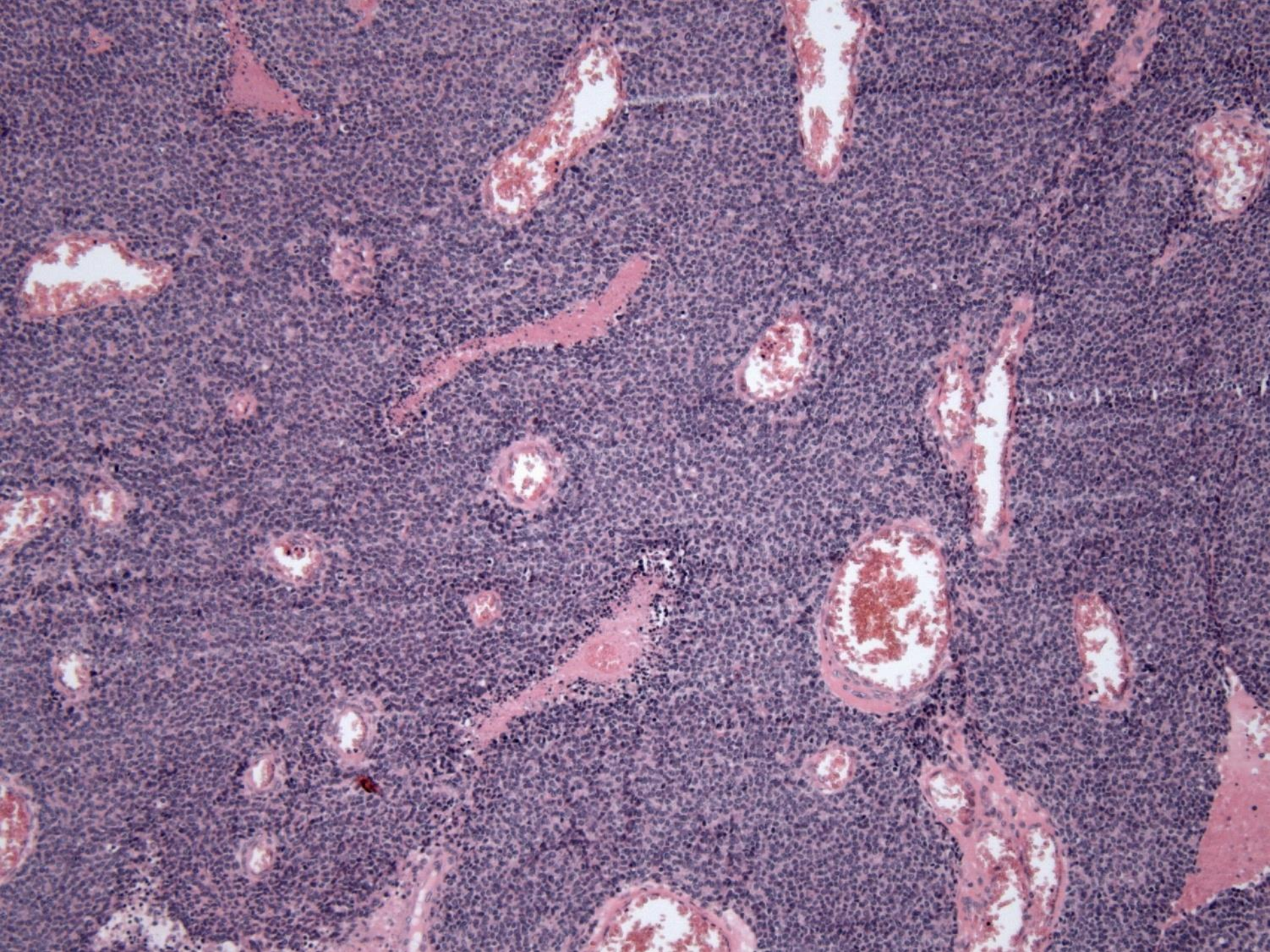


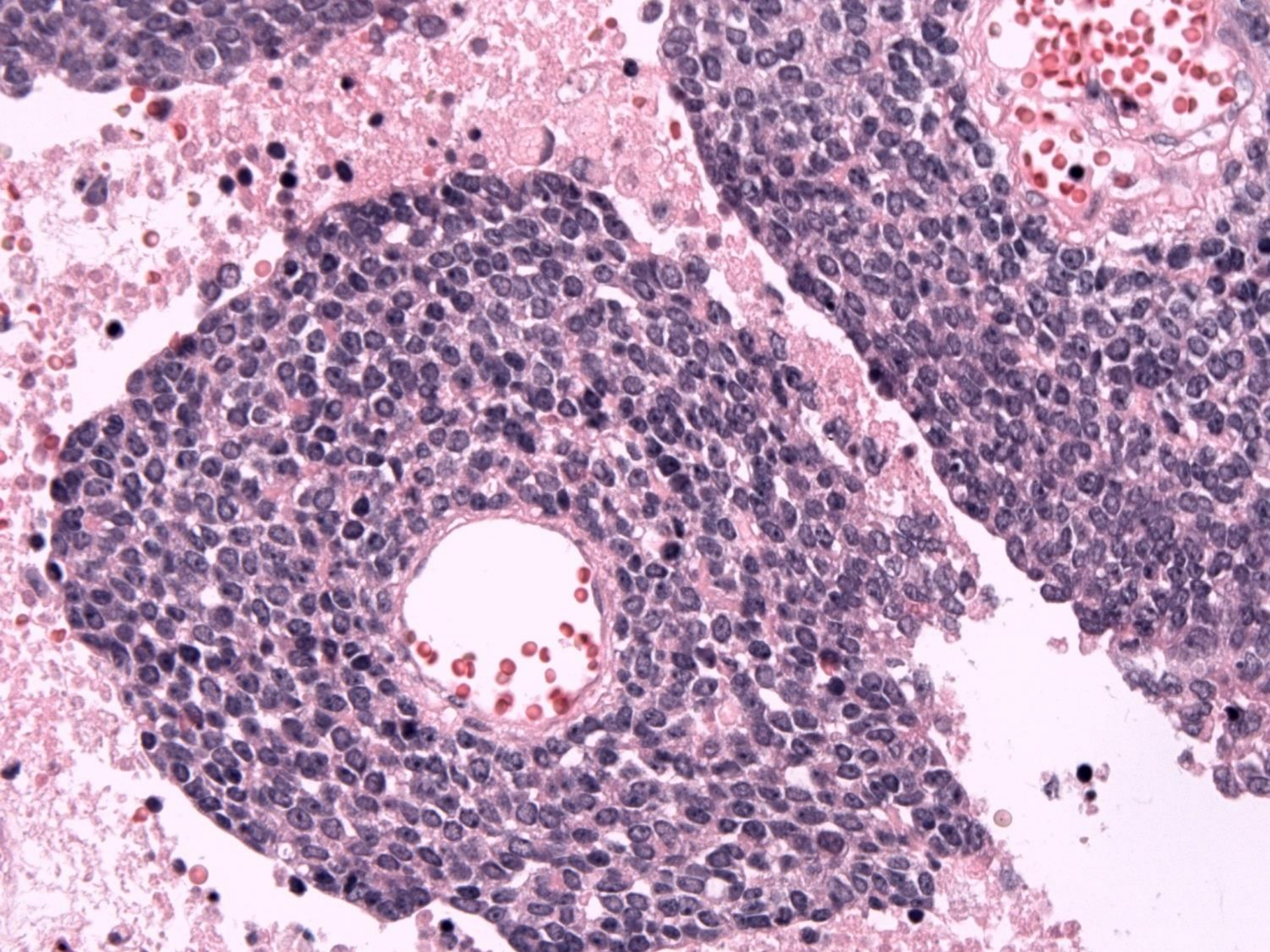
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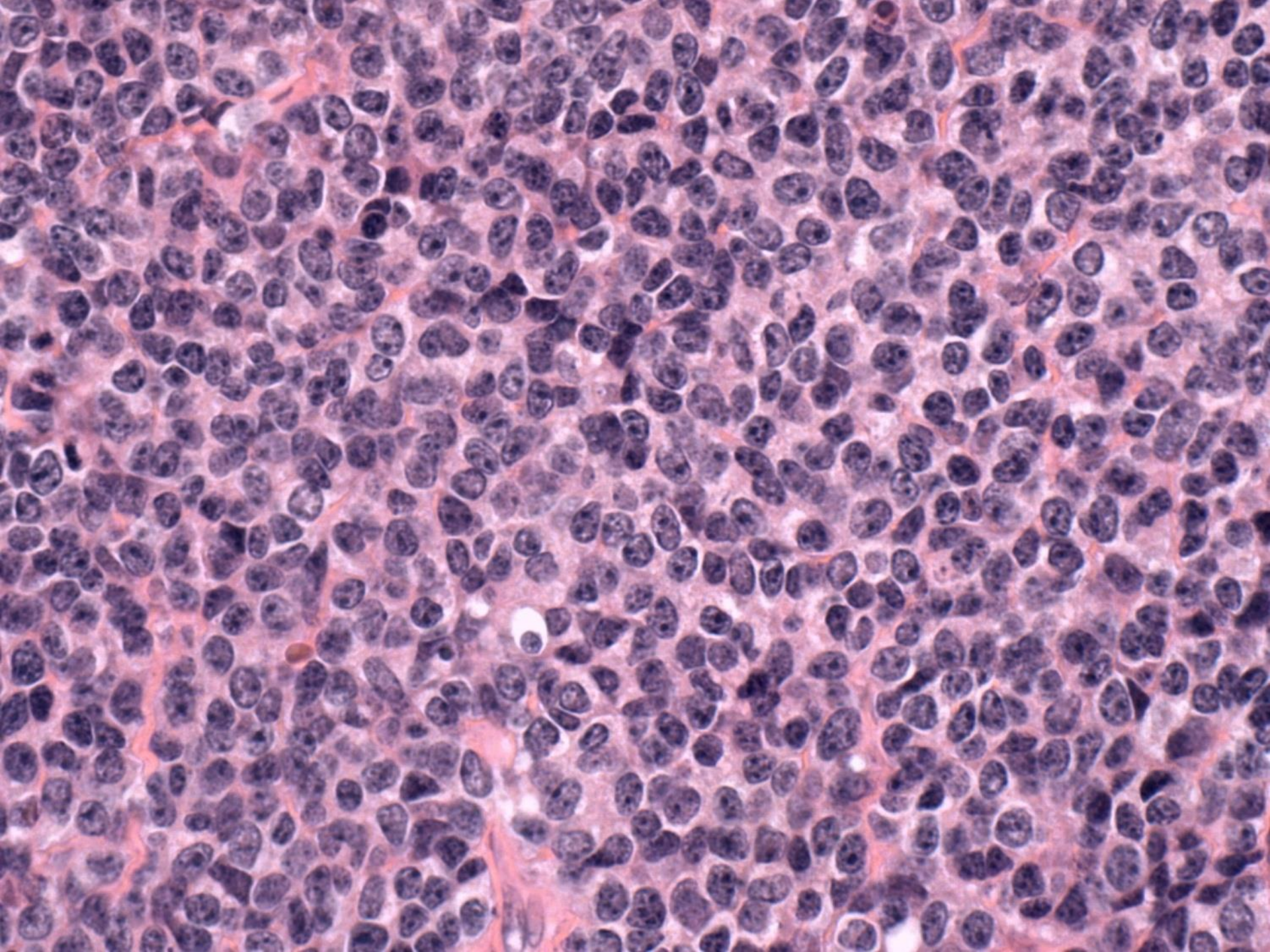


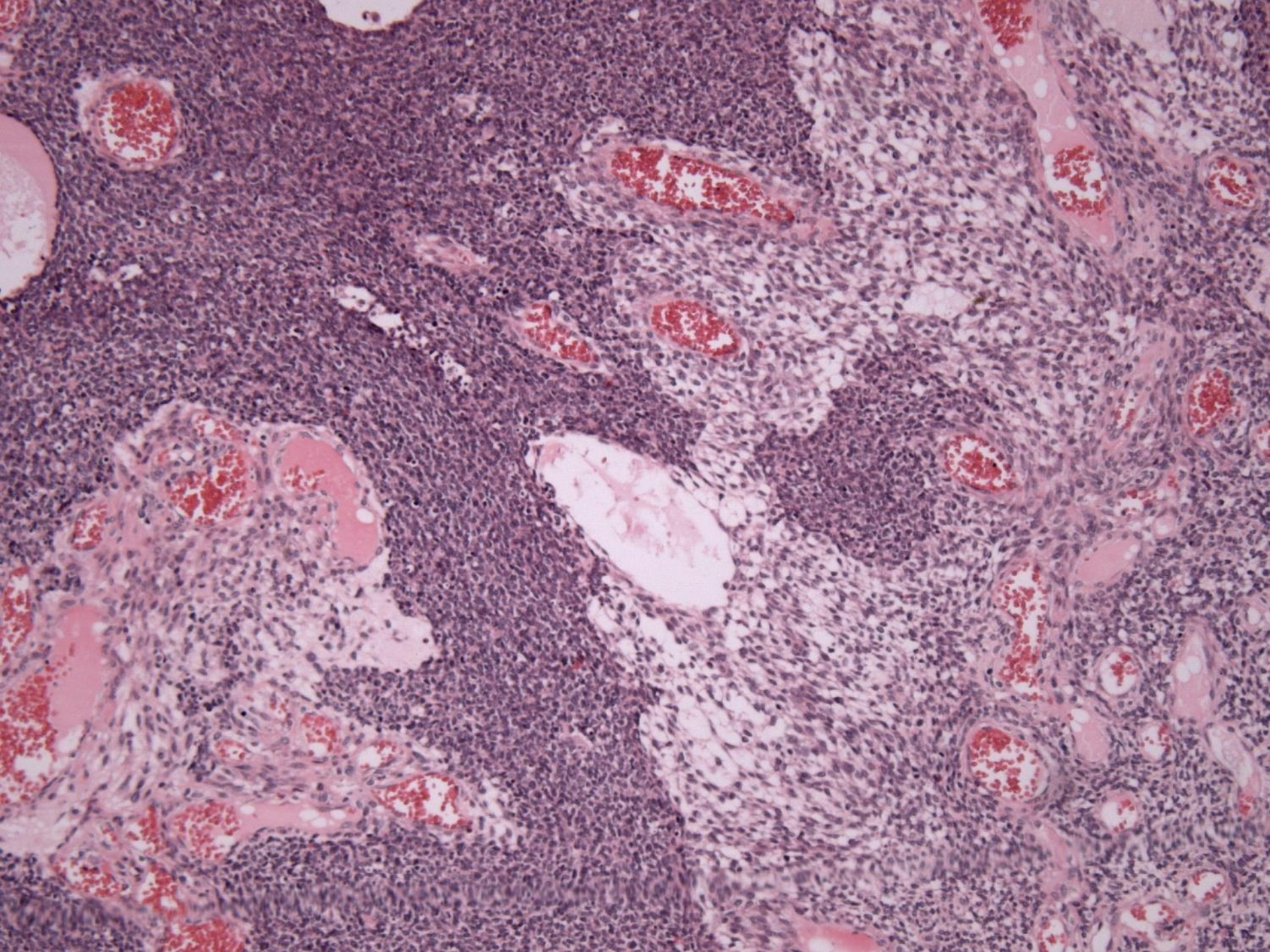




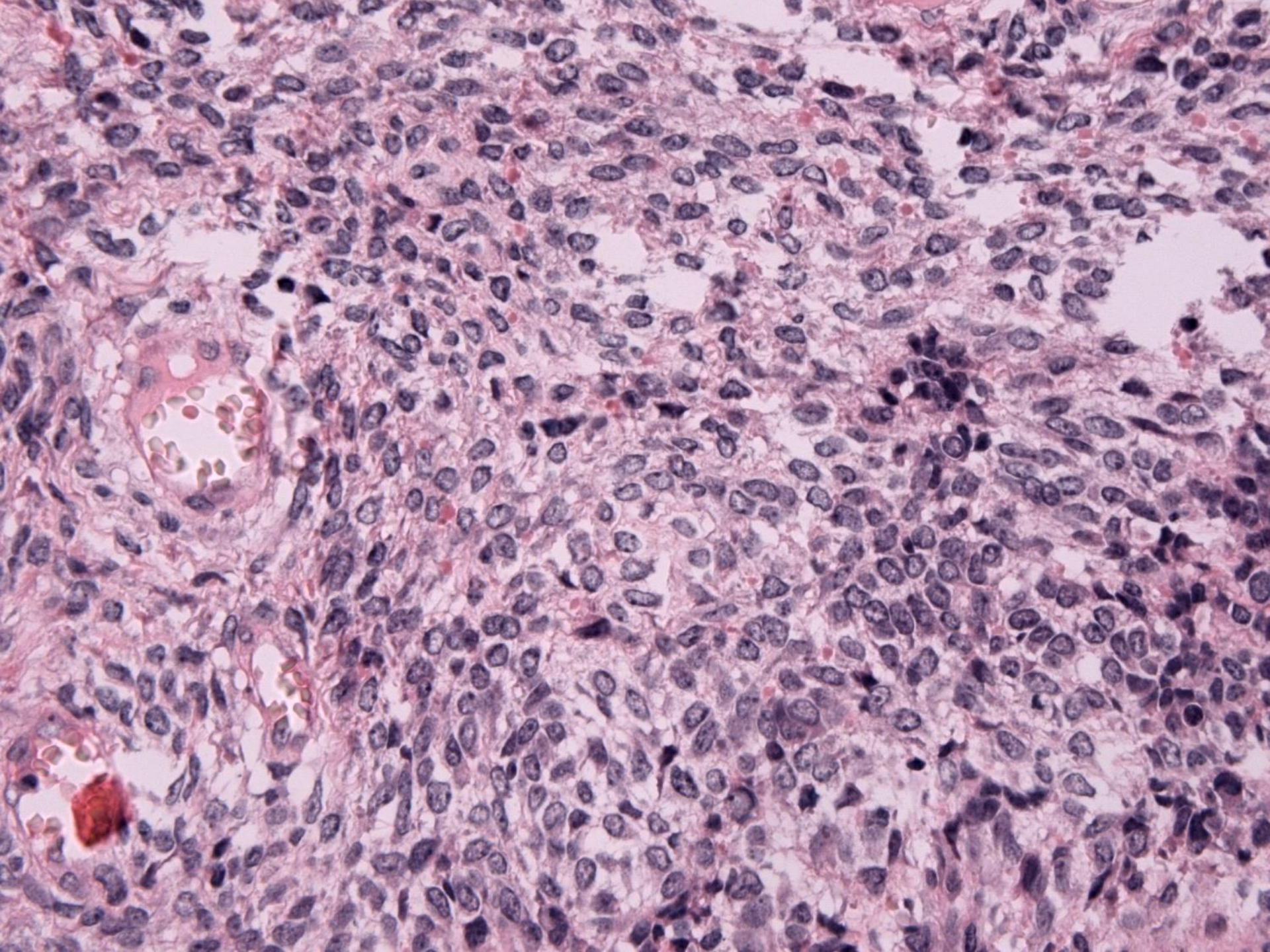


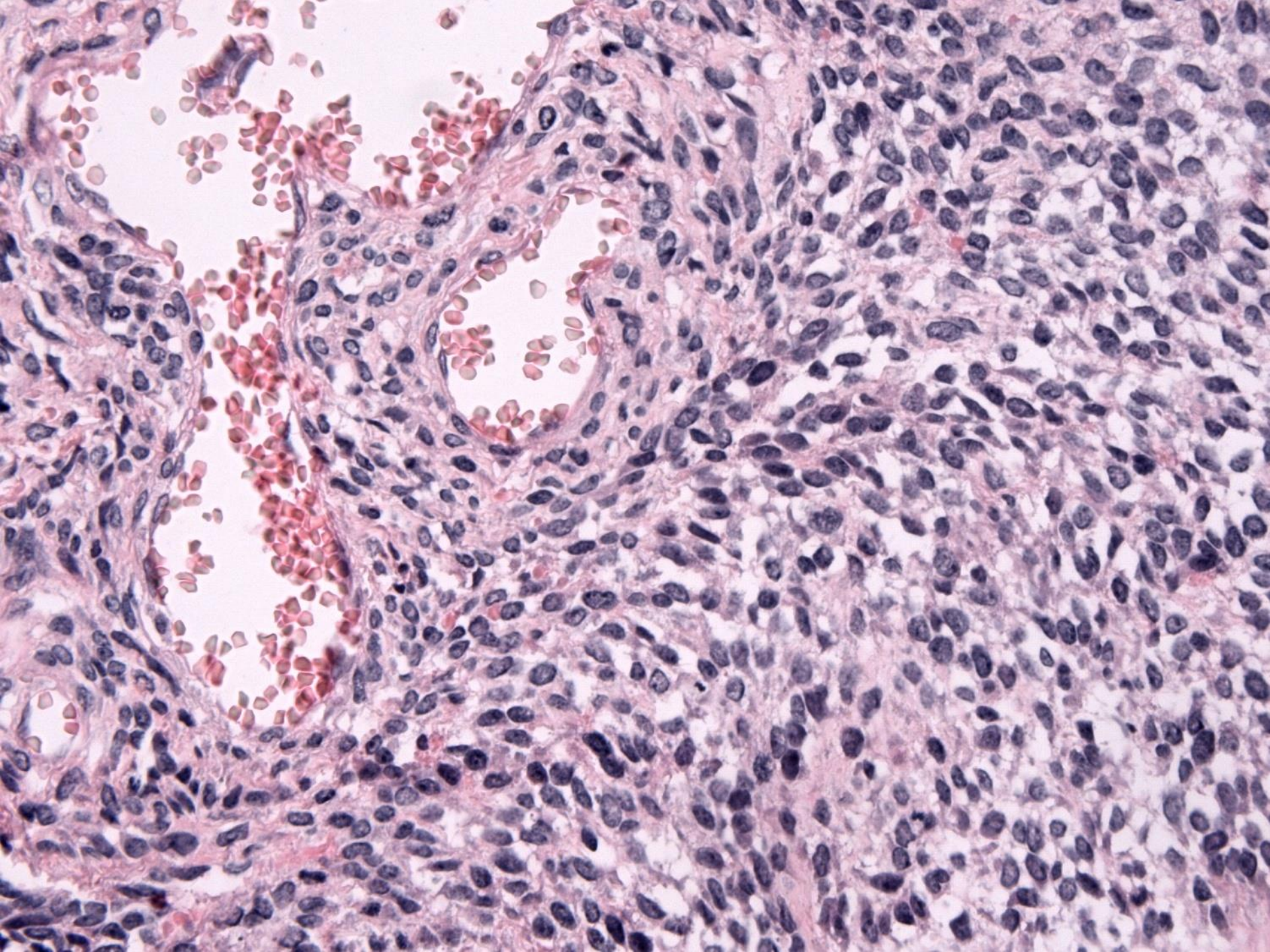


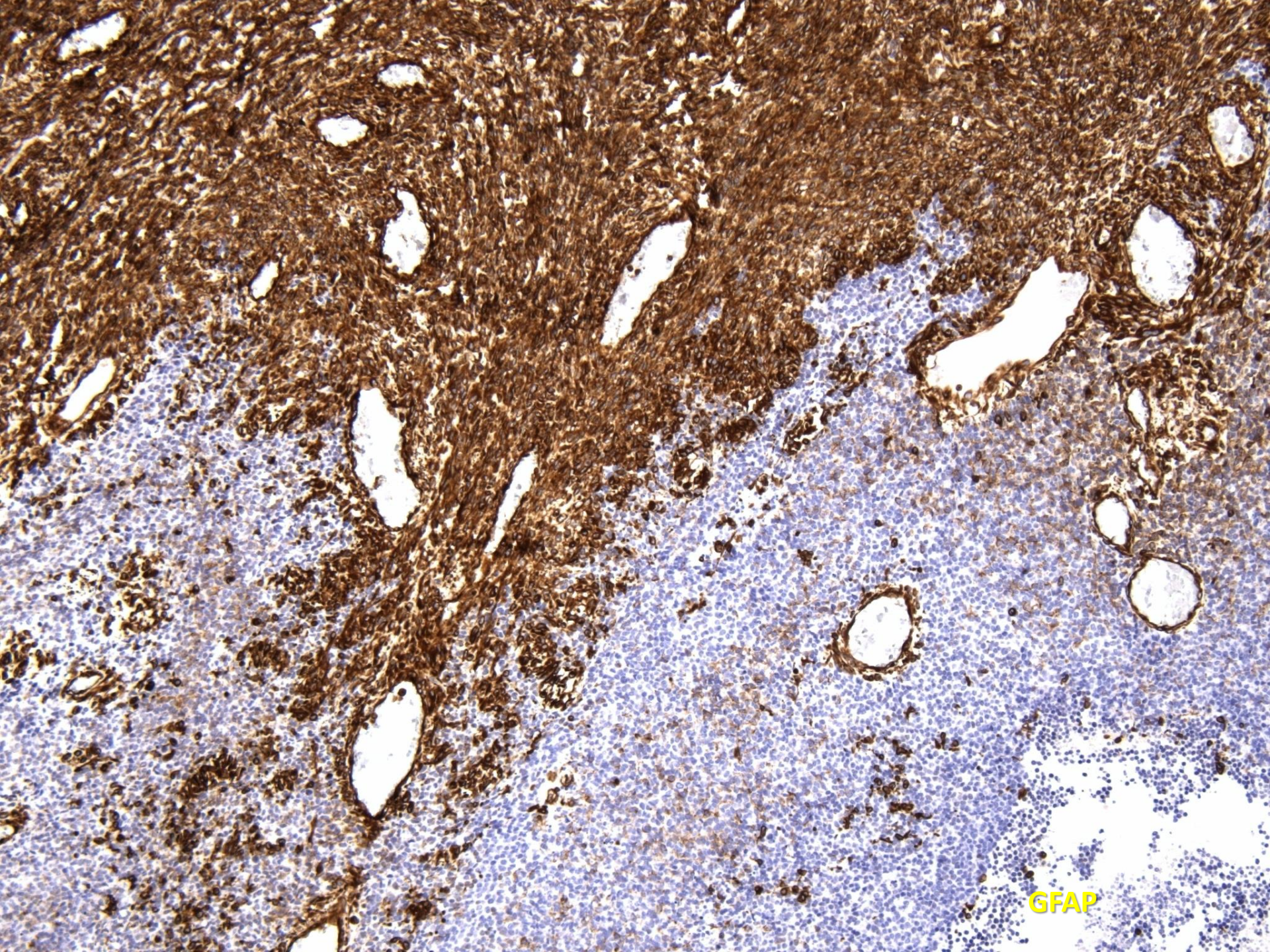




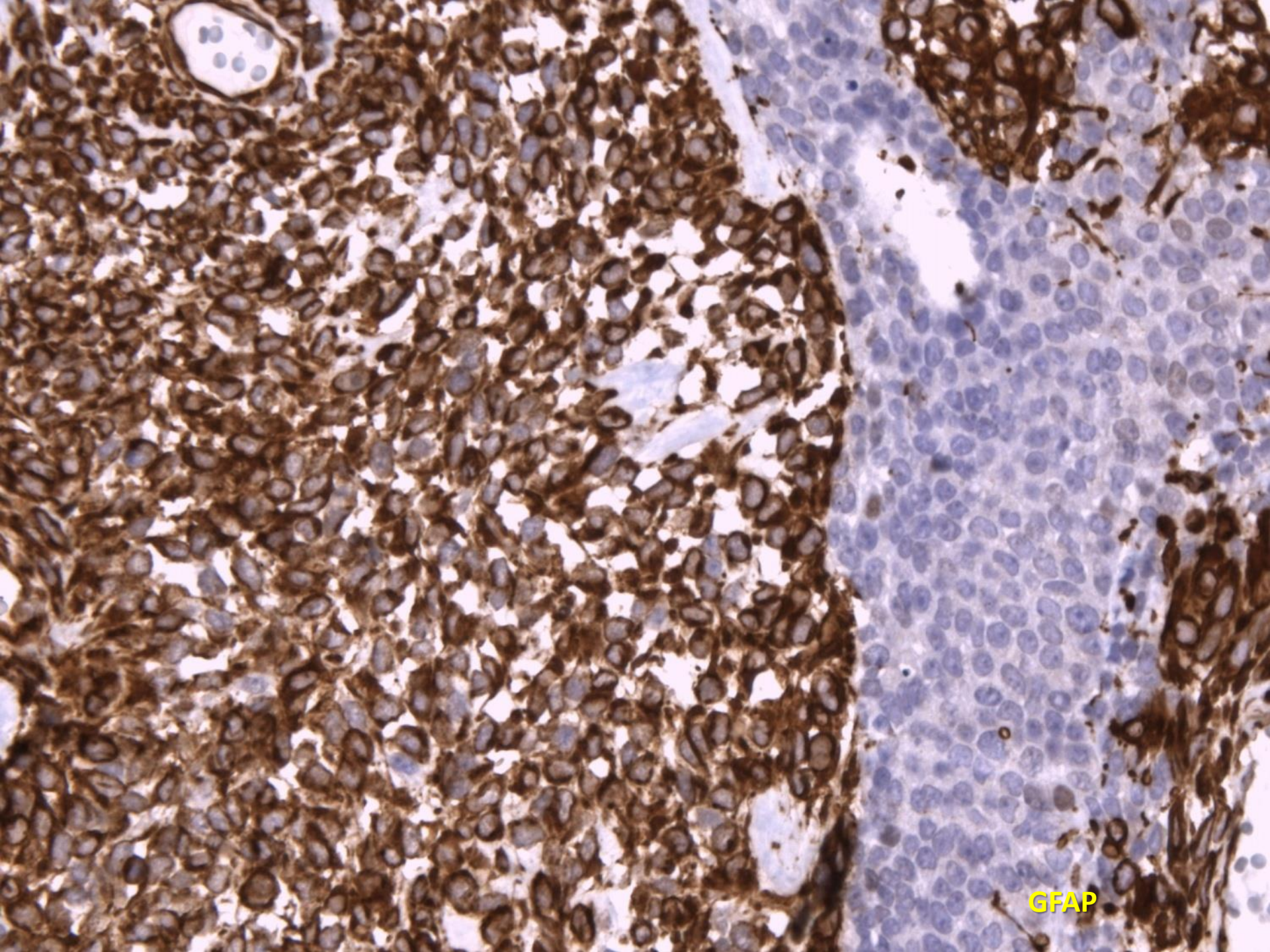




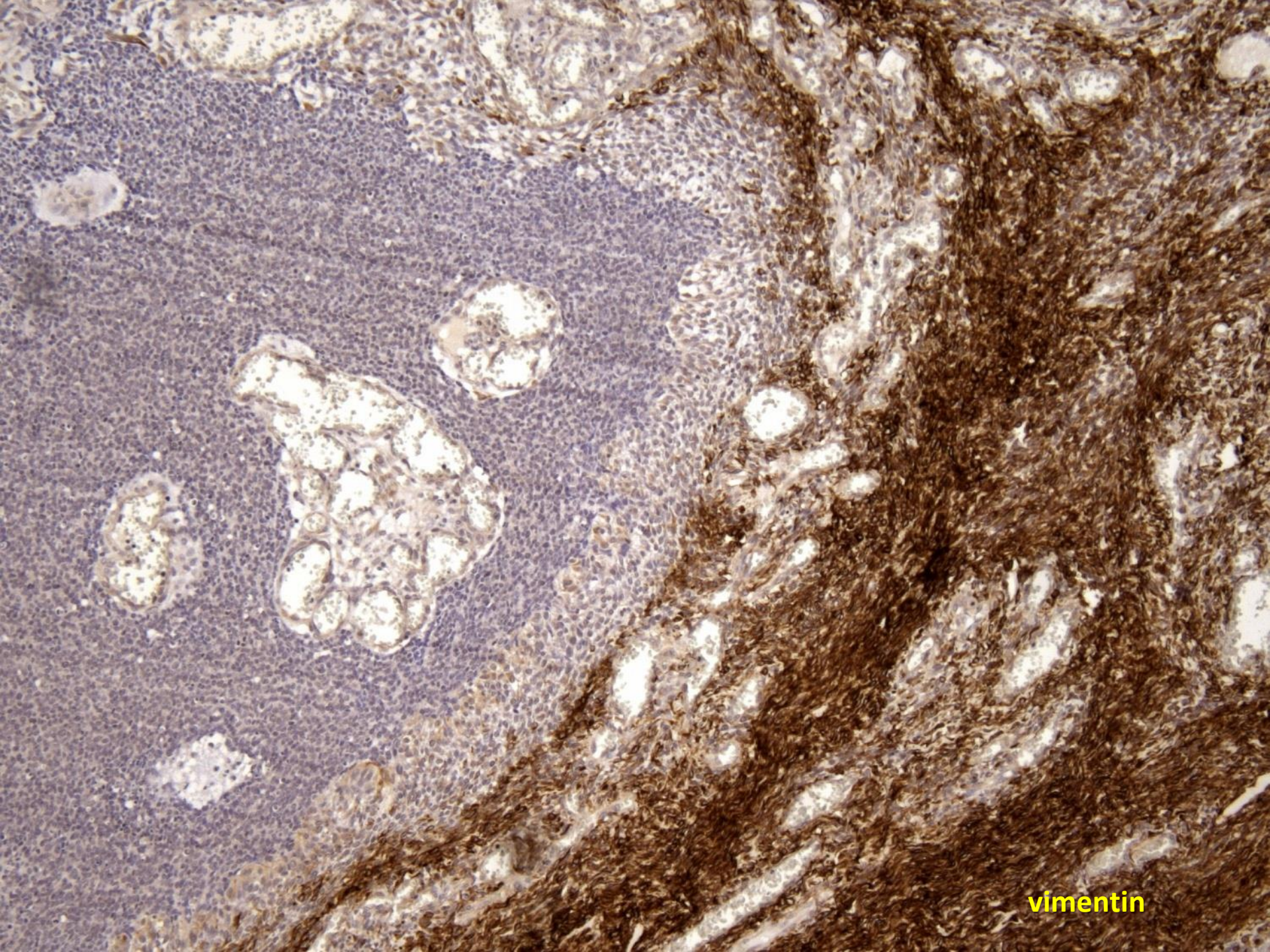




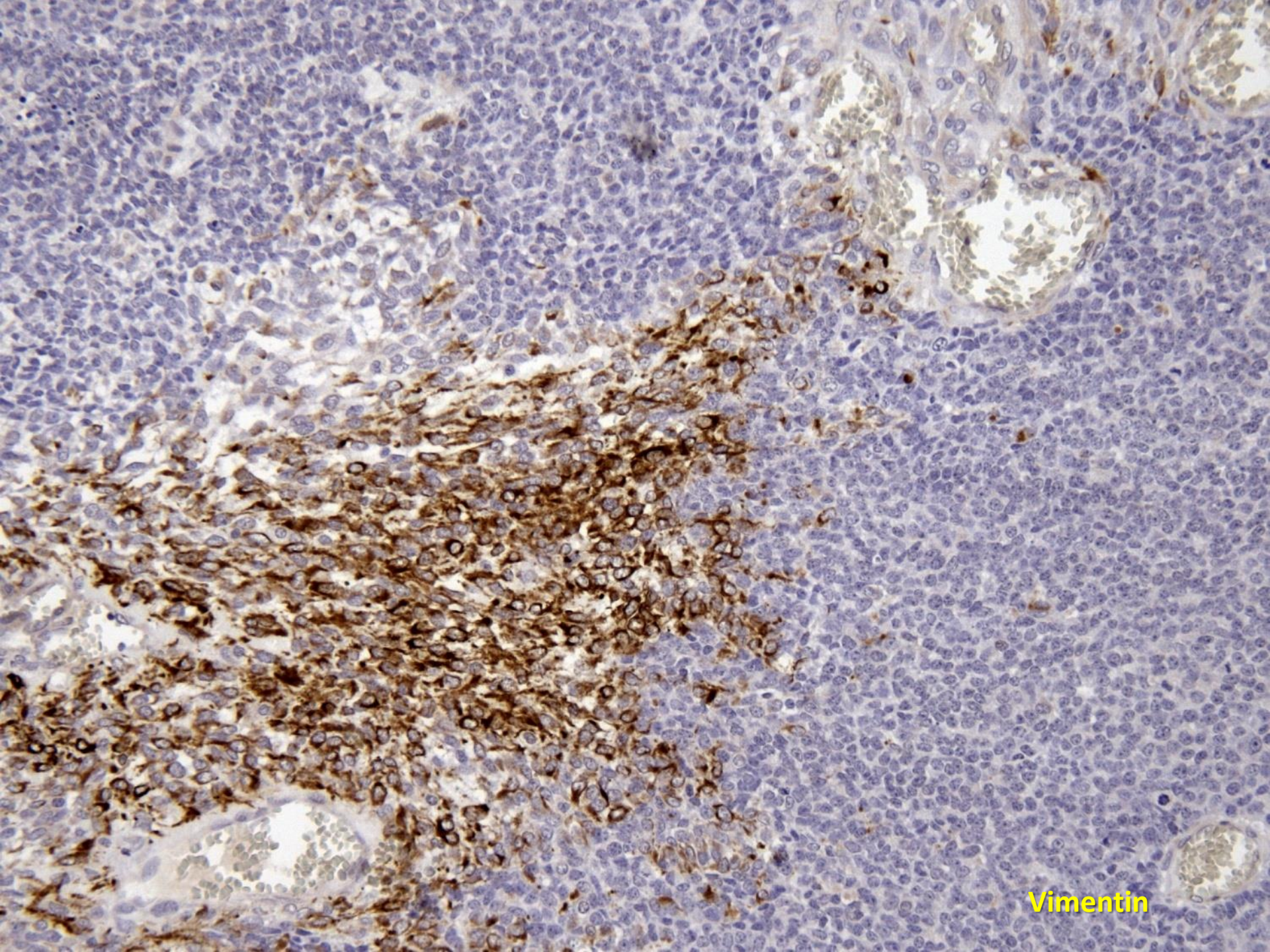
GFAP



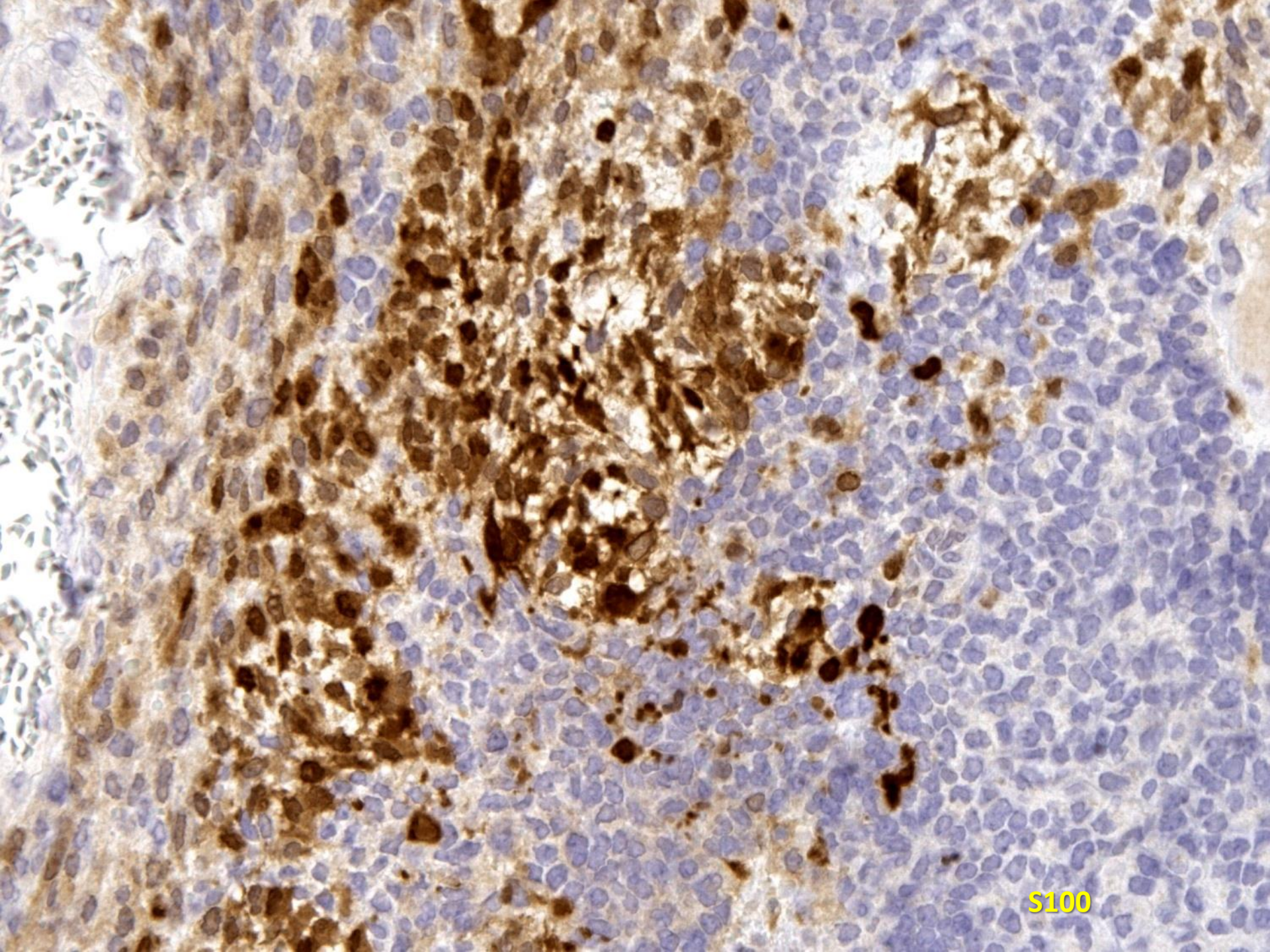
GFAP



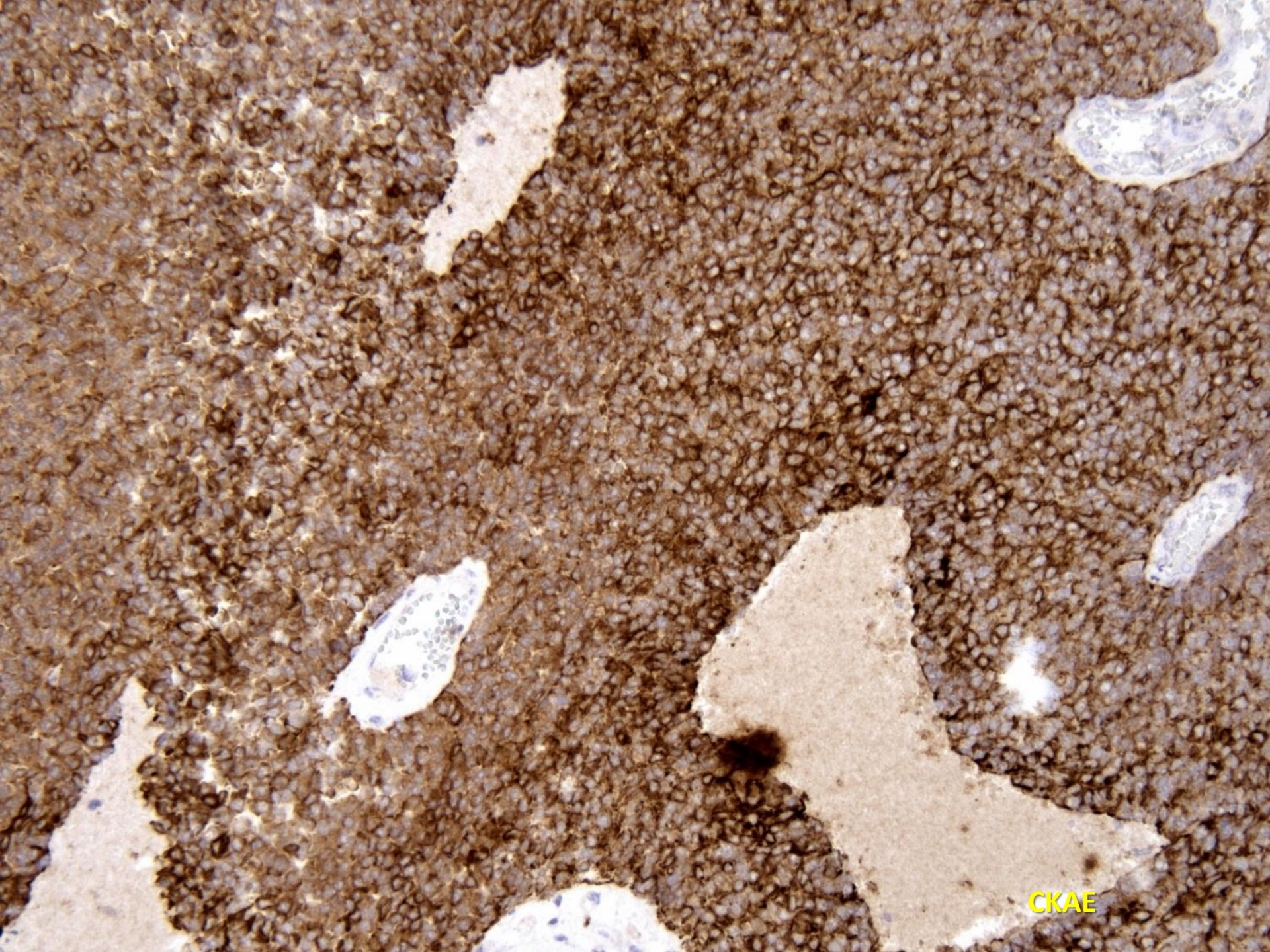
vimentin



Vimentin

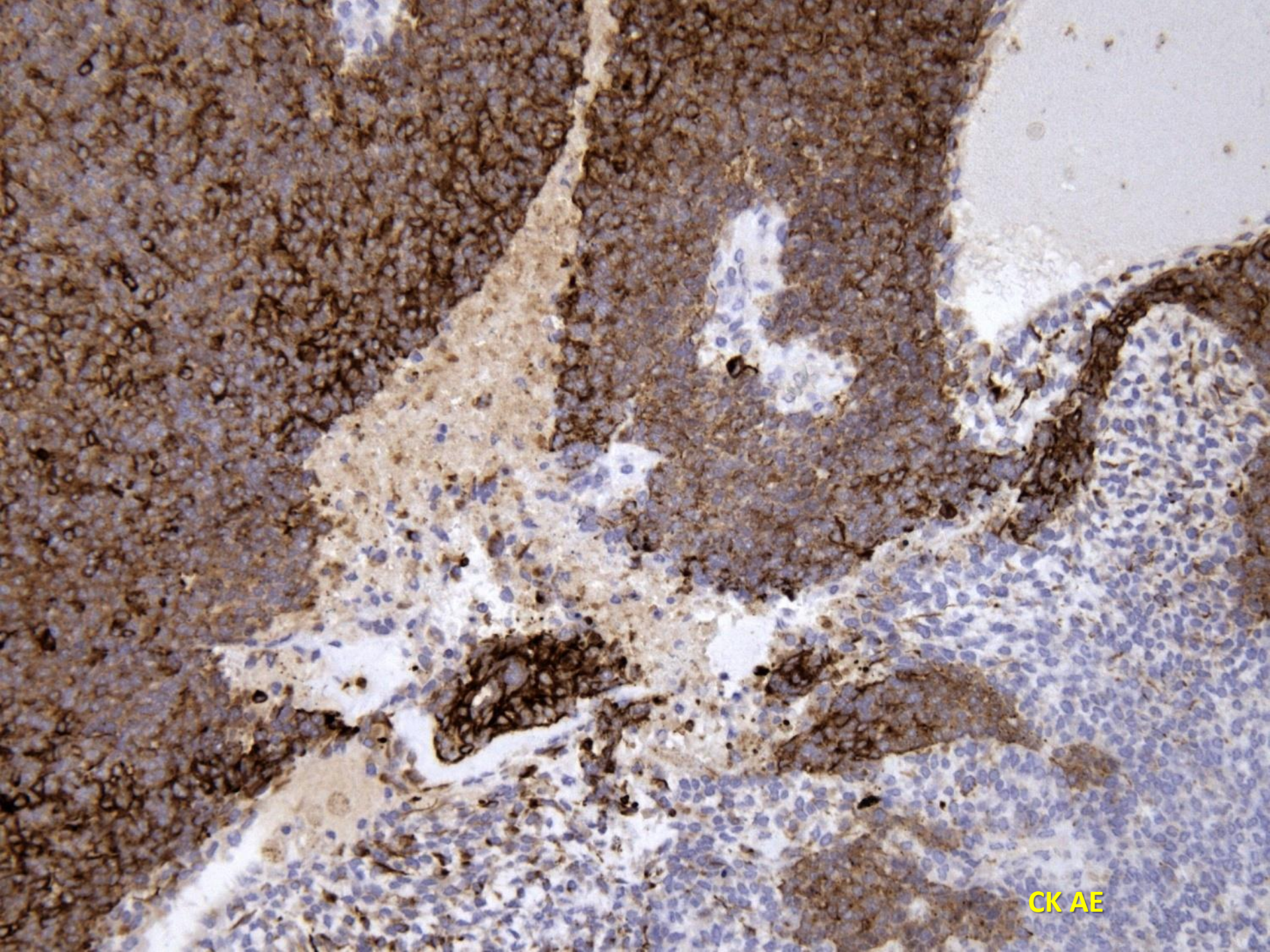


S100

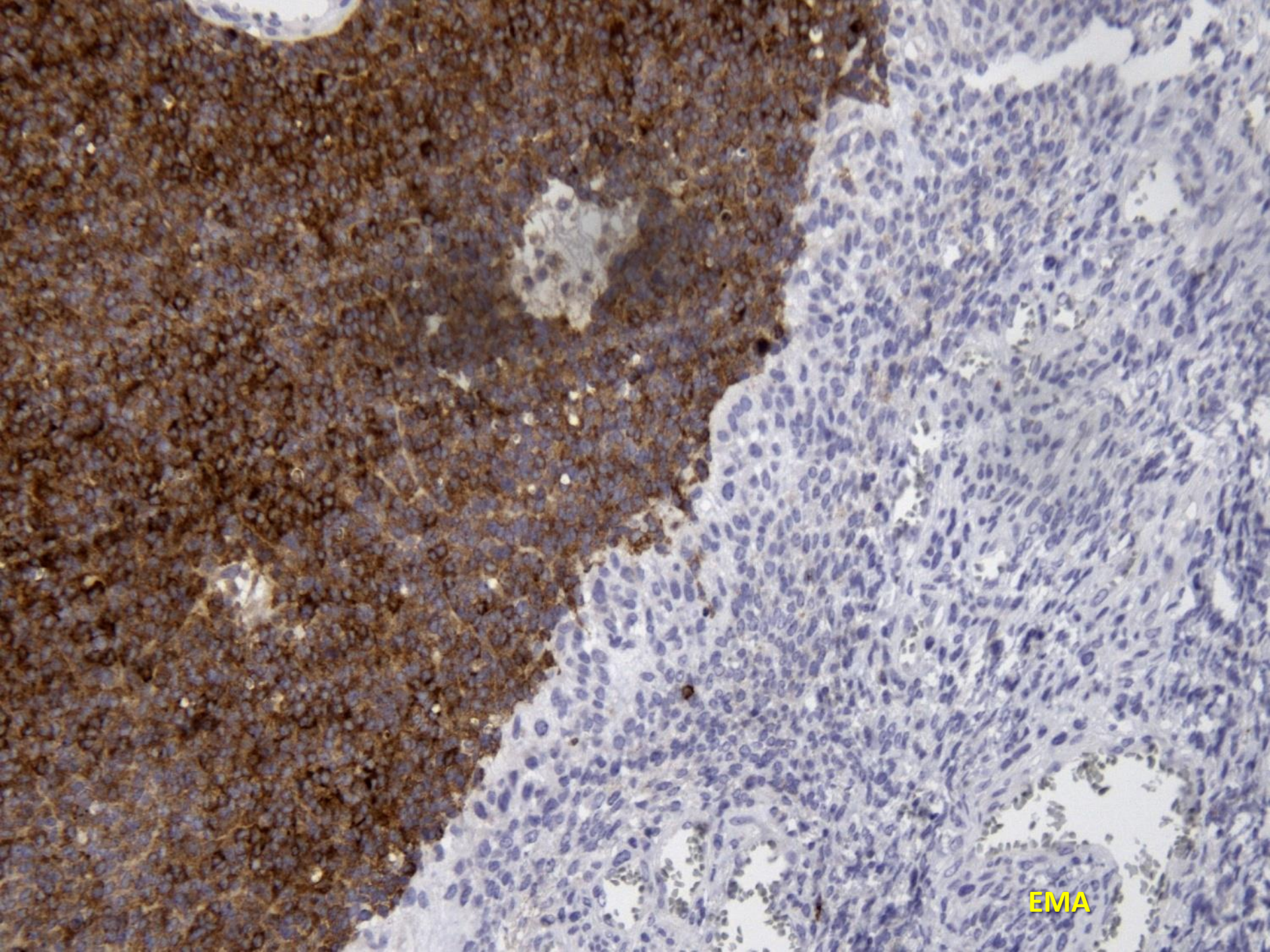


CKAE

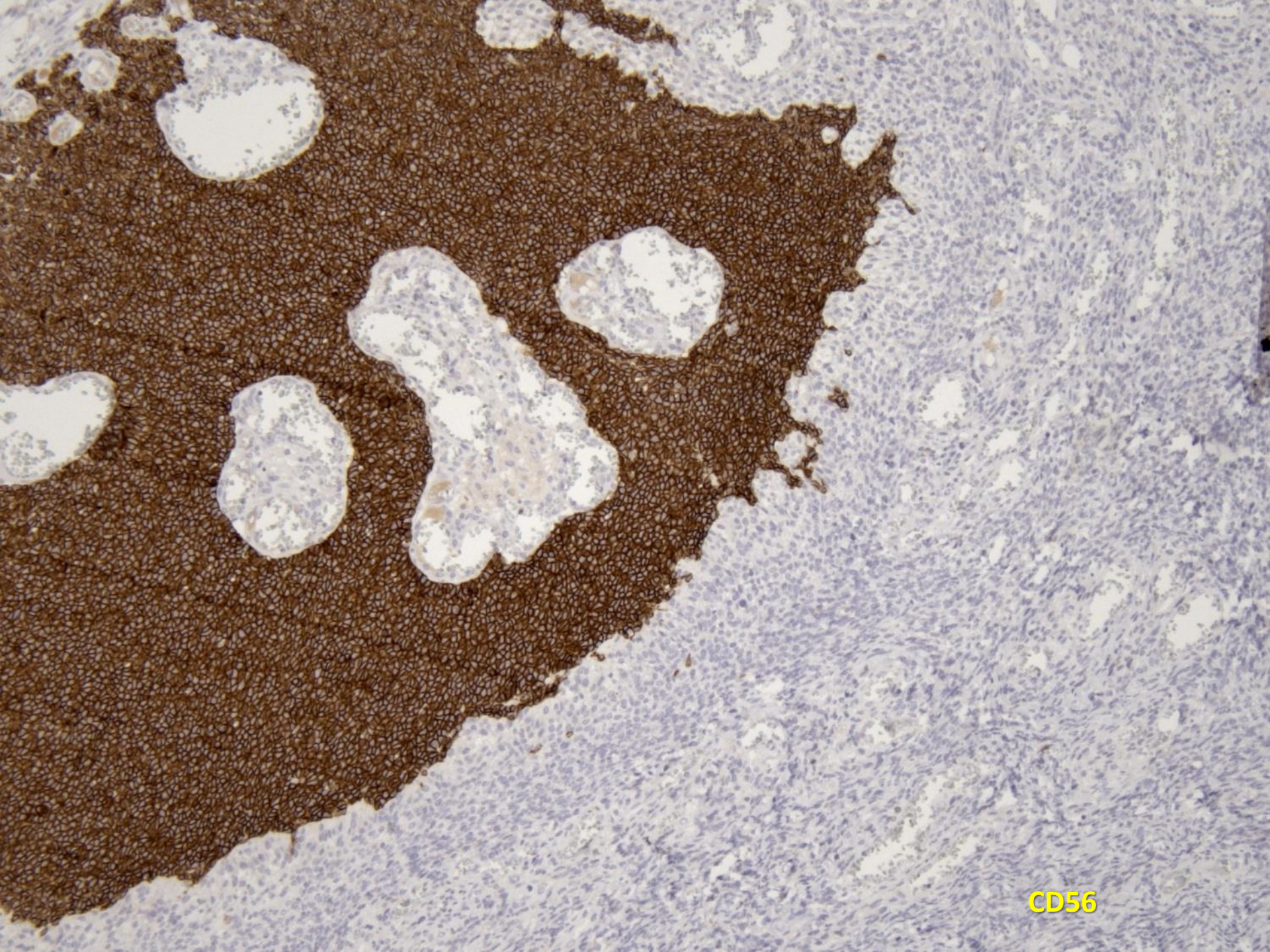




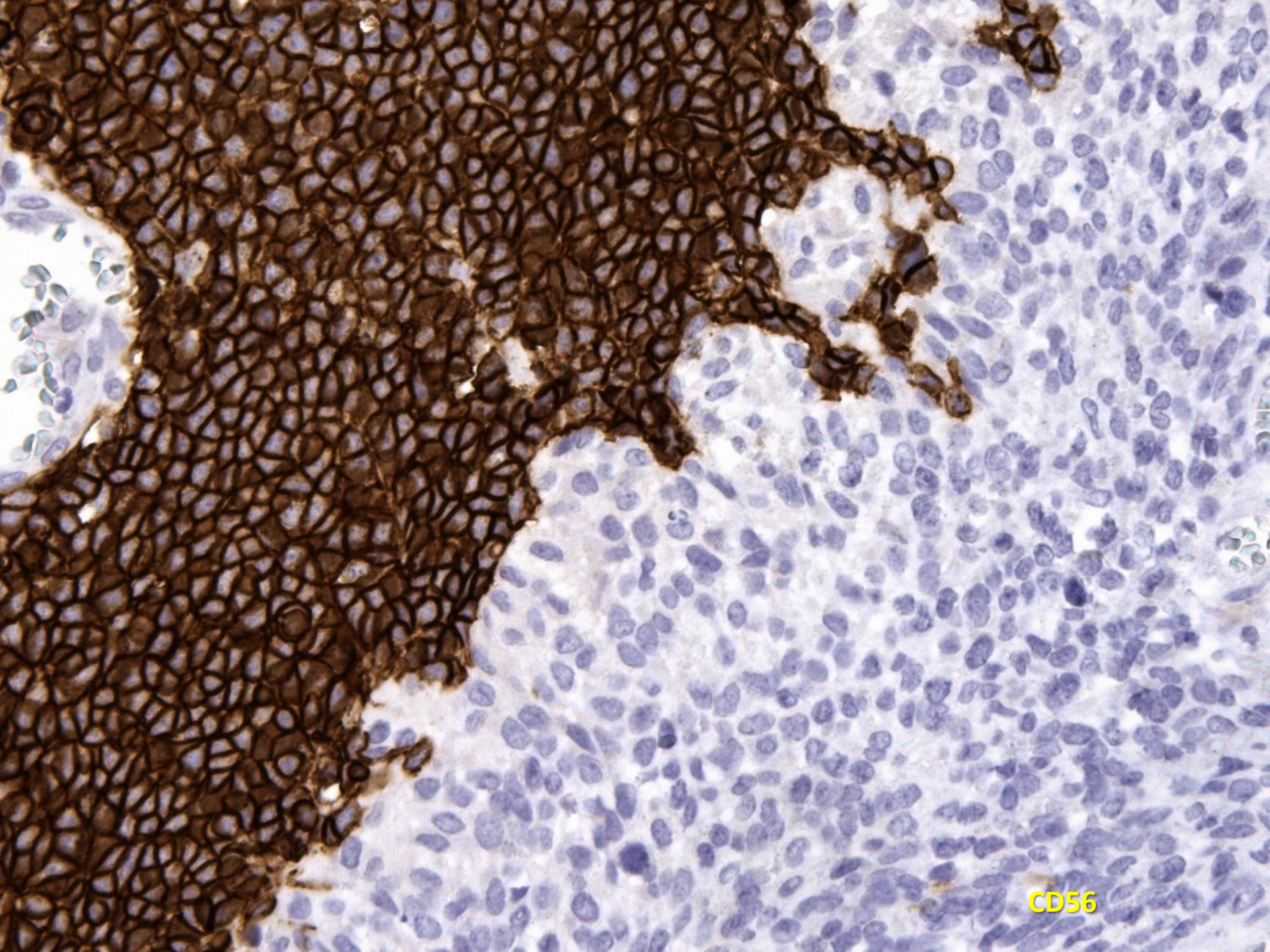
CK AE



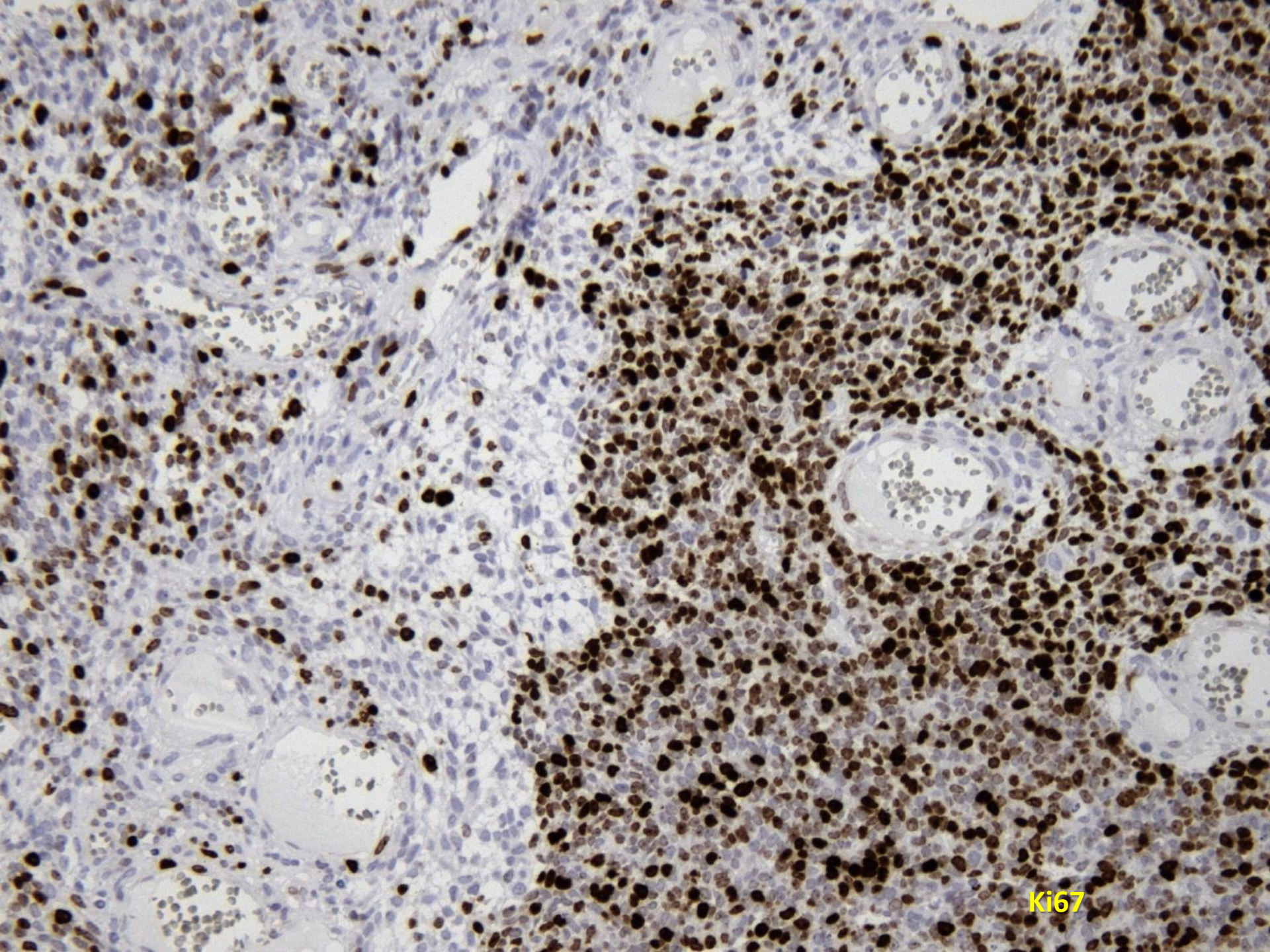
EMA



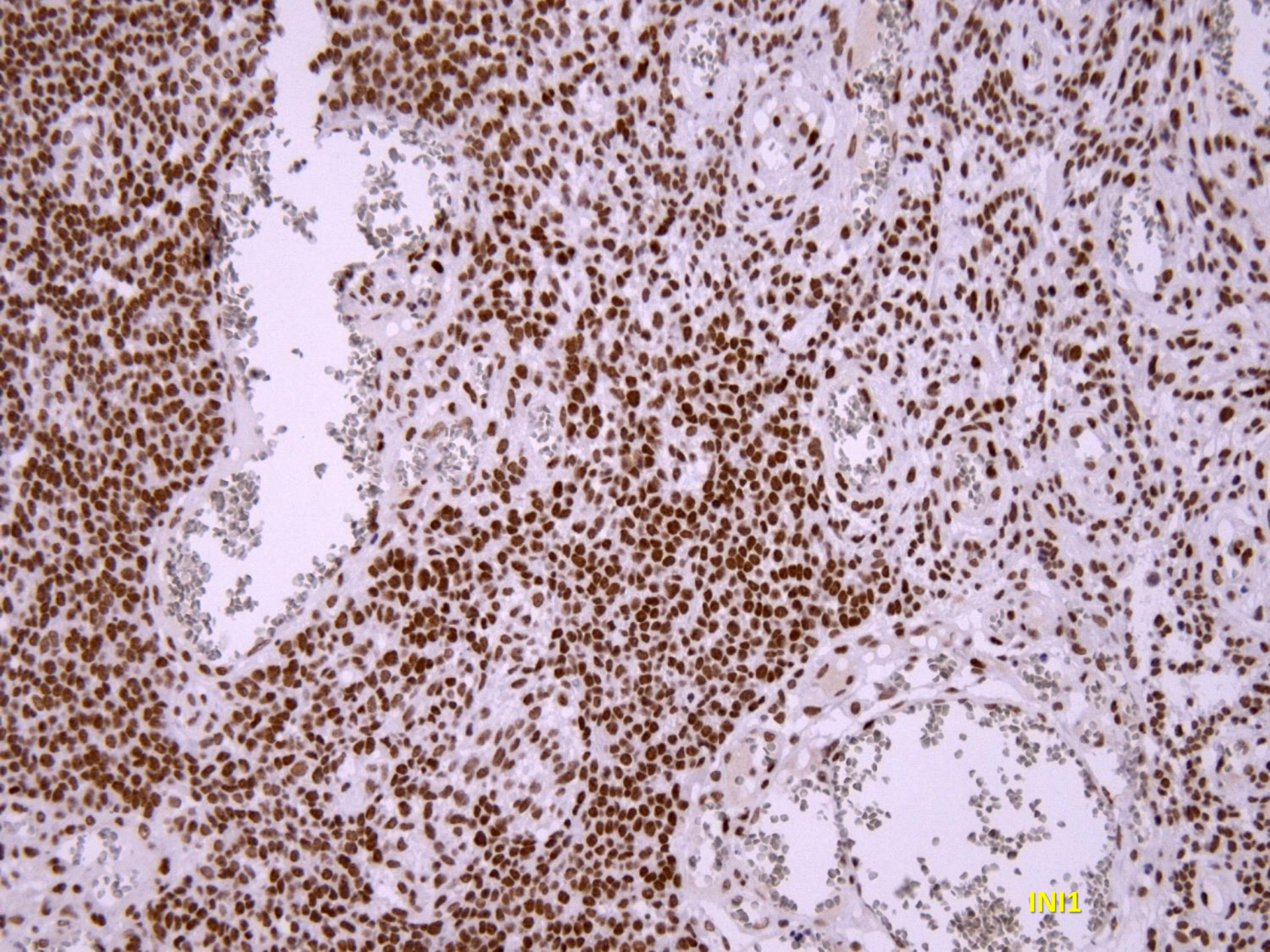
CD56



CD56



Ki67



INI1

## ***Diferenciálně diagnosticky :***

-Embryonální nádor typu ***atypického teratoidního/rhabdoidní nádor*** - exprese INI1 proteinu je ale normálně přítomna.

-Nelze vyloučit, že by nádor mohl morfologicky být součástí germinálního nádoru charakteru ***nezralého teratomu*** - imunohistochemické markery germinálních nádorů jsou však negativní.

-Také jsme zvažovali diagnózu ze skupiny ***desmoplastického infantilního astrocytomu/gangliogliomu (DIA/DIG)***, který může také mít minoritní nezralou komponentu. Ta zde však zcela dominuje.



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**Final Surgical Pathology Consultation Report**  
**Referral Case**

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HISTORY NO.: 20170502  
RECEIVED: 2017-05-02  
REPORTED: 2017-05-15  
TAKEN: 2017-05-02

BIRTH DATE: 2016-02-05  
SOURCE: Referred In  
SUBMITTING PHYSICIAN: Eric Bouffet  
RESP. PHYSICIAN:

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**Microscopic Description:**

Sections show a biphasic neoplasm composed of sheets of small cells with fairly uniform round hyperchromatic nuclei sharply interfacing with more mesenchymal appearing areas with larger nuclei with more open chromatin. Both areas have a prominent vasculature, in some cases forming hemangioma-like structures. Mitotic figures are readily identified in the epithelial areas and necrosis are present and calcifications are also noted. Reticulin deposition is abundant in both areas with perhaps slightly more in the mesenchymal area. BAF47/INI1 staining is positive in the epithelial areas and focally for pan-keratin. Whereas the mesenchymal areas are diffusely positive for vimentin with focal GFAP positivity. SMA highlights the extensive vasculature. Lymphocytic infiltrates are seen in the epithelial cell areas and ~15% in the mesenchymal areas. The tumour is diffusely immunonegative for synaptophysin, Olig-2, WT1 and CD99.

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**Comment:**

The tumour shows an immature epithelial component along with a mesenchymal and vasculature component. Morphologically it does not resemble an atypical teratoid/rhabdoid tumour. It is immunohistochemically negative for CD99 as well as WT-1. Overall, the appearance is most in keeping with component of an immature teratoma. This case was internally peer-reviewed at P

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**Final Diagnosis:**

**Fronto-parietal tumour (referred in 3777/17):**  
**- Consistent with immature teratoma (see comment) |**

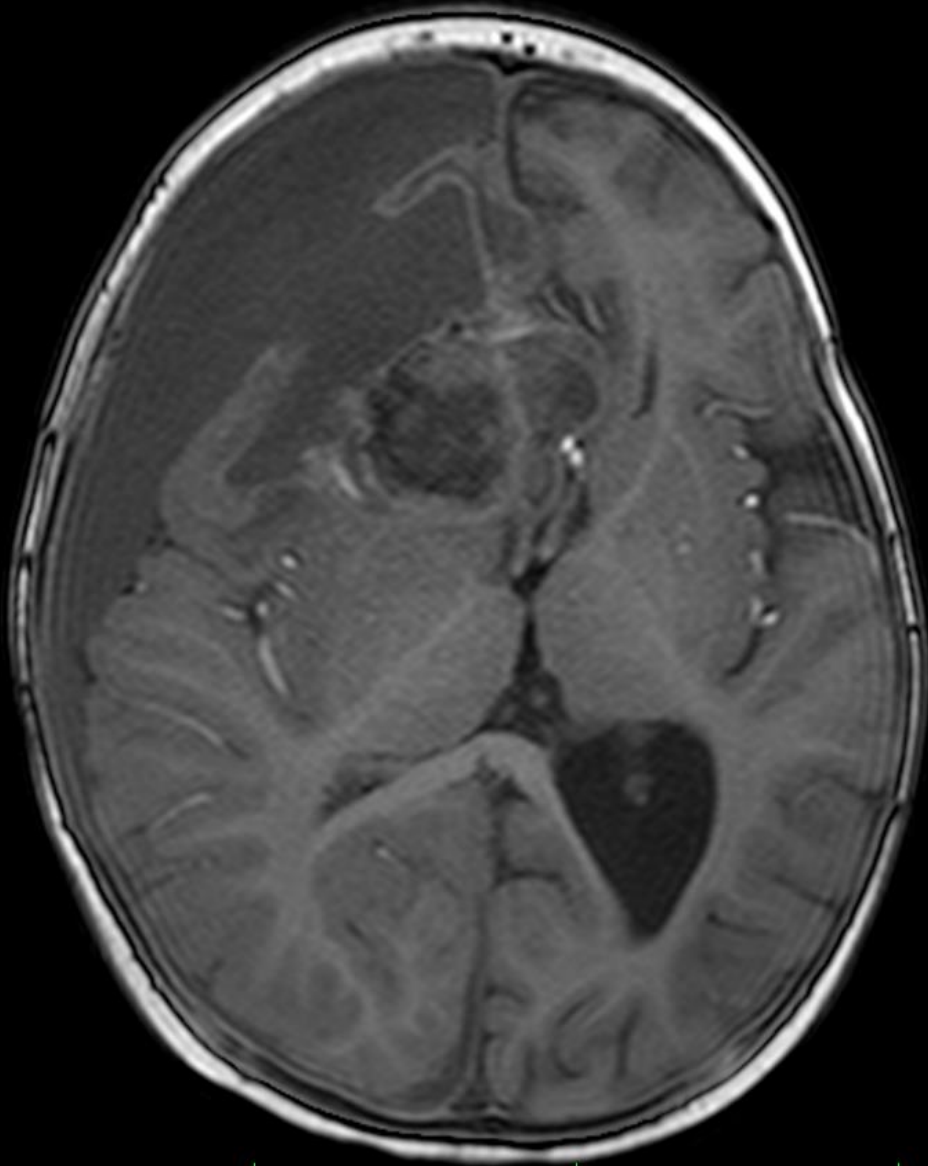


1. operace

Pooperačně – reziduum

+ progrese

A

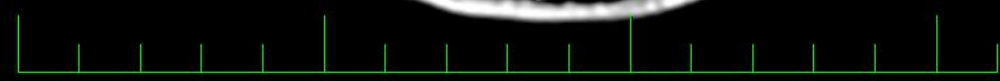


P

A



P



vyrazna progrese velikosti rezidua tumoru v  
okraji poop.dutiny

2. operace

A



P

01/2018 – progrese

+ rozsahlá infiltrace mening paterniho kanalu  
drobnoložiskovým procesem + infratentorialne kolem  
mozecku, mozkového kmene i supratentorialne v prave  
postranni komore.







*2 roky chlapec*

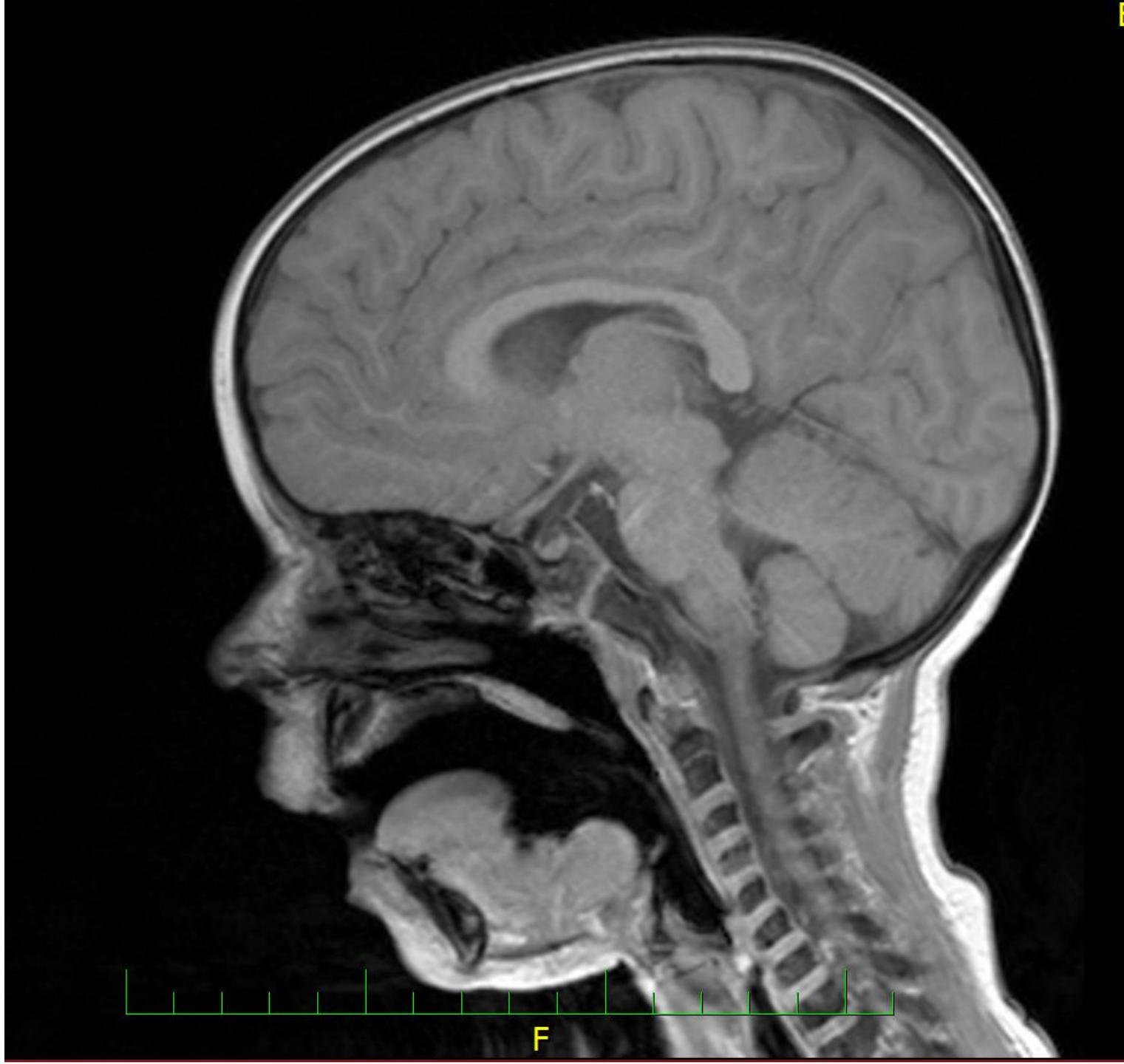
**tu mostomozečkového koutu dx.**

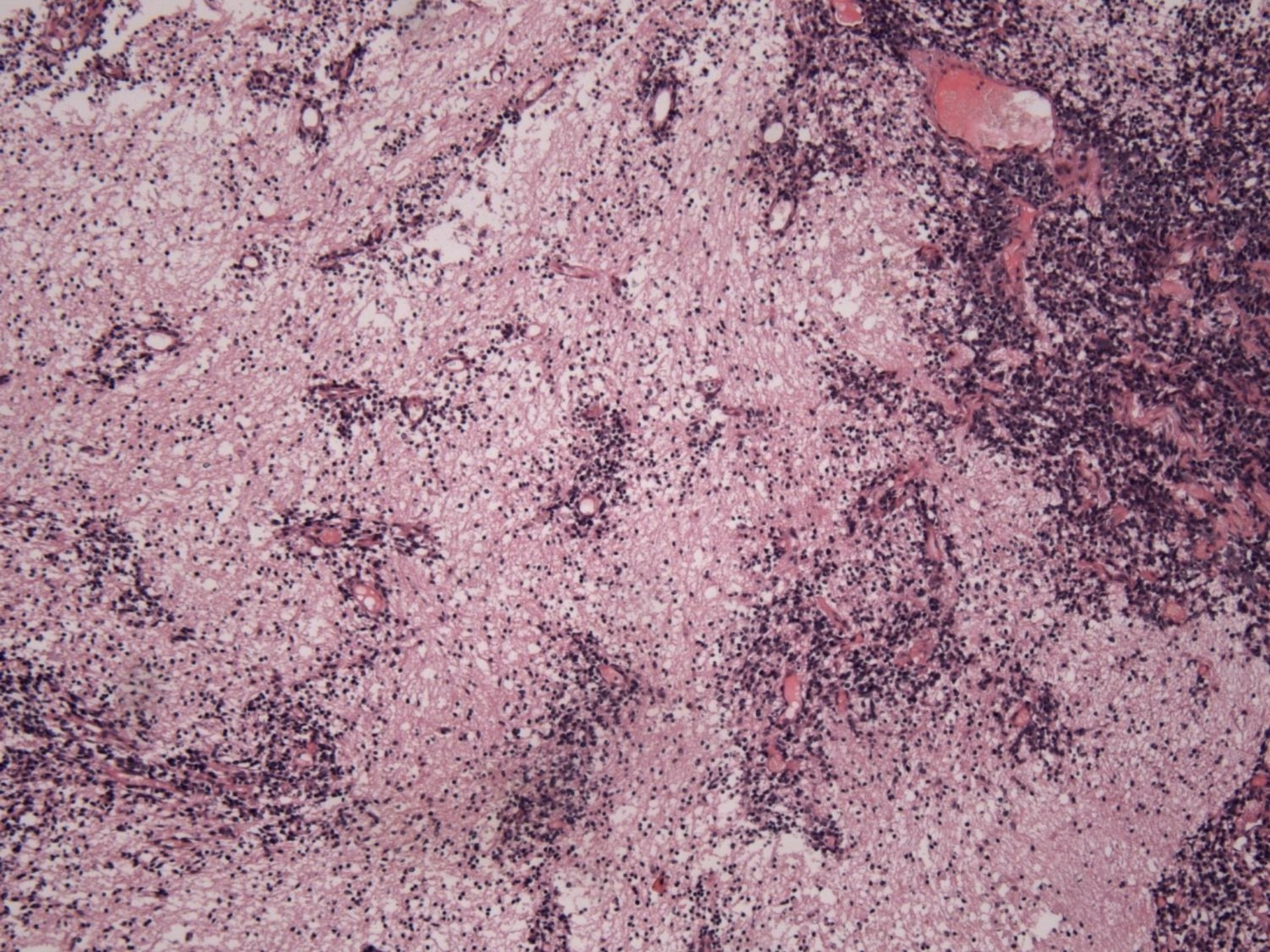
H

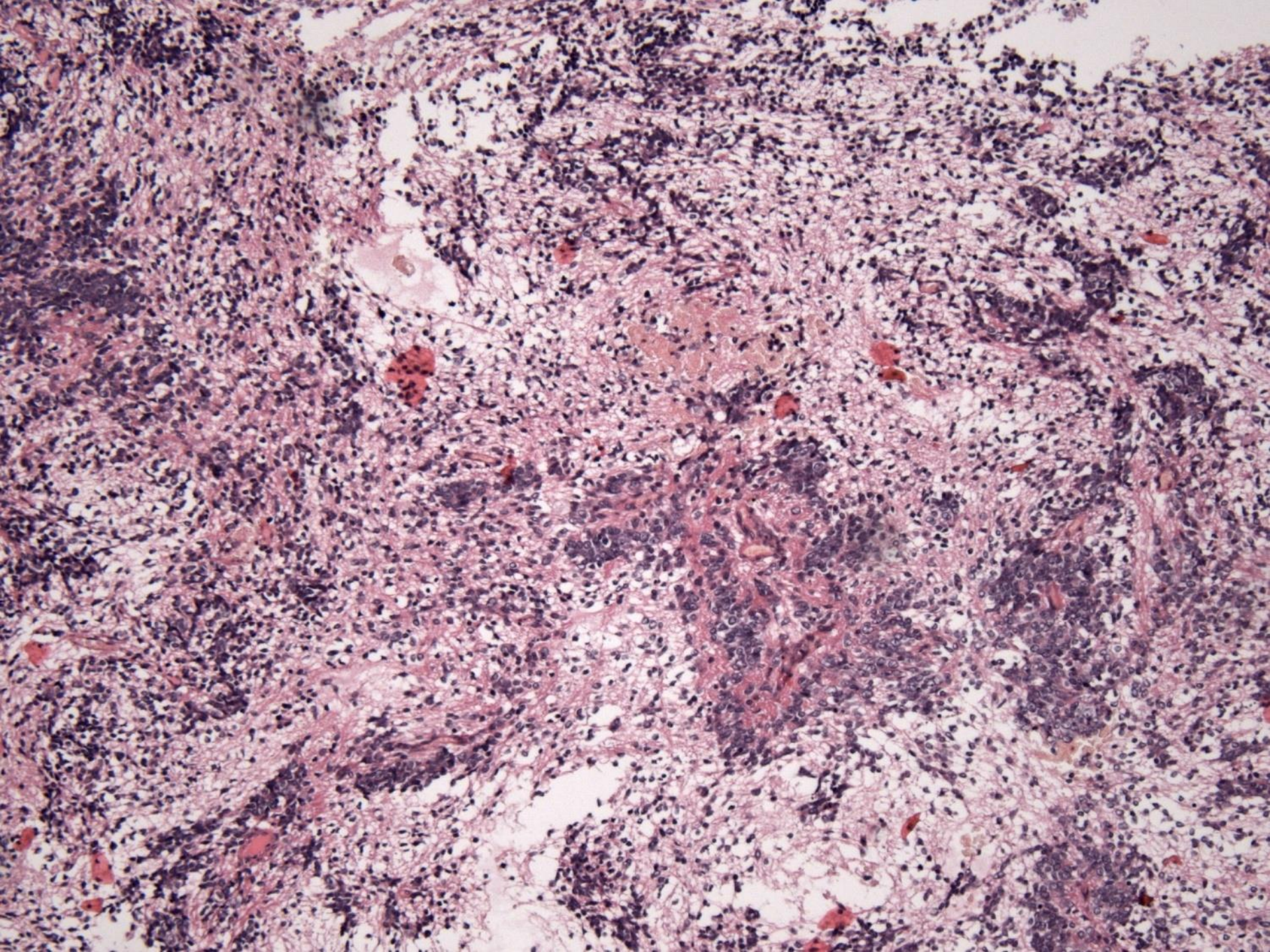
Fakult  
Bi

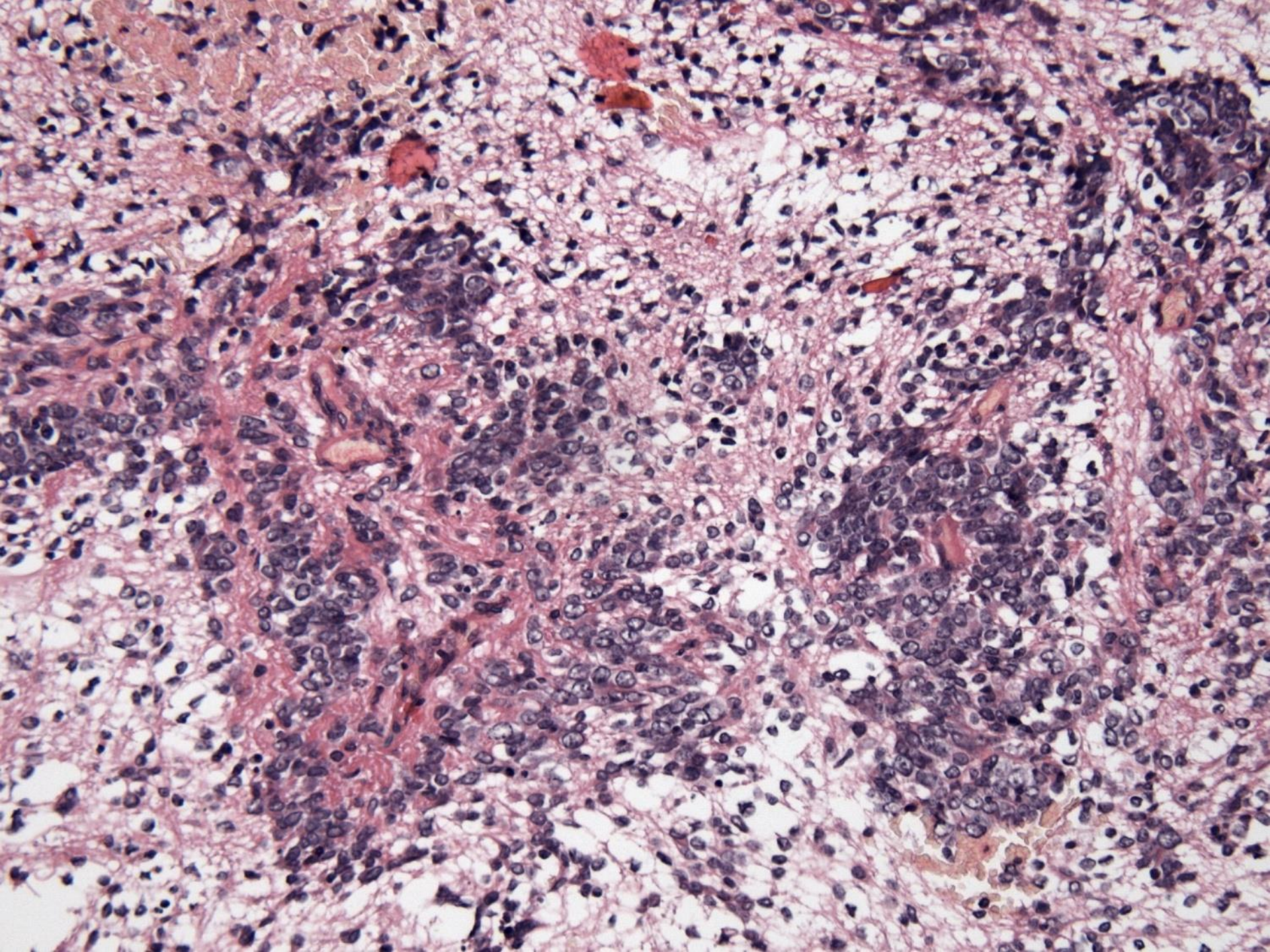


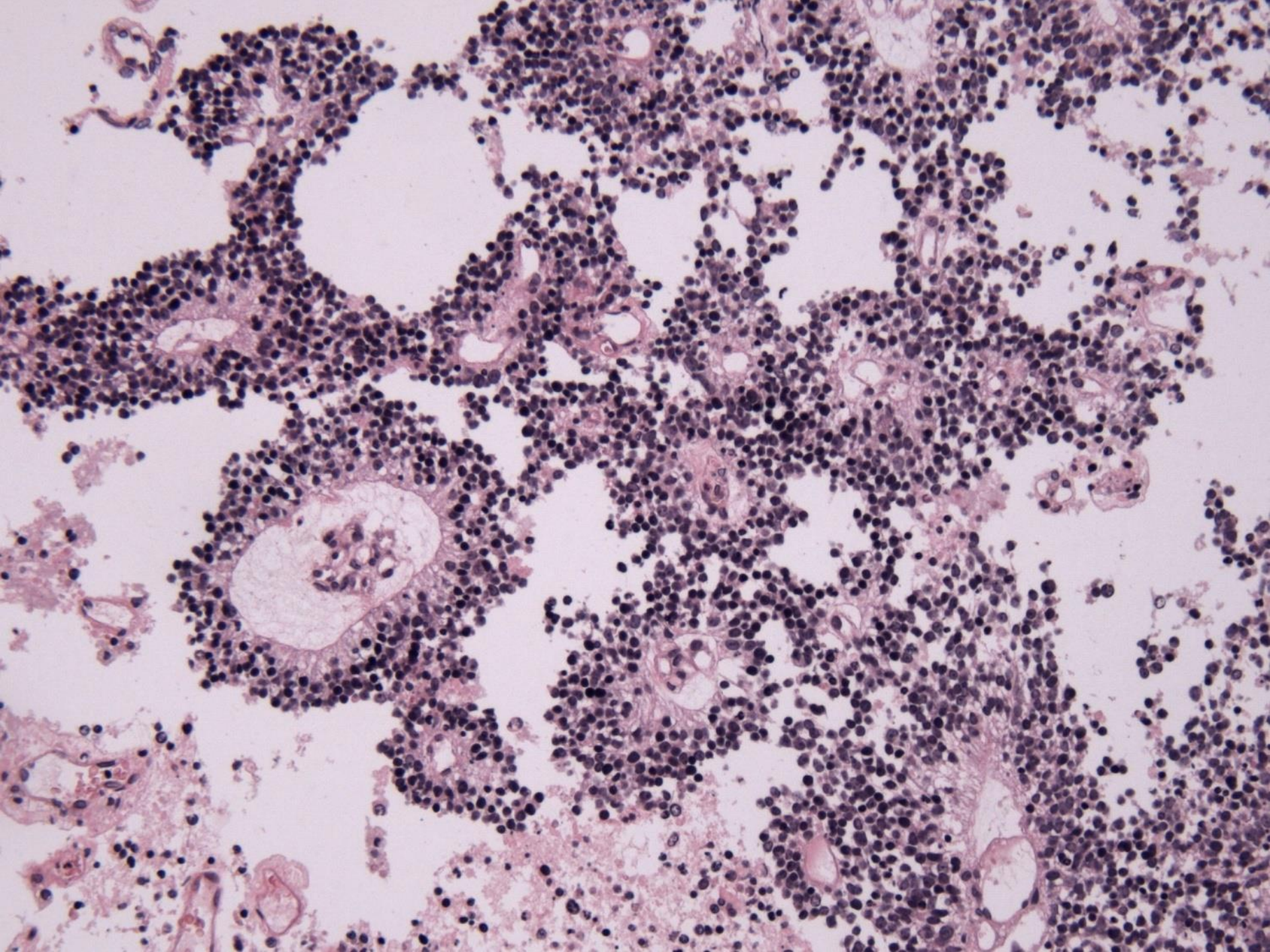
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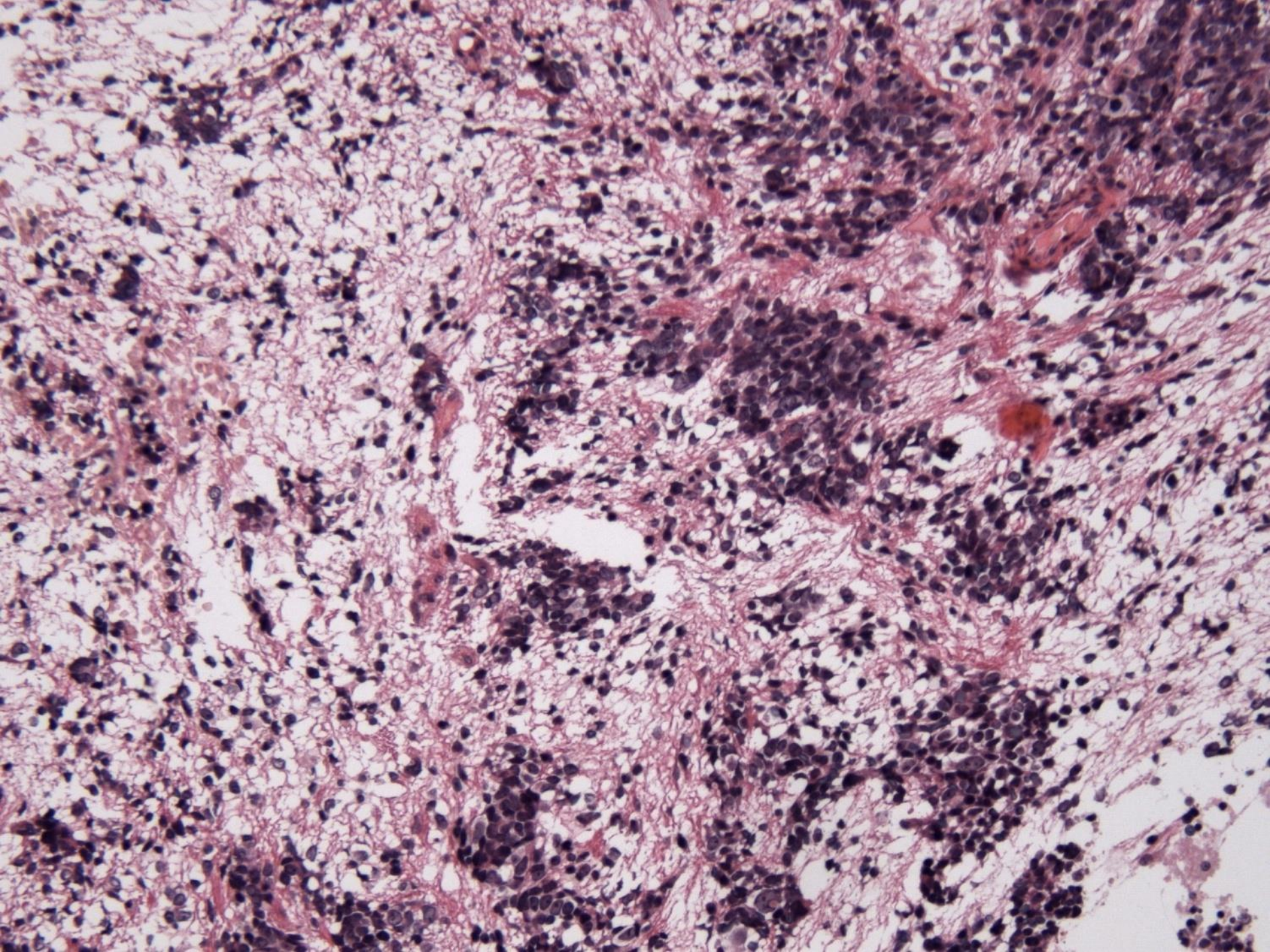




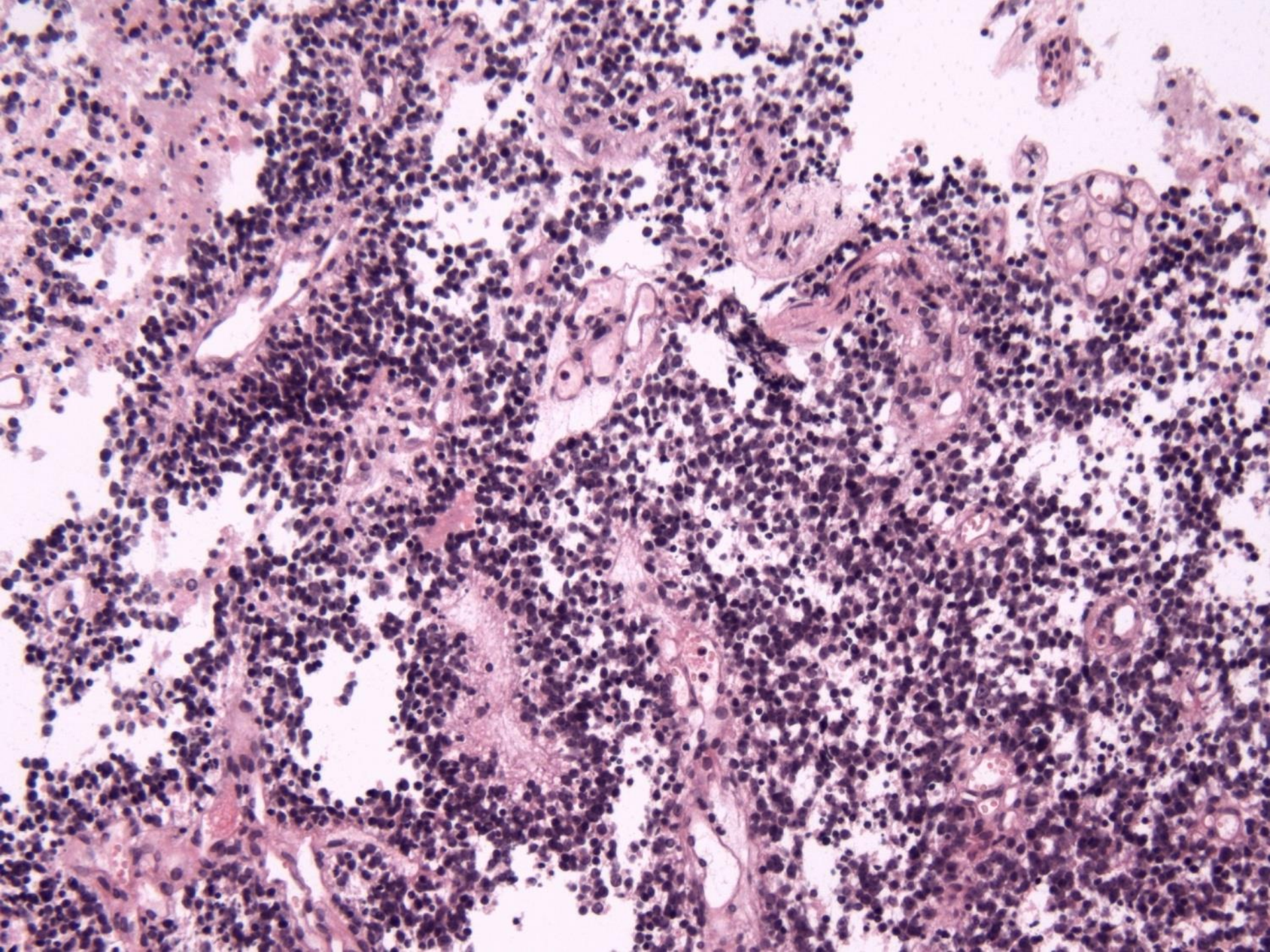


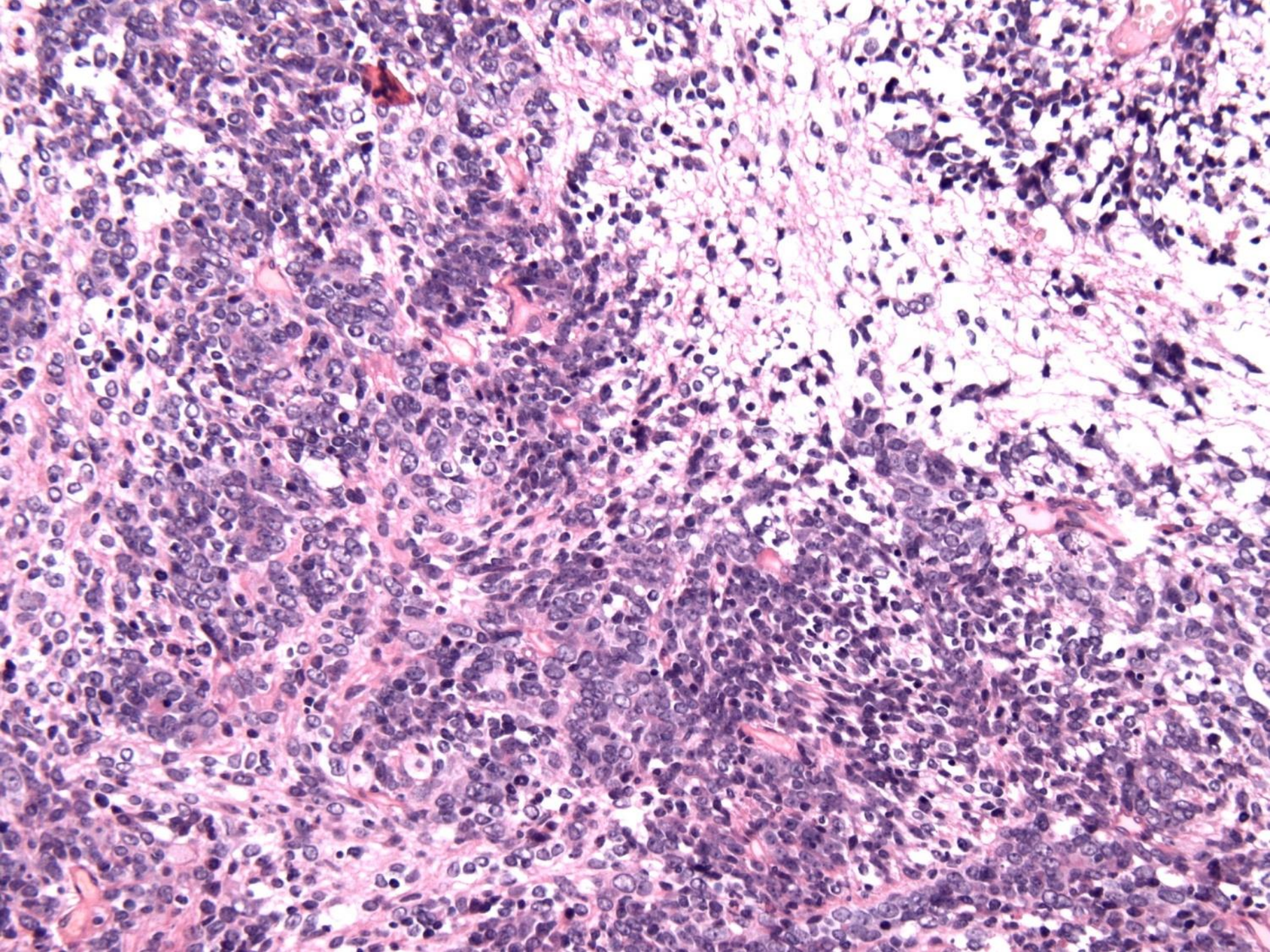


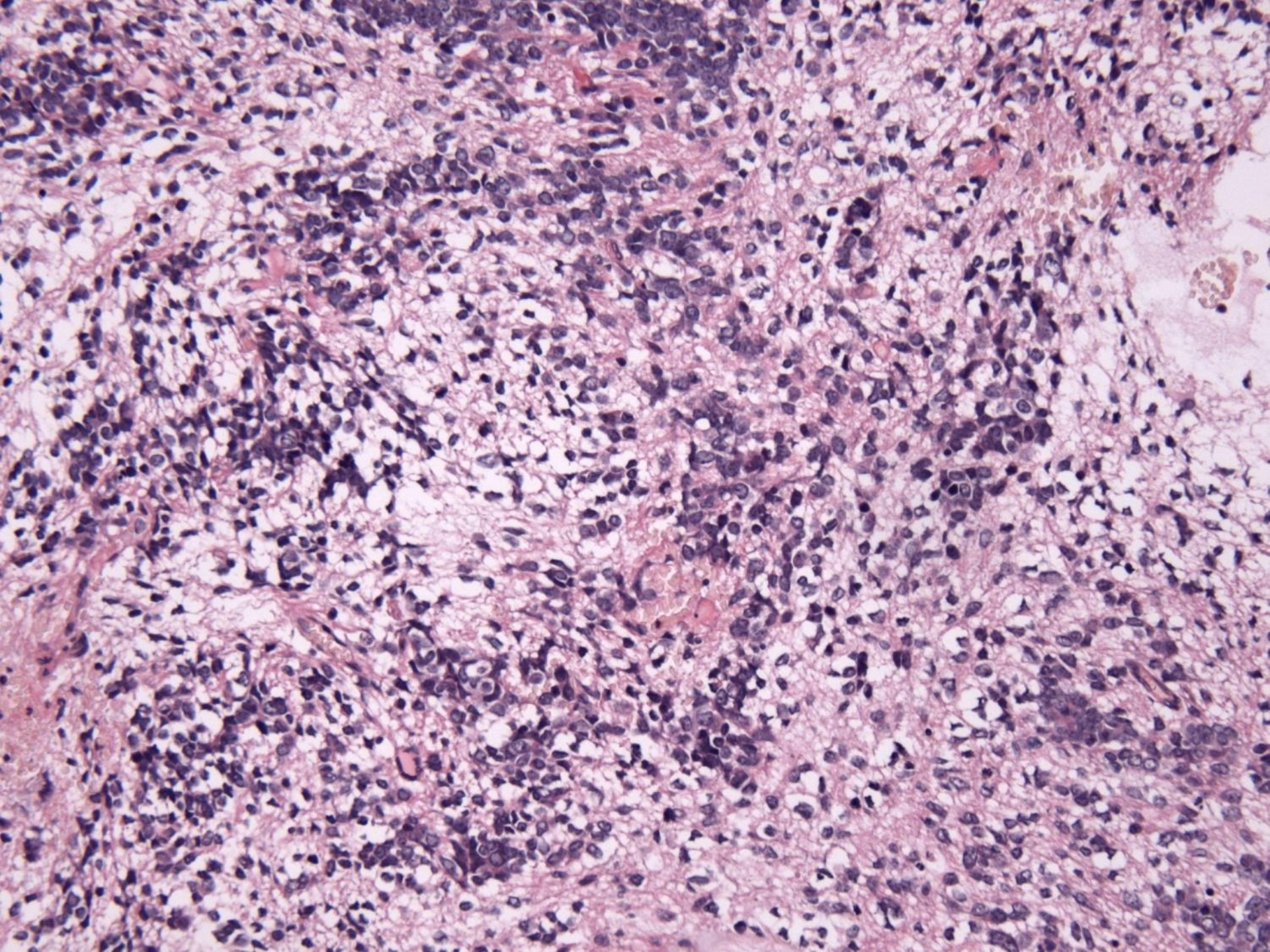


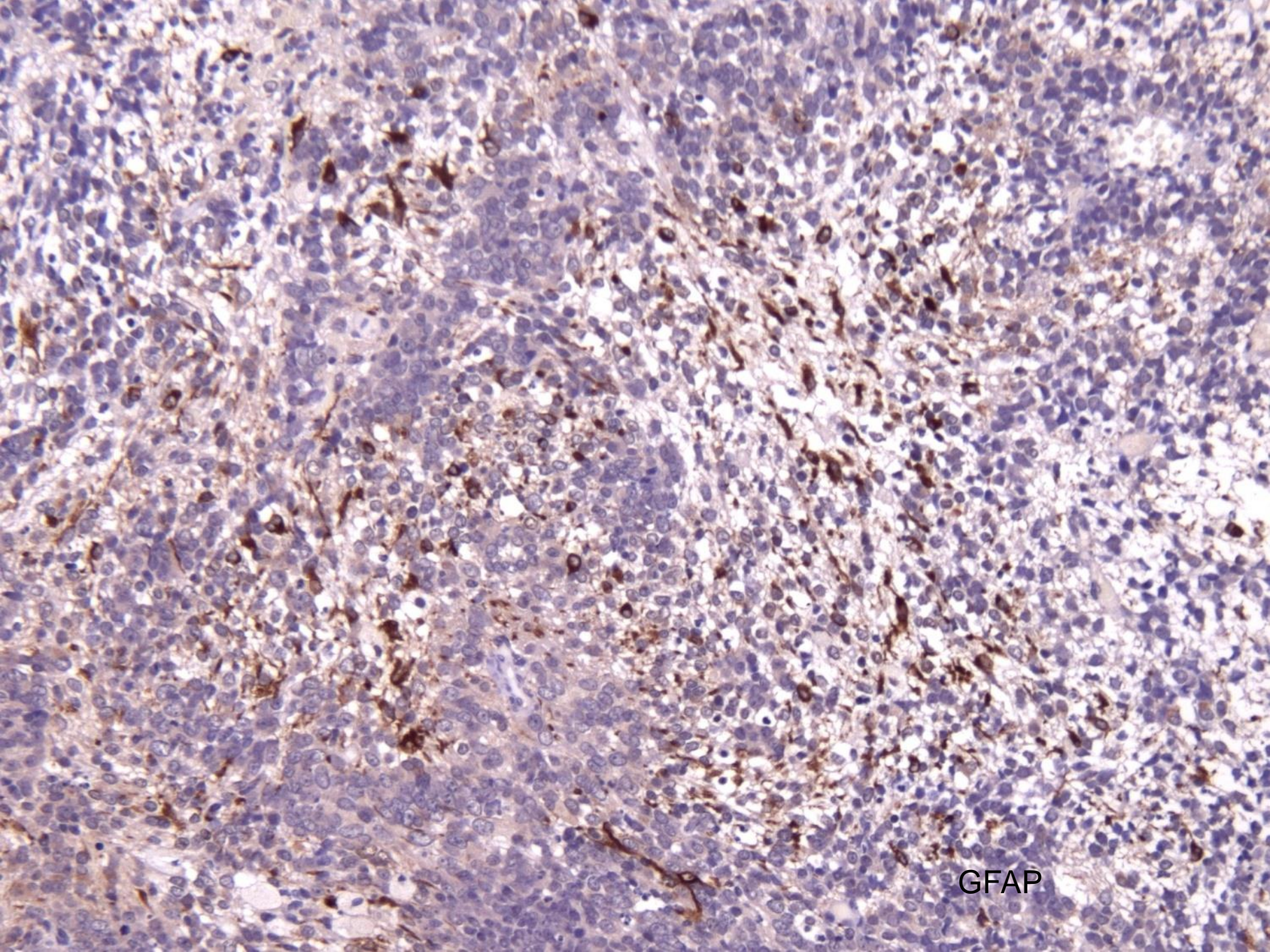




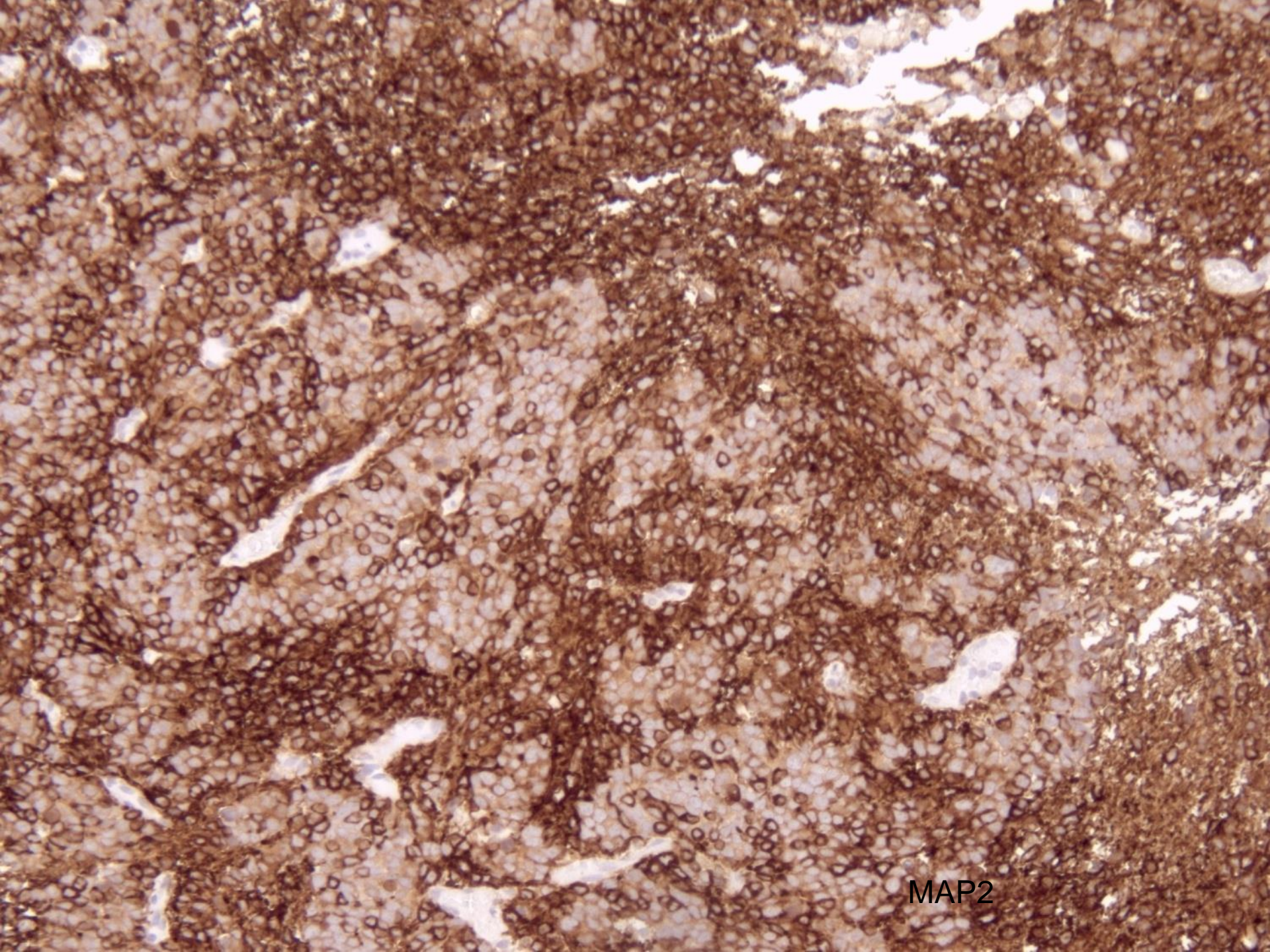




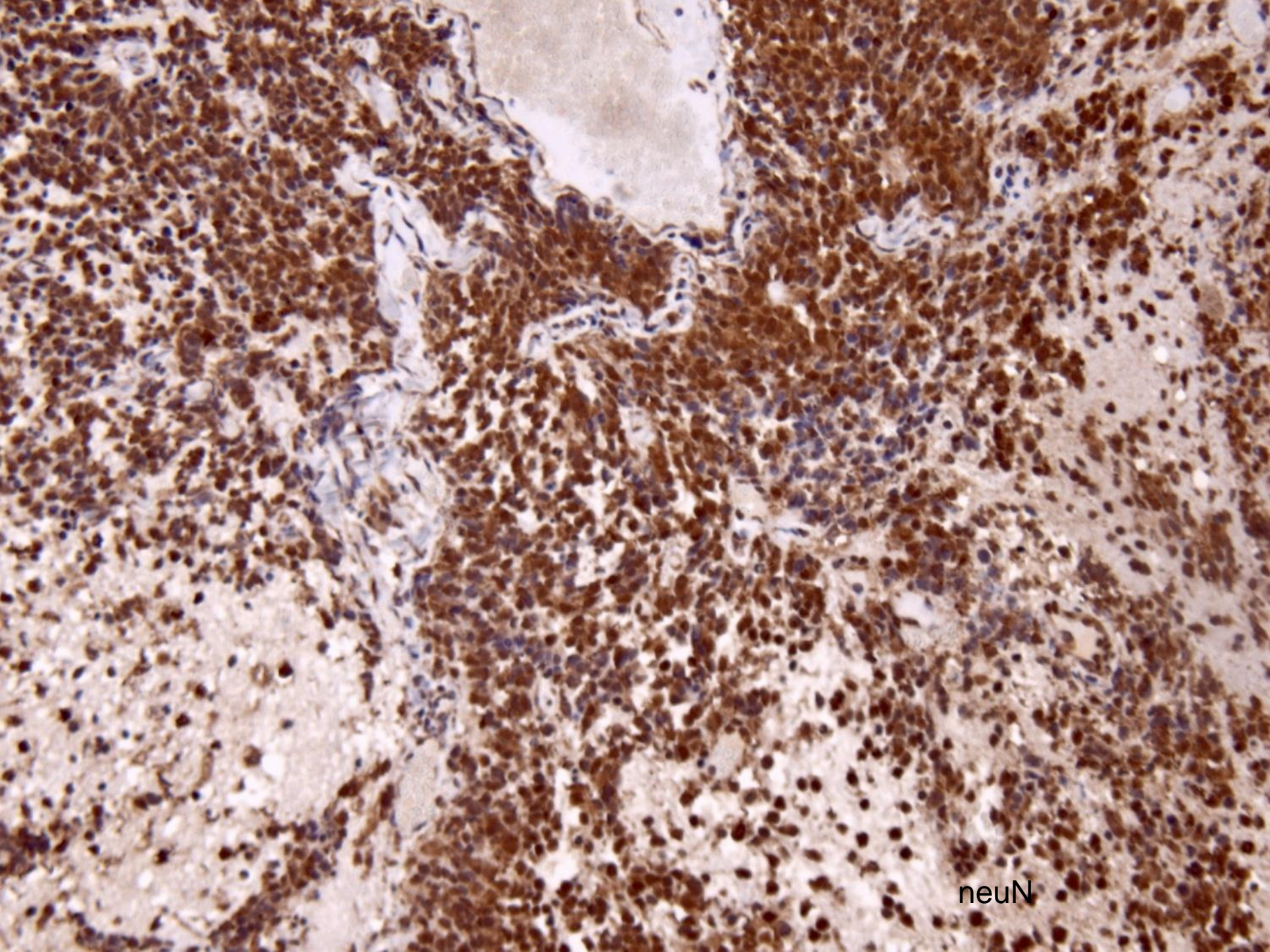




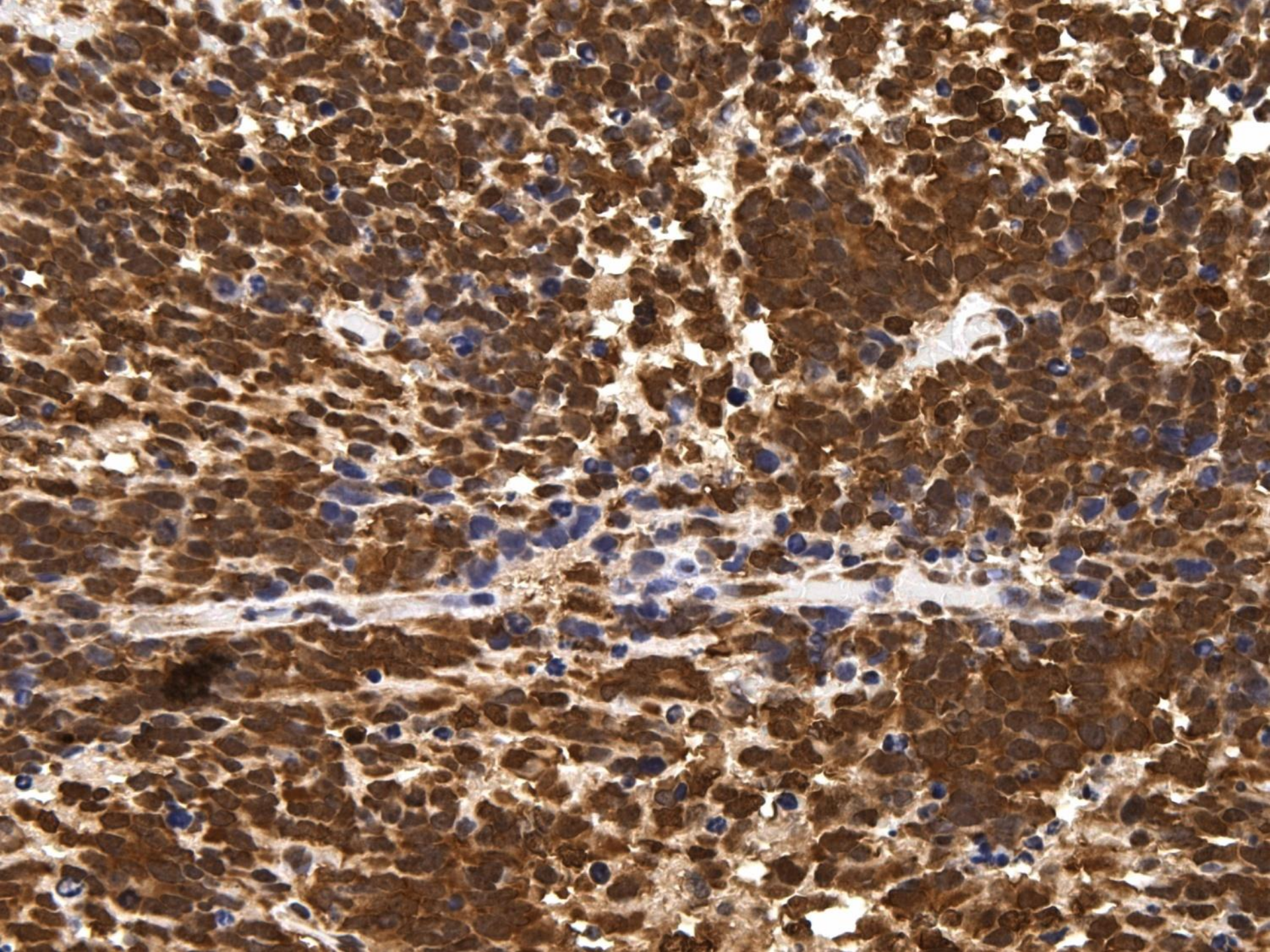
GFAP

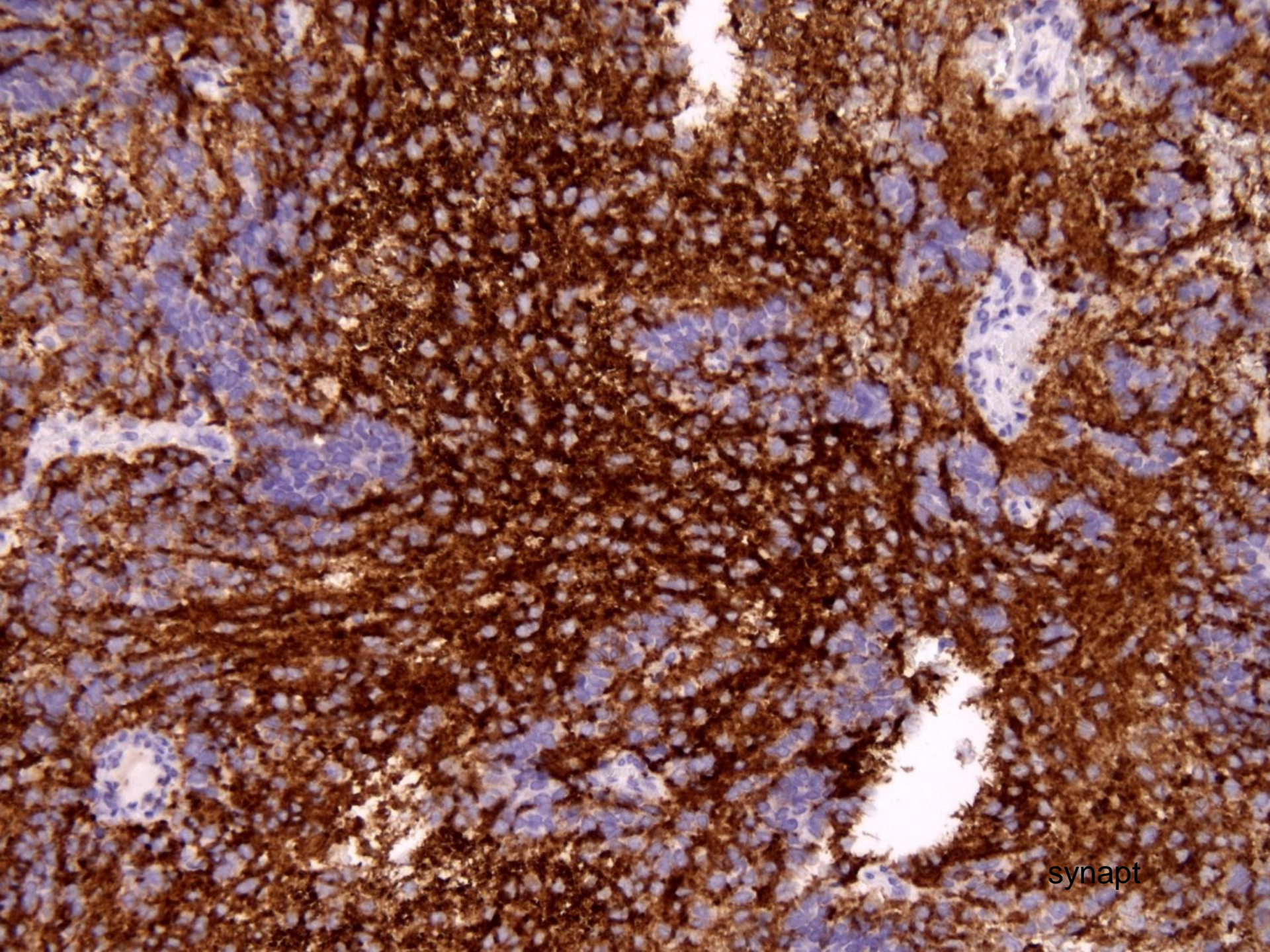


MAP2



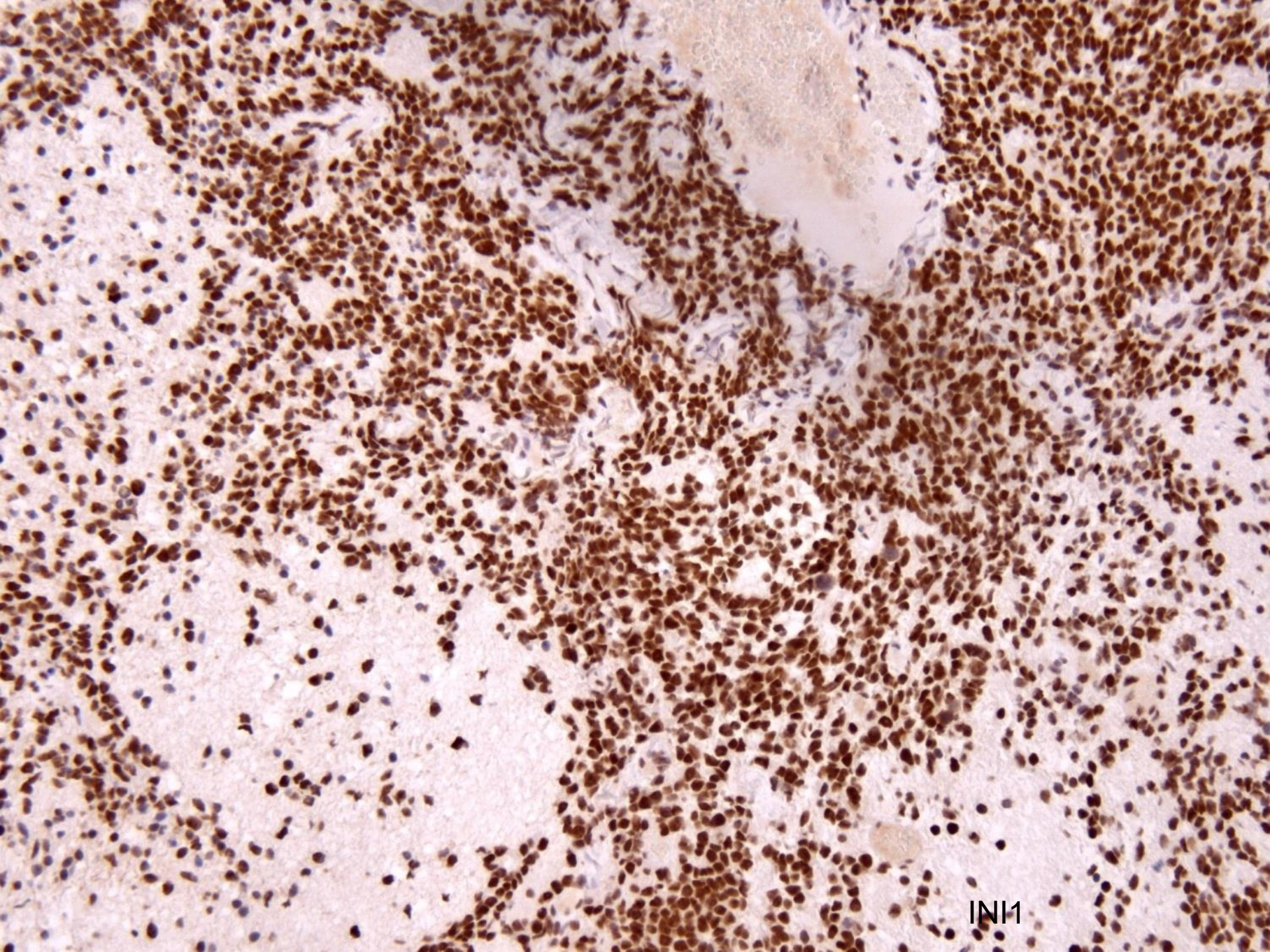
neuN



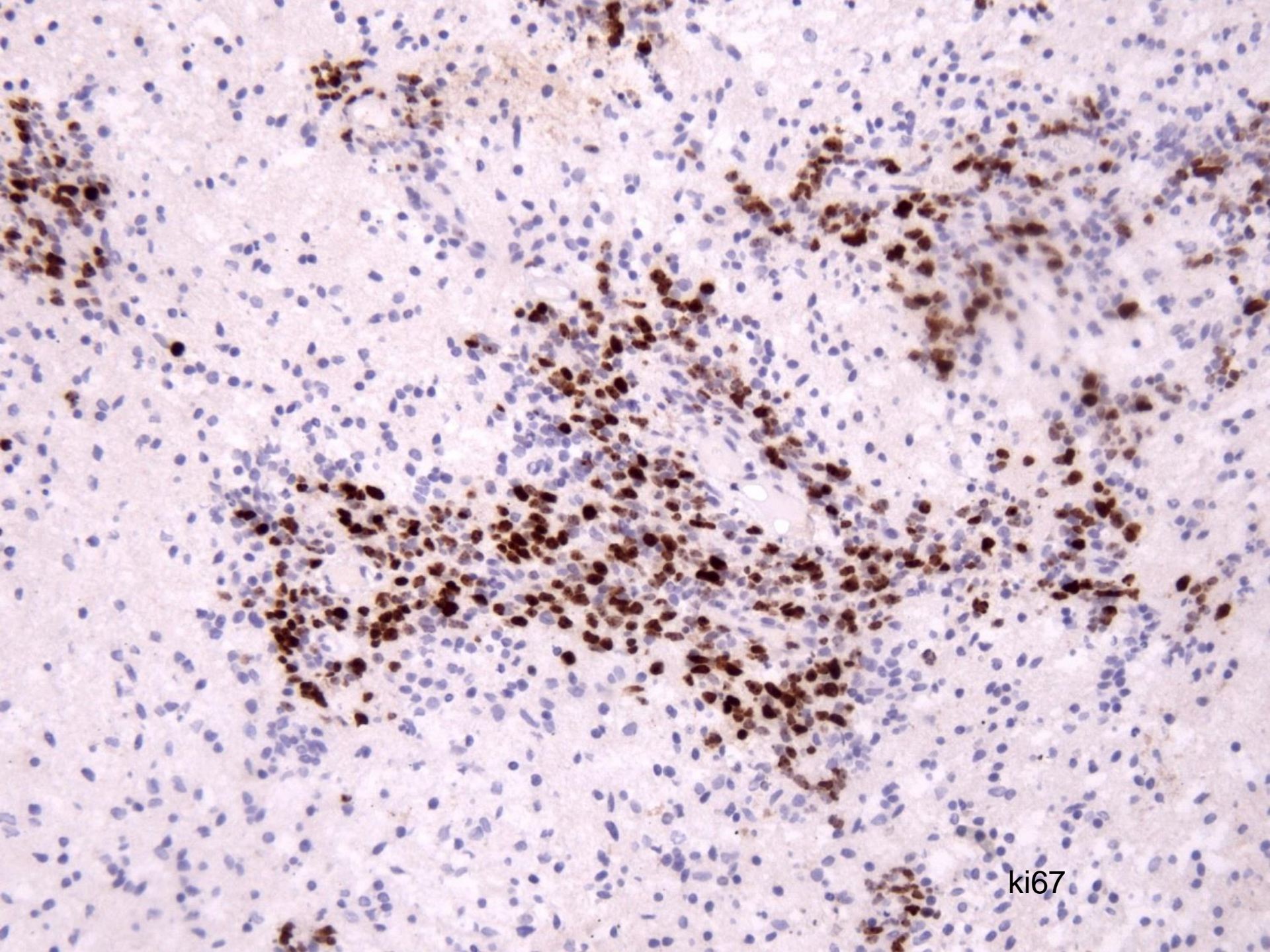


synapt

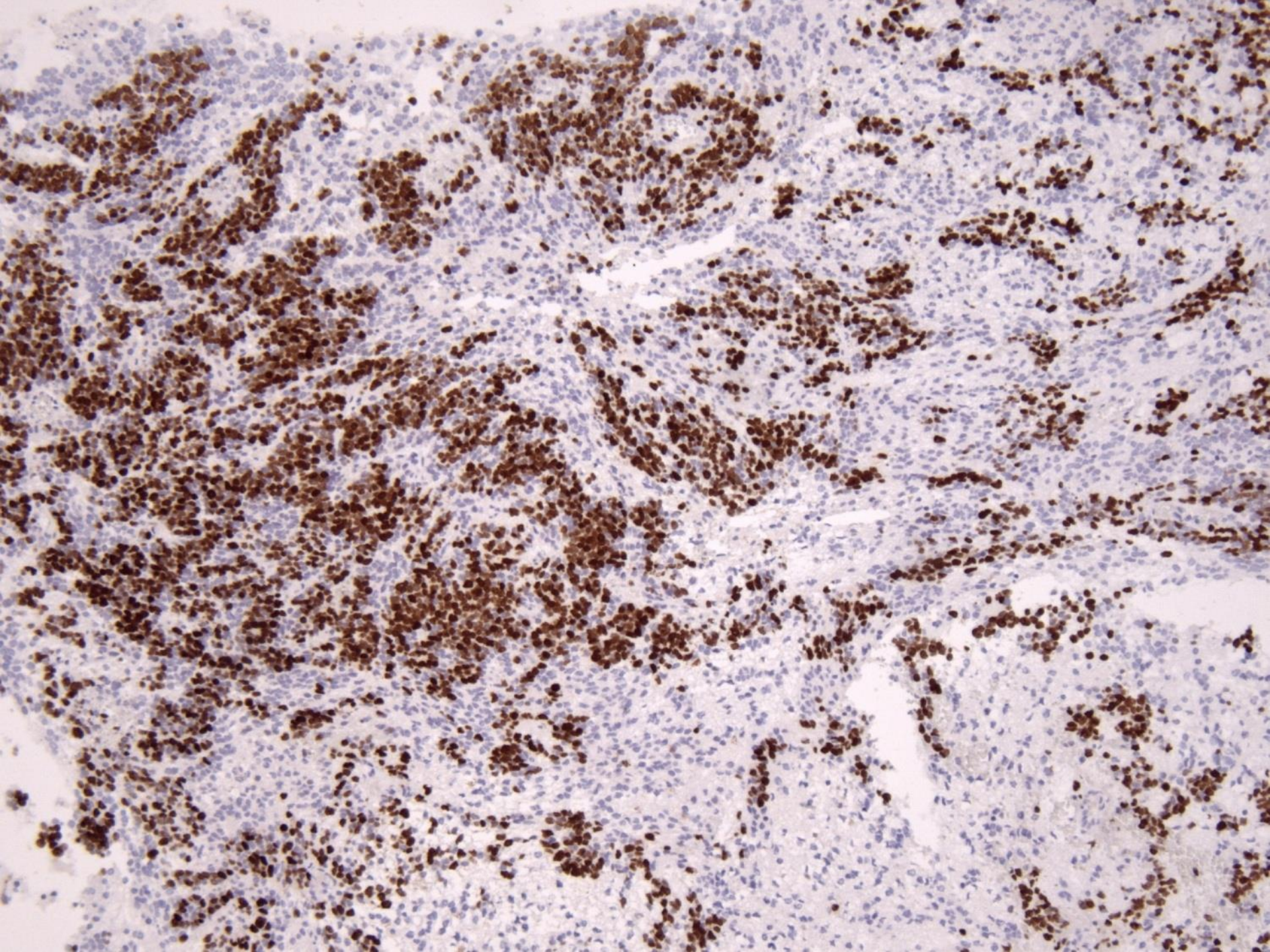




INI1



ki67



***embryonální tumor s abundantním  
neuropilem a pravými rozetami - ETANTR,  
WHO grade IV.***

**C19MC alterovaný**

Chromozomální změny	Výsledek:
Amplifikace 19q13.42	Pozitivní

#### **ZÁVĚR VYŠETŘENÍ:**

Ve vyšetřované nádorové tkáni (2E04) jsme prokázali chromosomální změny typické pro ETANTR.

Pro Ependyblastom (EBL) a embryonální tumor s abundantním neuropilem a pravými rozetami (ETANTR) byla prokázána specifická fokální amplifikace 19q13.42, která nenyla prokázána u jiných embryonálních nádorů CNS. Korshunov et al ji prokázal u 93% EBL a ETANTR případů. Stejnou změnu, fokální amplifikaci 19q13.42, prokázal u ETANTR ve své práci Nobusawa et al. (Korshunov A, et al. Acta Neuropathol. 2010, Nobusawa S, et al. Pathol Int. 2014)

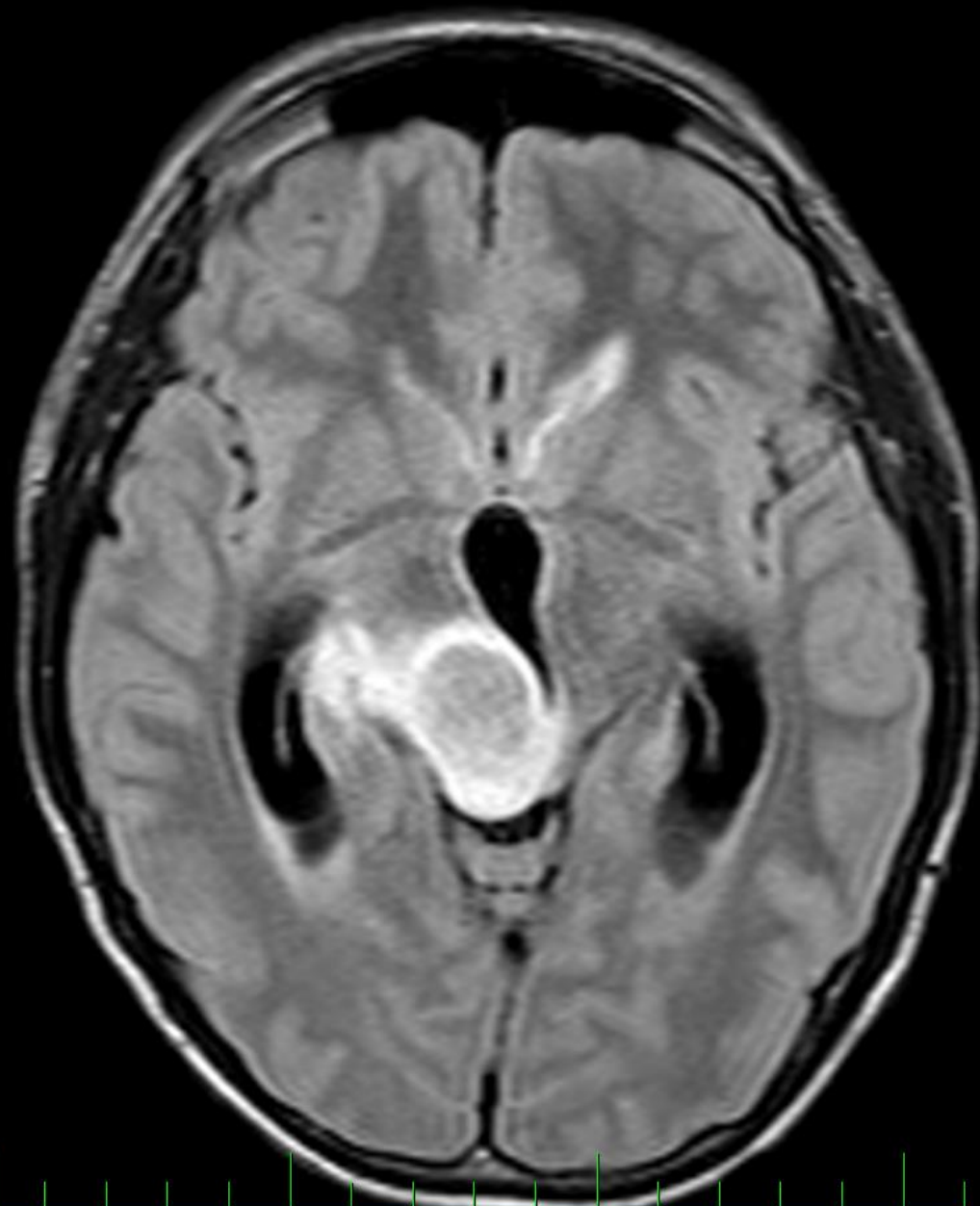


# 14 let, chlapec

## MR mozku

- Ovalné lozisko 3,2 x 2,2 x 2,5 cm v pravem thalamu., zasahuje do obl. mezencefala, s liquidnim podilem
- V lozisku nenin pritomna restrikce difuze.
- supratentorialni hydrocefalus,
- dif. dg. low grade gliom

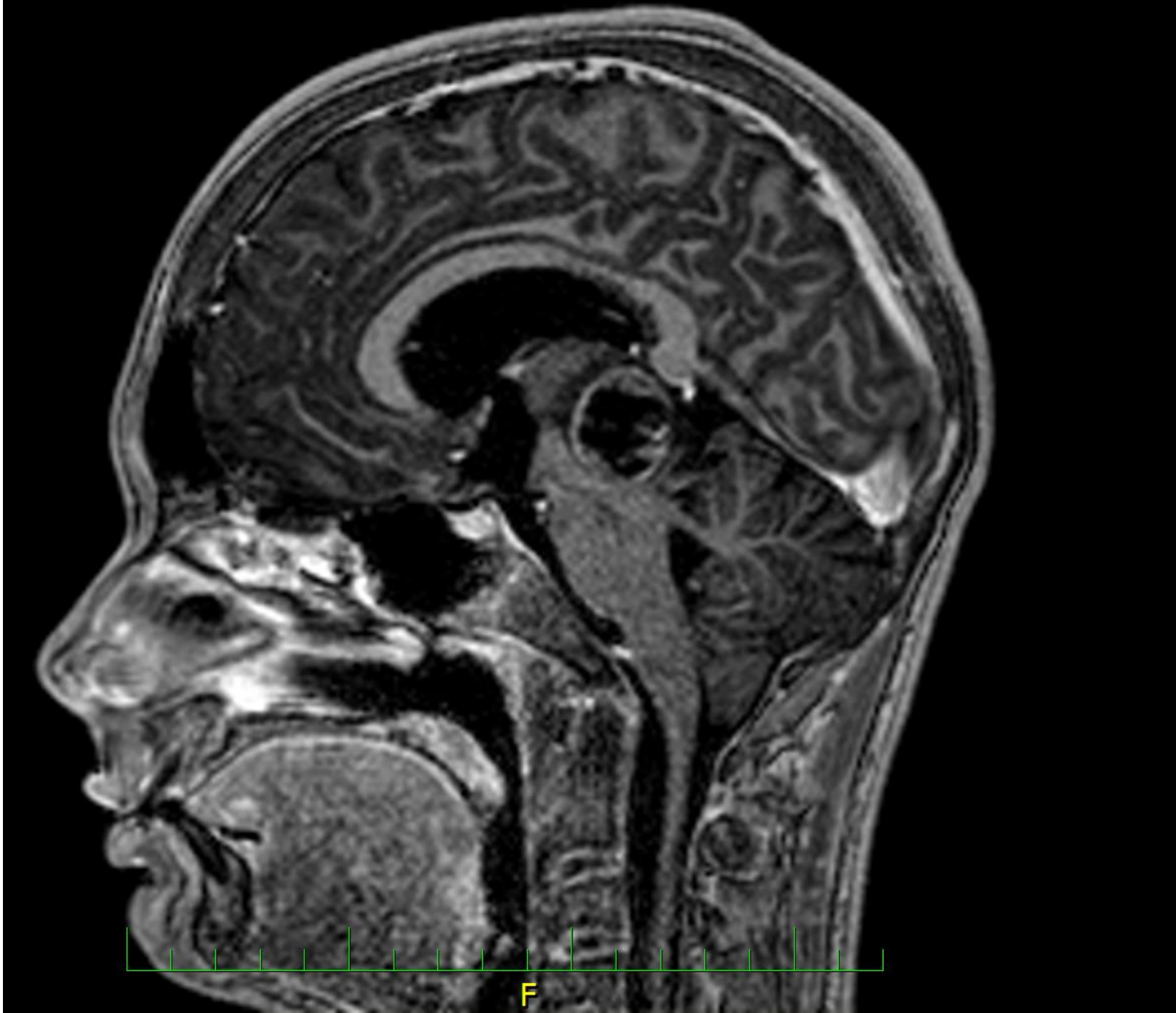
A



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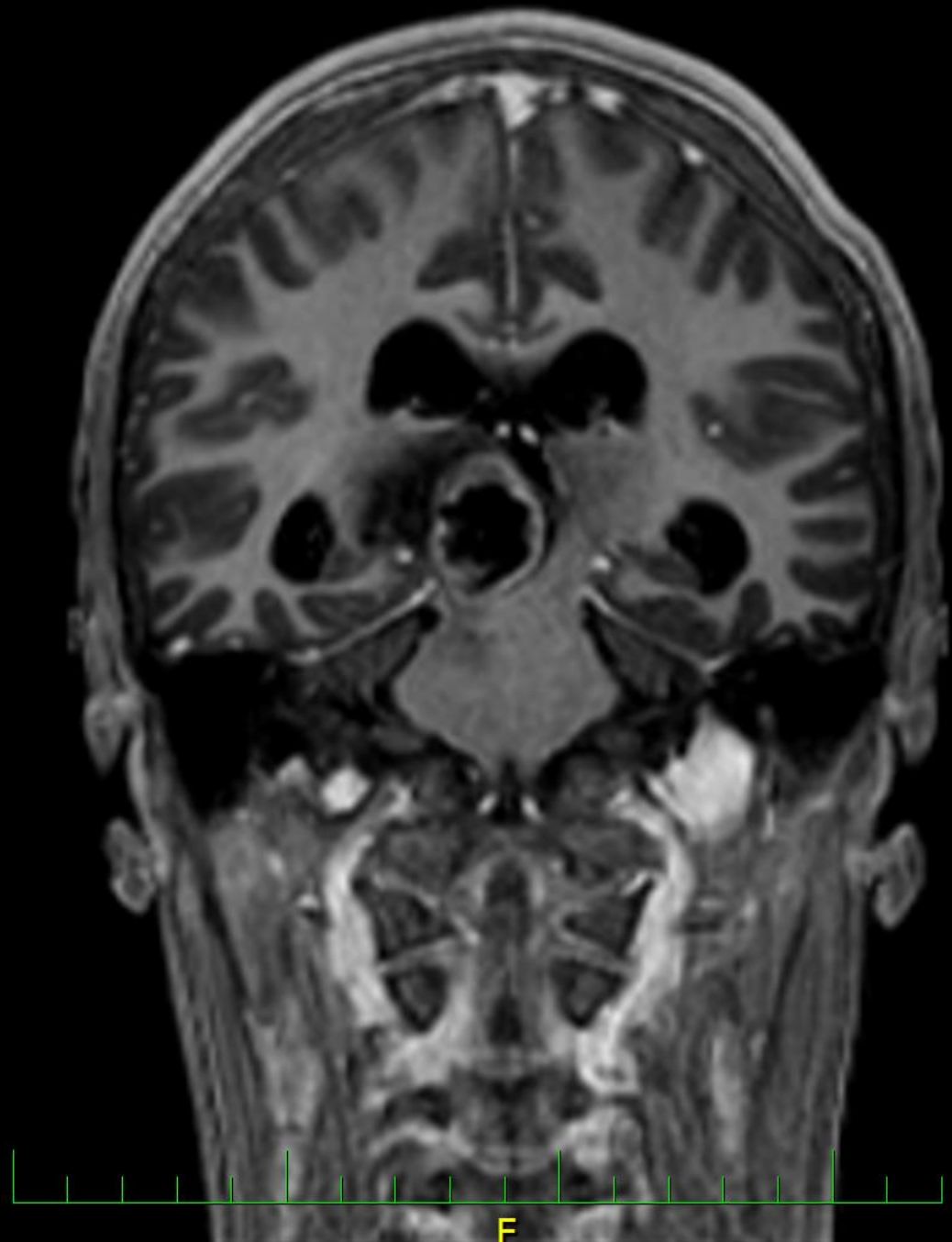


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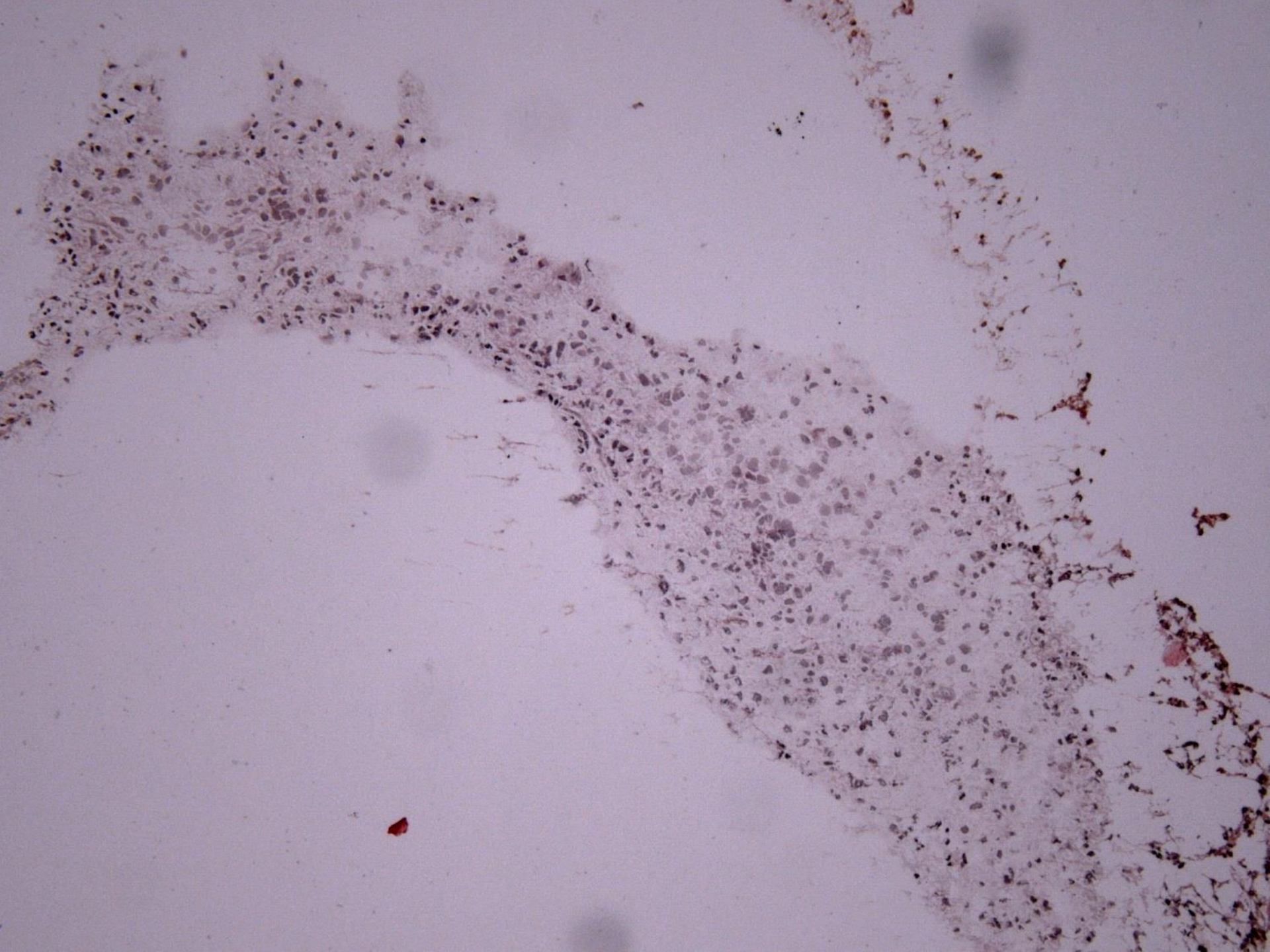


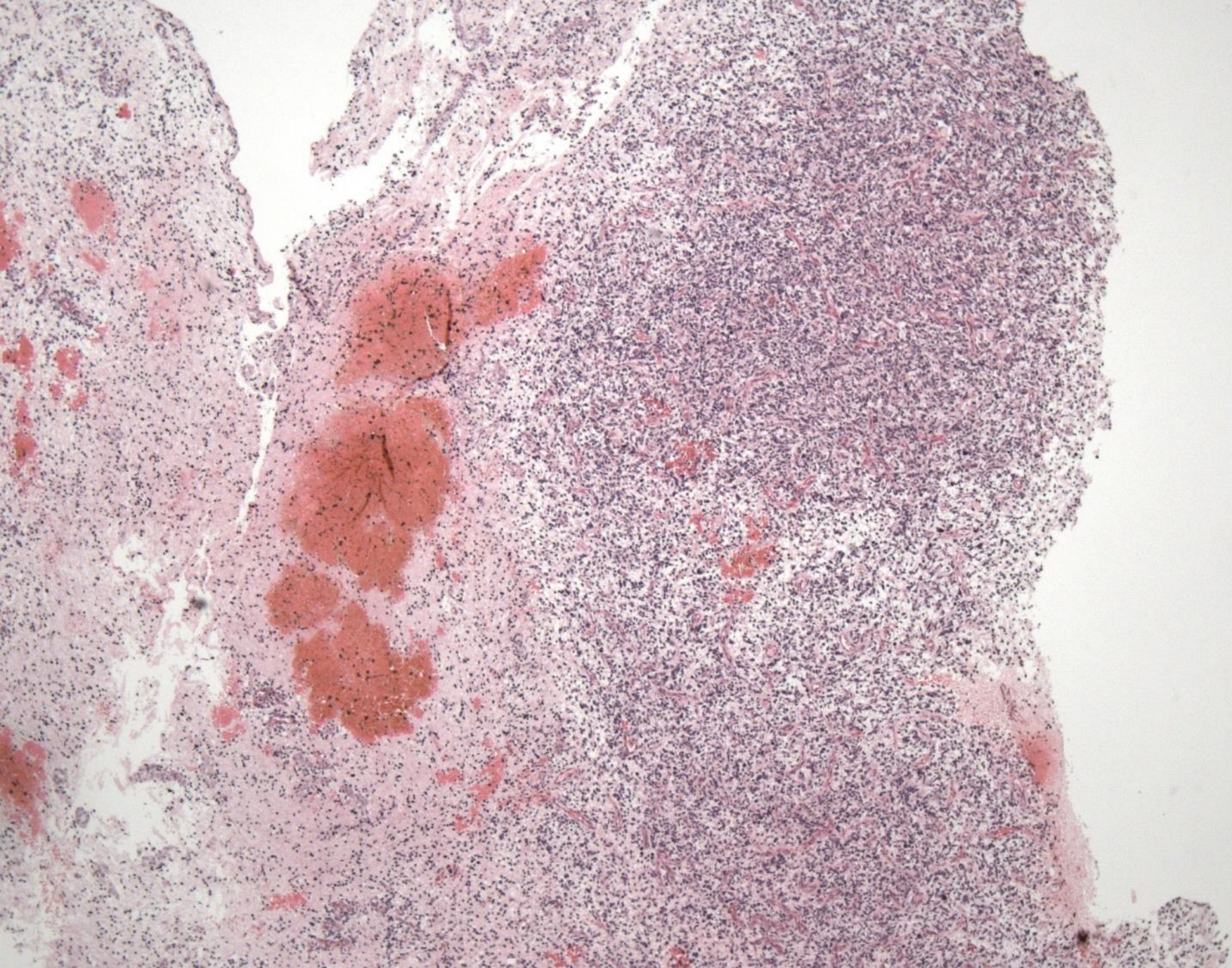
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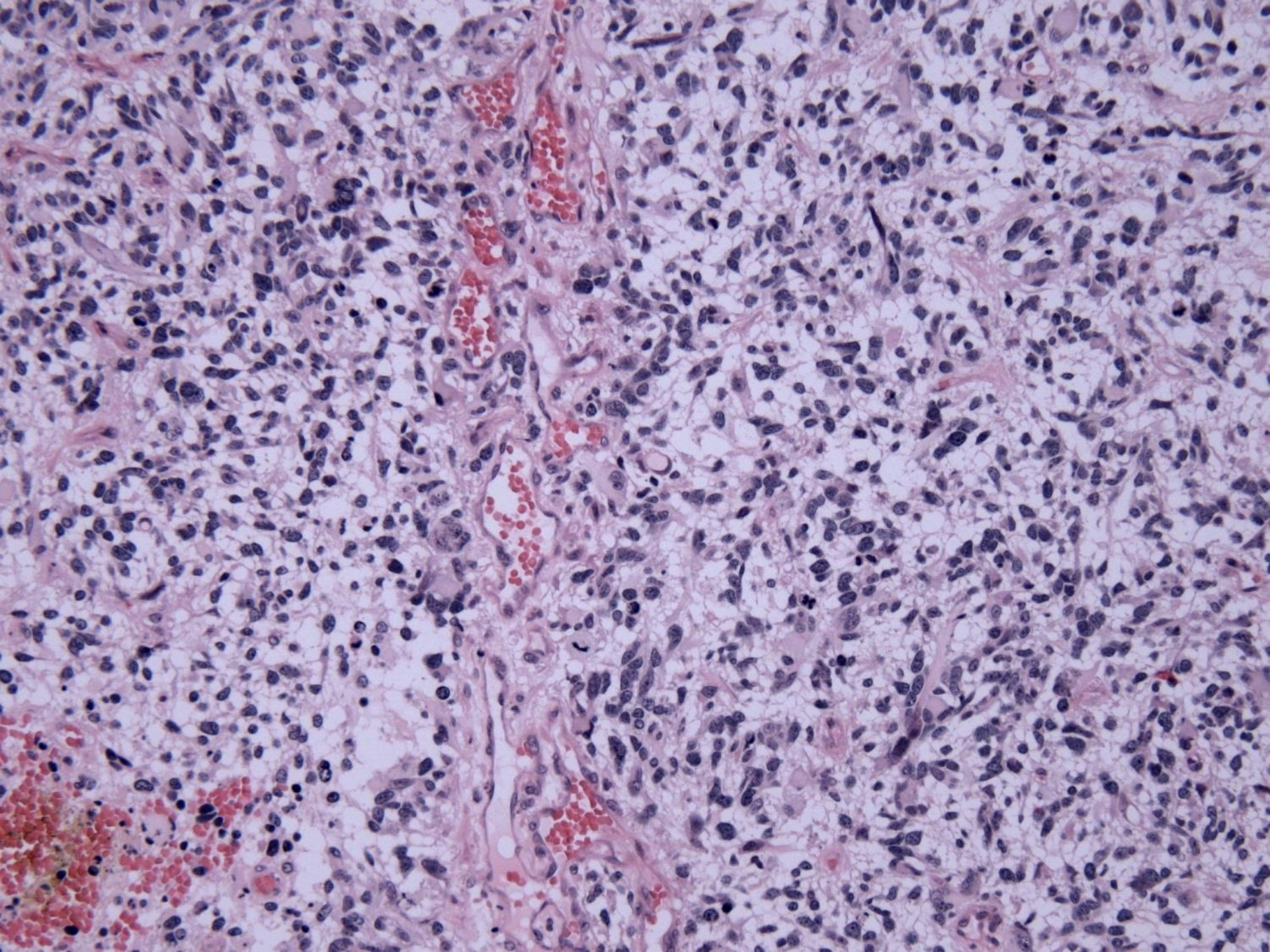
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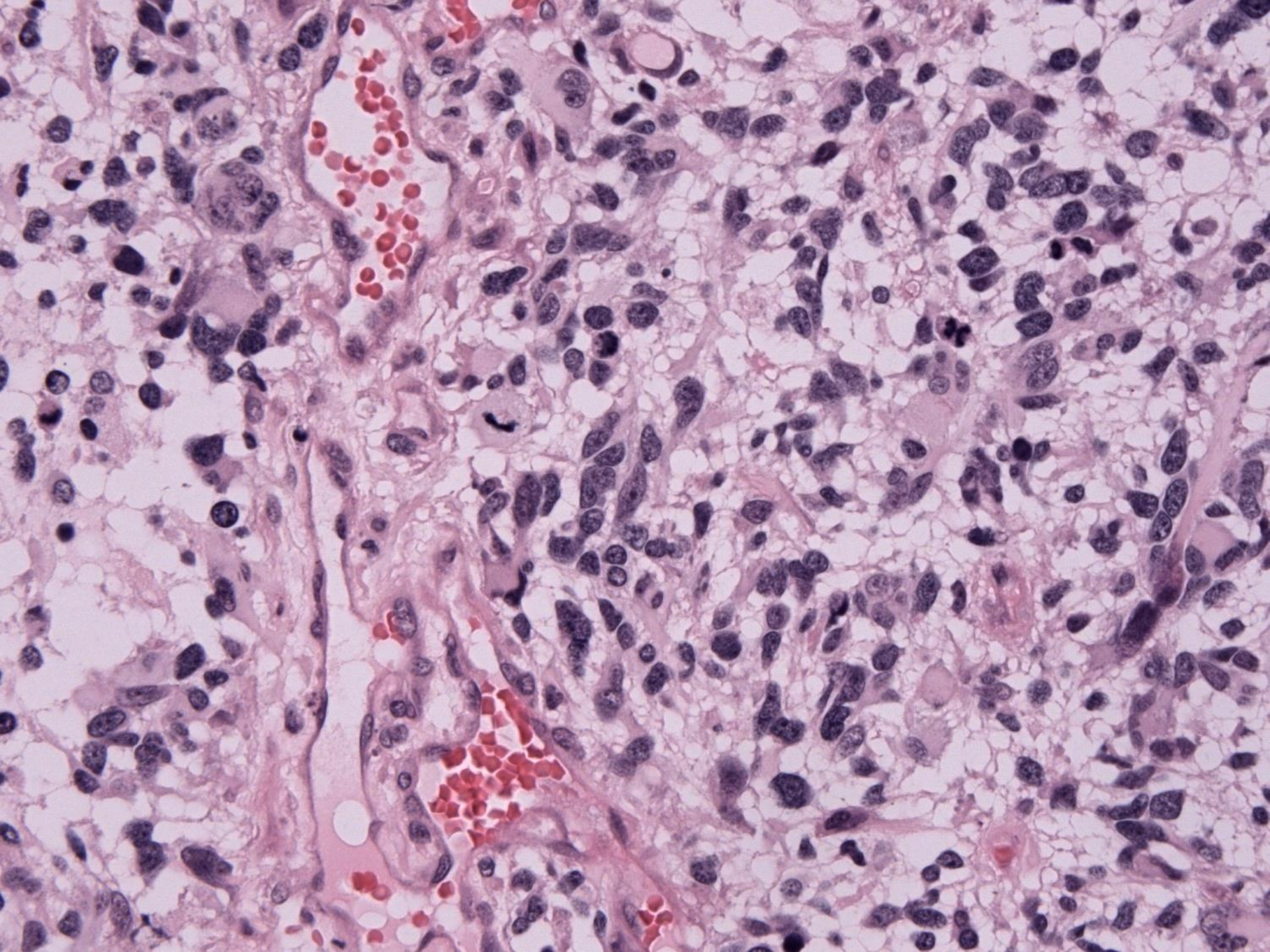


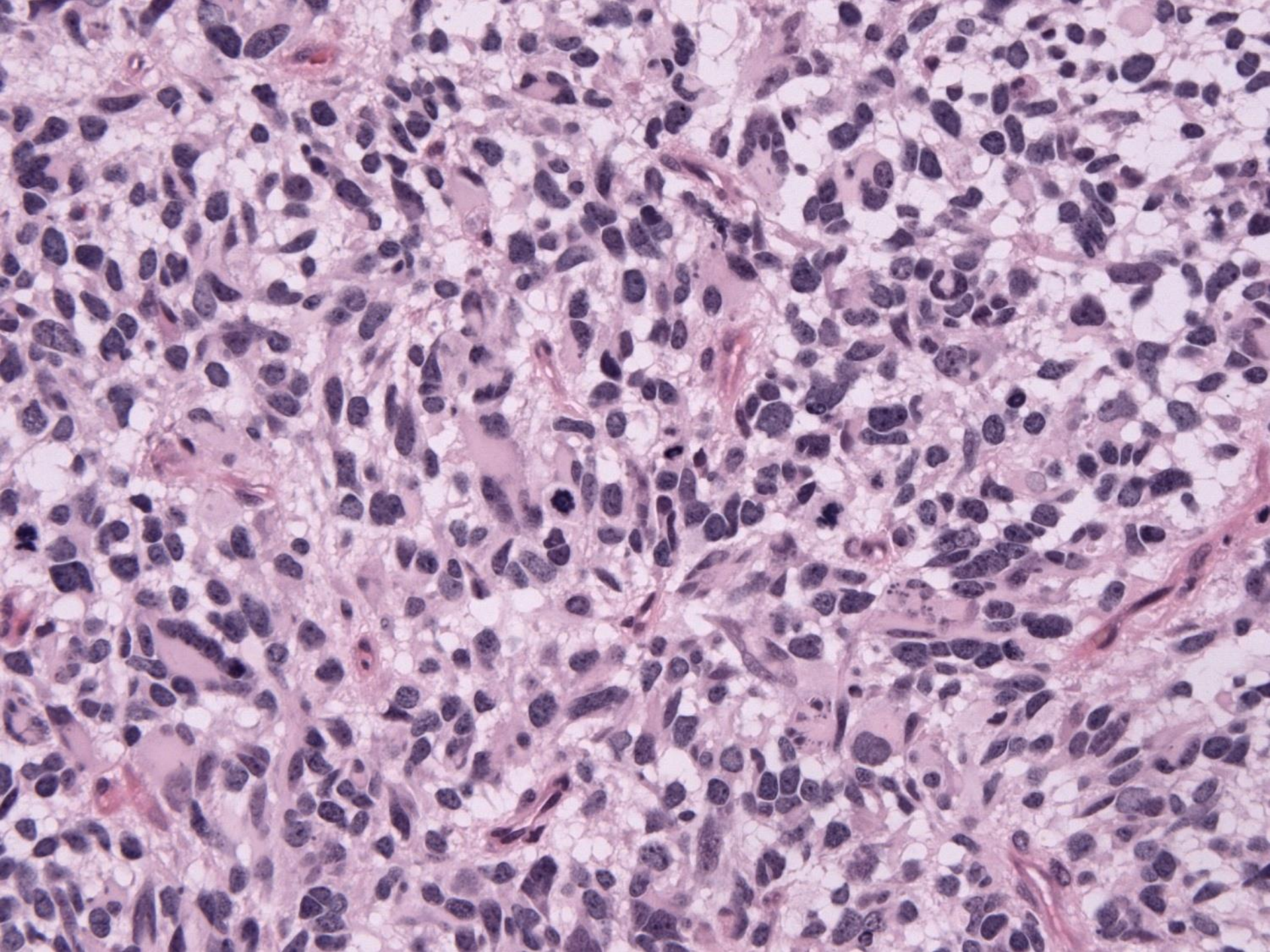
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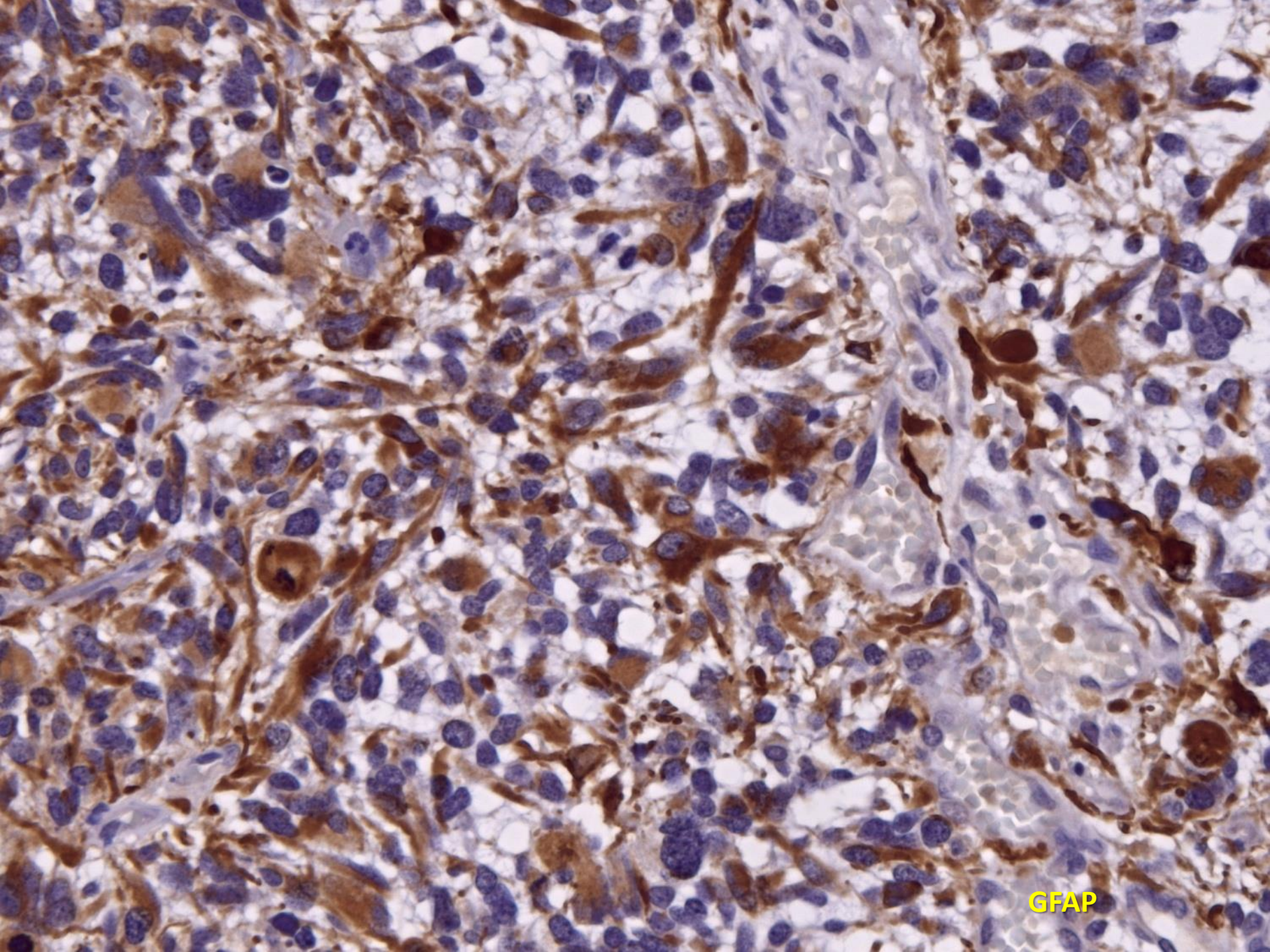






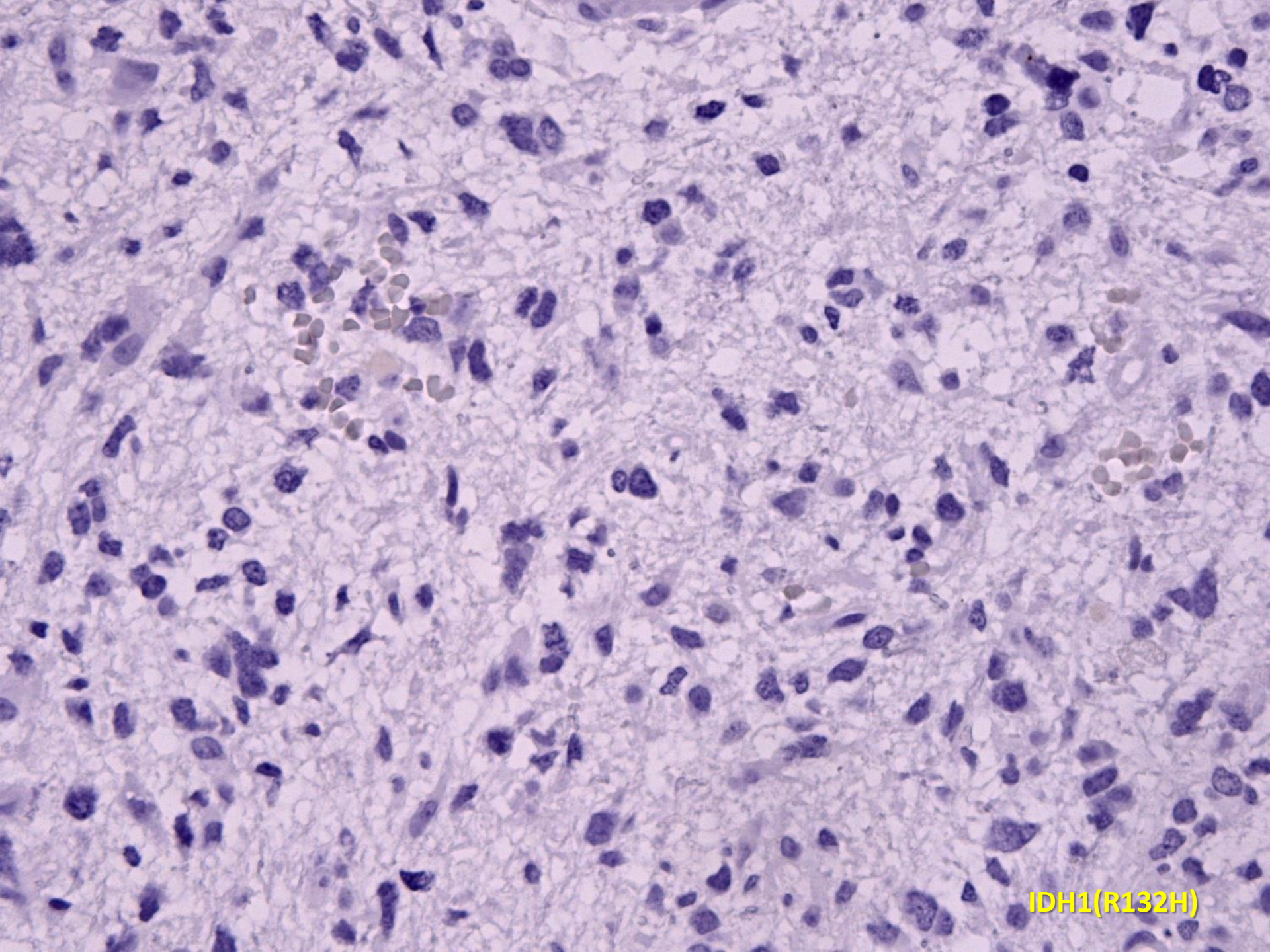




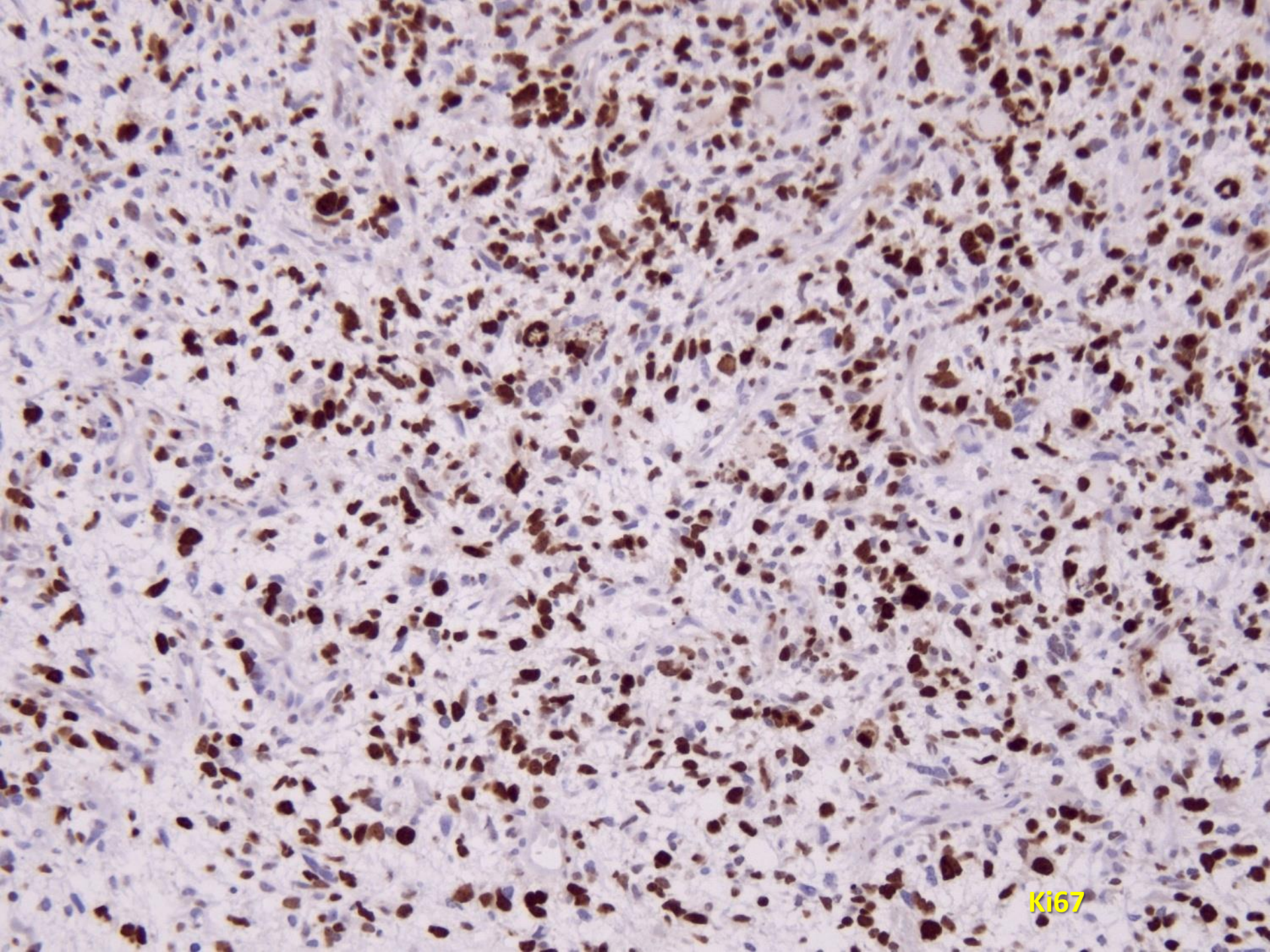


GFAP





IDH1(R132H)



Ki67

*Souhrn provedených molekulárně patologických vyšetření:*

- Nebyla zjištěna přítomnost aktivační mutace genu BRAF V600E
- Nebyla detekována žádná z hot spot mutací genu IDH1 a IDH2
- Byla detekována **mutace K27M H3.3 (H3F3A) exonu 1**

*Z á v ě r:*

Nález odpovídá *high grade gliomu, který má morfologii glioblastomu.*

Vzhledem k průkazu mutace H3 K27M:

**difúzní středočárový gliom, grade IV, K27M mutovaný.**

**5182-18**

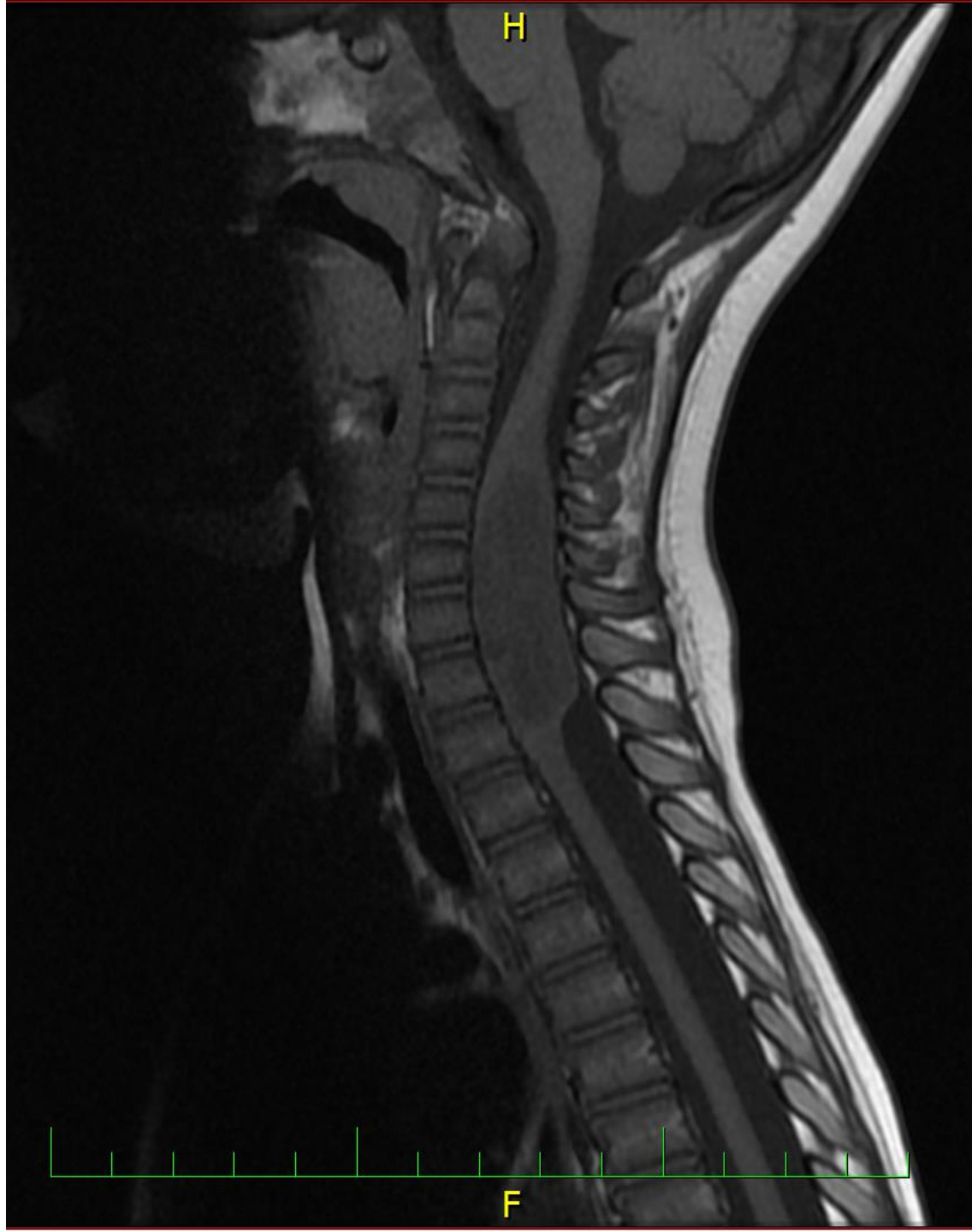
## **Intramedulární tumor C5-Th3**

v prosinci jí začaly bolesti krční míchy, ukazovala si dozadu na krka a stěžovala si na bolesti , plačtivá, nedokázala otočit krkem, při bolestivých záchvatech i nejistá chůze o široké bazi MR páteře s nálezem intramedulárního nádorového ložiska C4-Th3 s cystickou složkou s přilehlým edémem, snímky při příjmu nejsou k dispozici

hospitalizována na Klinice dětské onkologie a hematologie v nemocnici Banská Bystrica, kde zaléčena retrovirová infekce a cestou konzultace na Neurochirurgické klinice v Bratislavě Kristína přijata k operačnímu výkonu na naší klinice



F





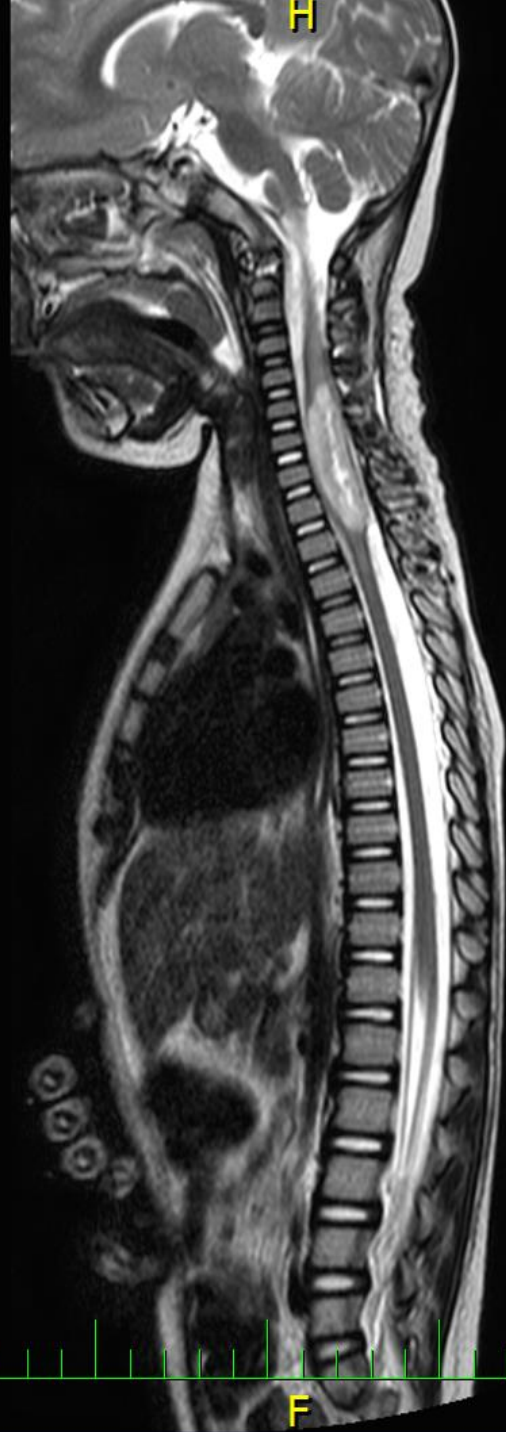
Indik. dg.: St.p. odstraneni intramedularniho tumoru C5-Th3. Poop. kontrola, k vyloucení rezidua či komplikace.

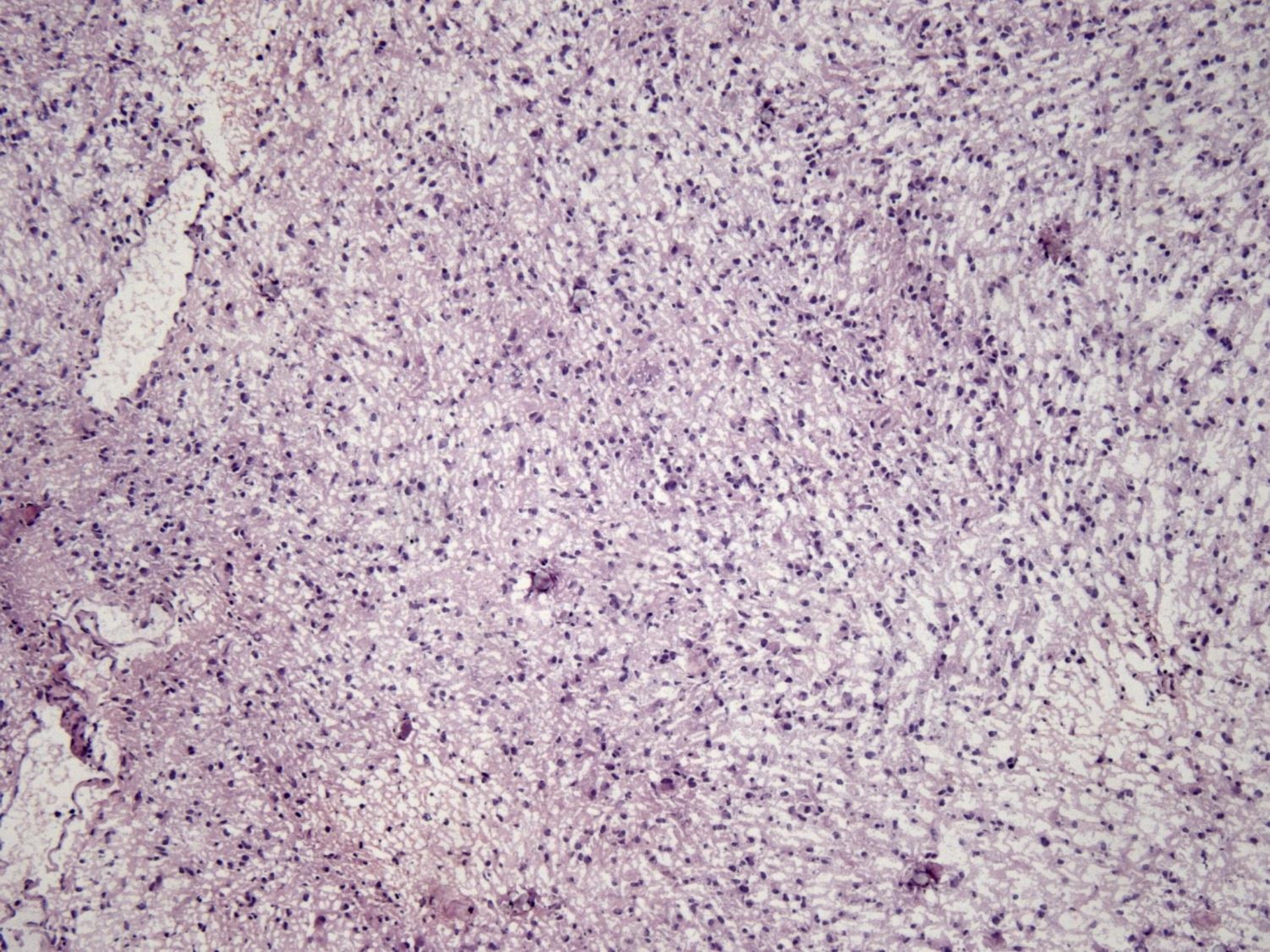
Vyšetřeno v základních anatomických rovinách v sekvencích 1W, T2W, STIR, DWI, VISTA, THRIVE nativně a po aplikaci 2 ml Prohance i.v., bez komplikací.

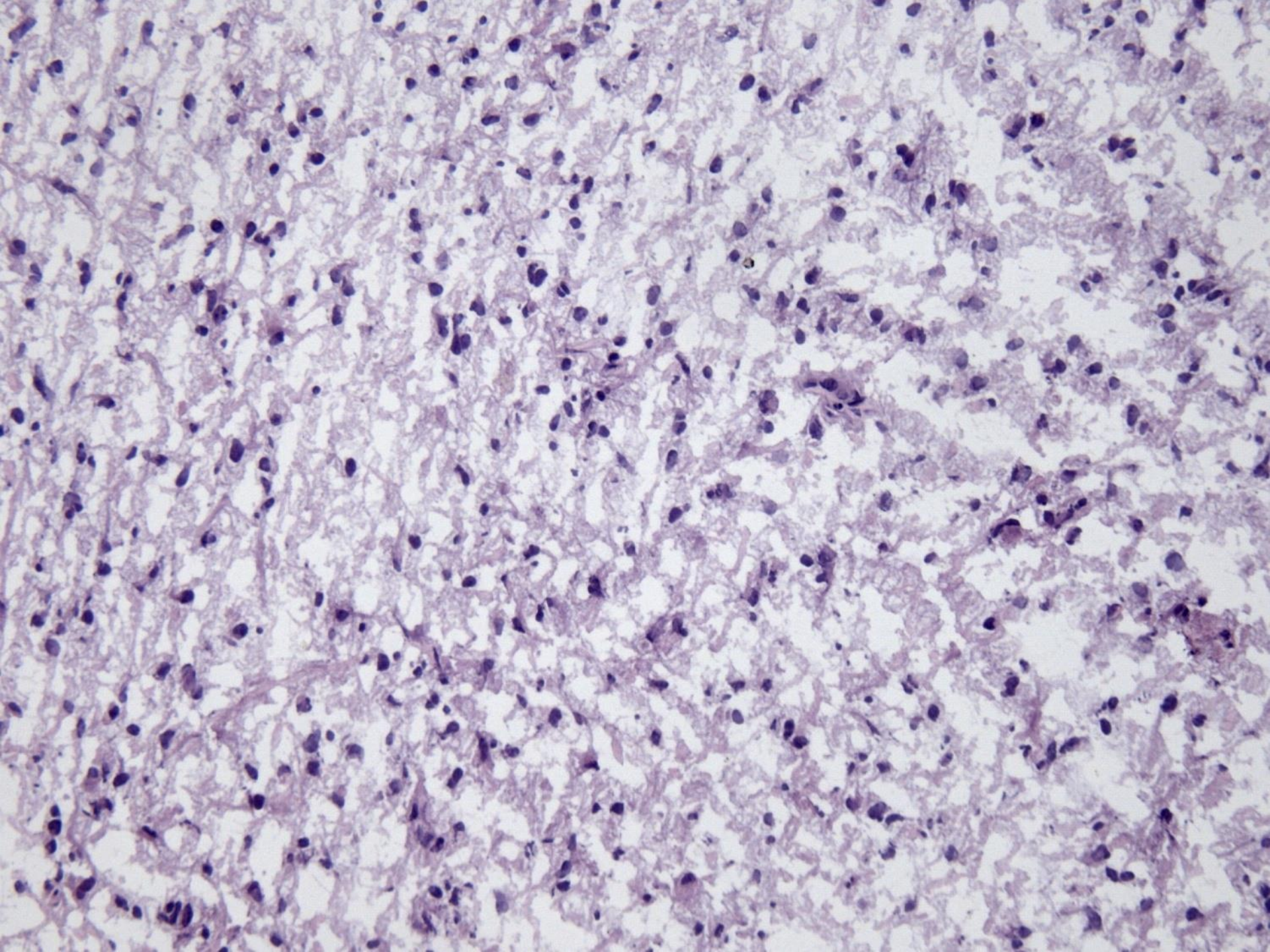
Stp. odstranění intramedularního TU. V úrovni C5 - Th2 se zobrazuje vretenovité rozšíření míchy, v rozsahu 16 x 12 x 45 mm. V T2 a STIR je hyperintenzní, v T1 hypointenzní. Centrálně je patrná nepravidelná, pruhovitá hyperintenzita v T2 i STIR. Po aplikaci KL dochází k reziduálnímu syčení. Nejsou patrné známky restrikce difuze. V obl. zadních oblouku obratlu C5 - Th3 se zobrazují nehomogenní poop. změny. Jinak čerstvou patol. strukturu v páteřním kanálu neprokazují. Kónus kaudy uložen v úrovni L1. Kauda bez patologických změn. Zakřivení páteře v sagitální rovině je primerené. Obratlová těla mají normální tvar a výšku, jsou bez signálových změn. Meziobratlové prostory jsou primerené šířky, meziobratlové plotenky jsou bez signálových změn. Intervertebrální foramina jsou volná.

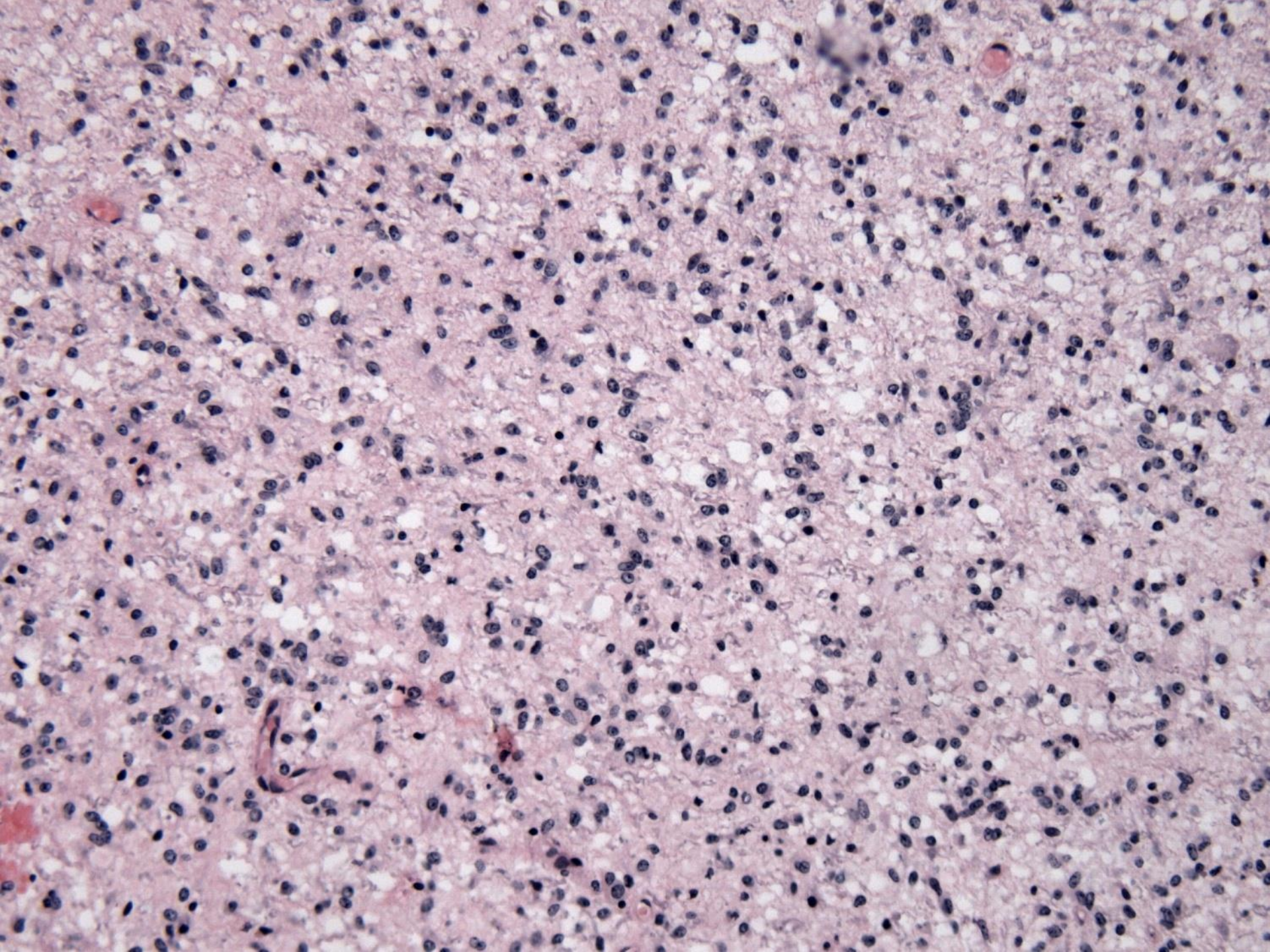
Závěr:

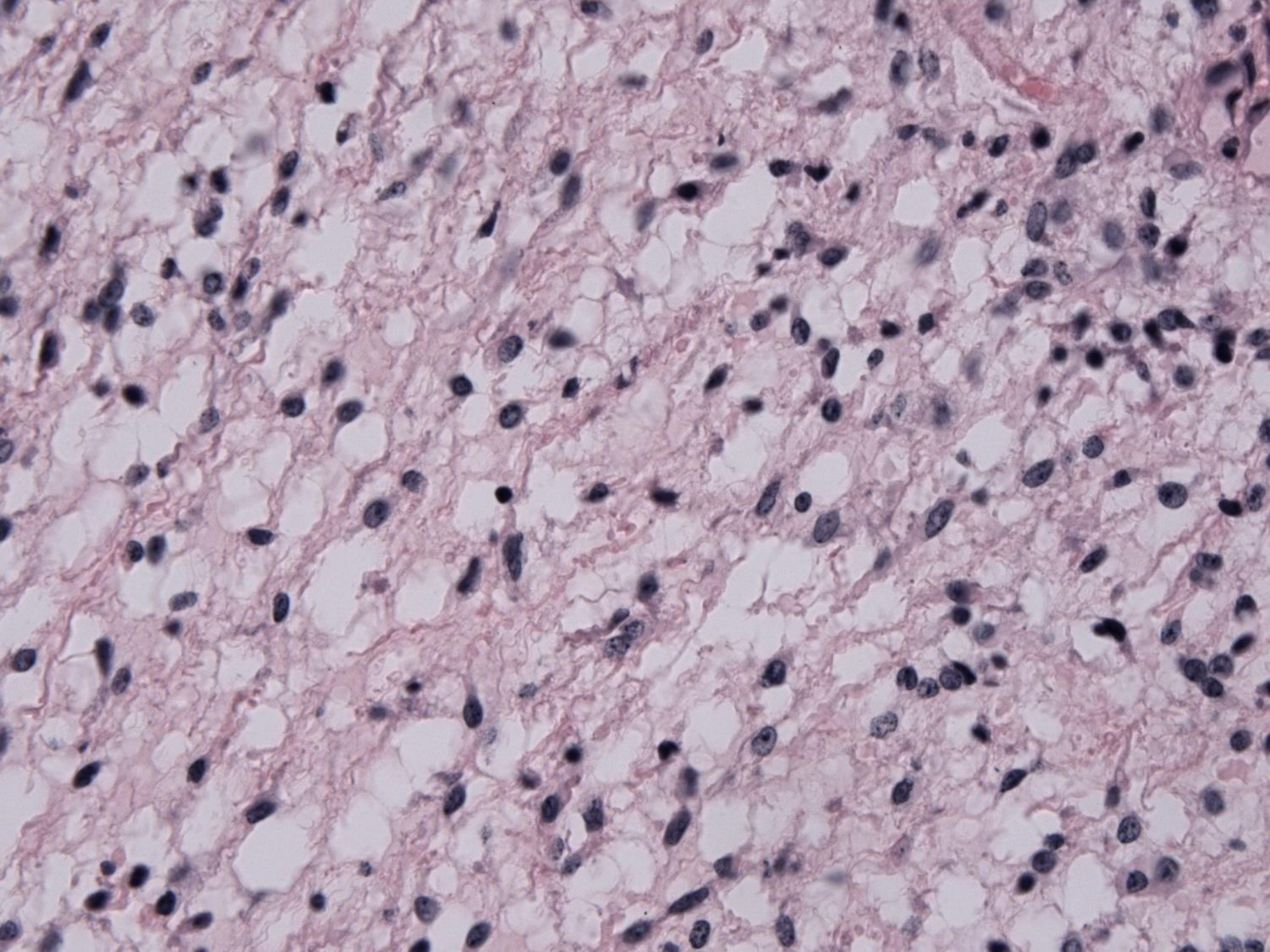


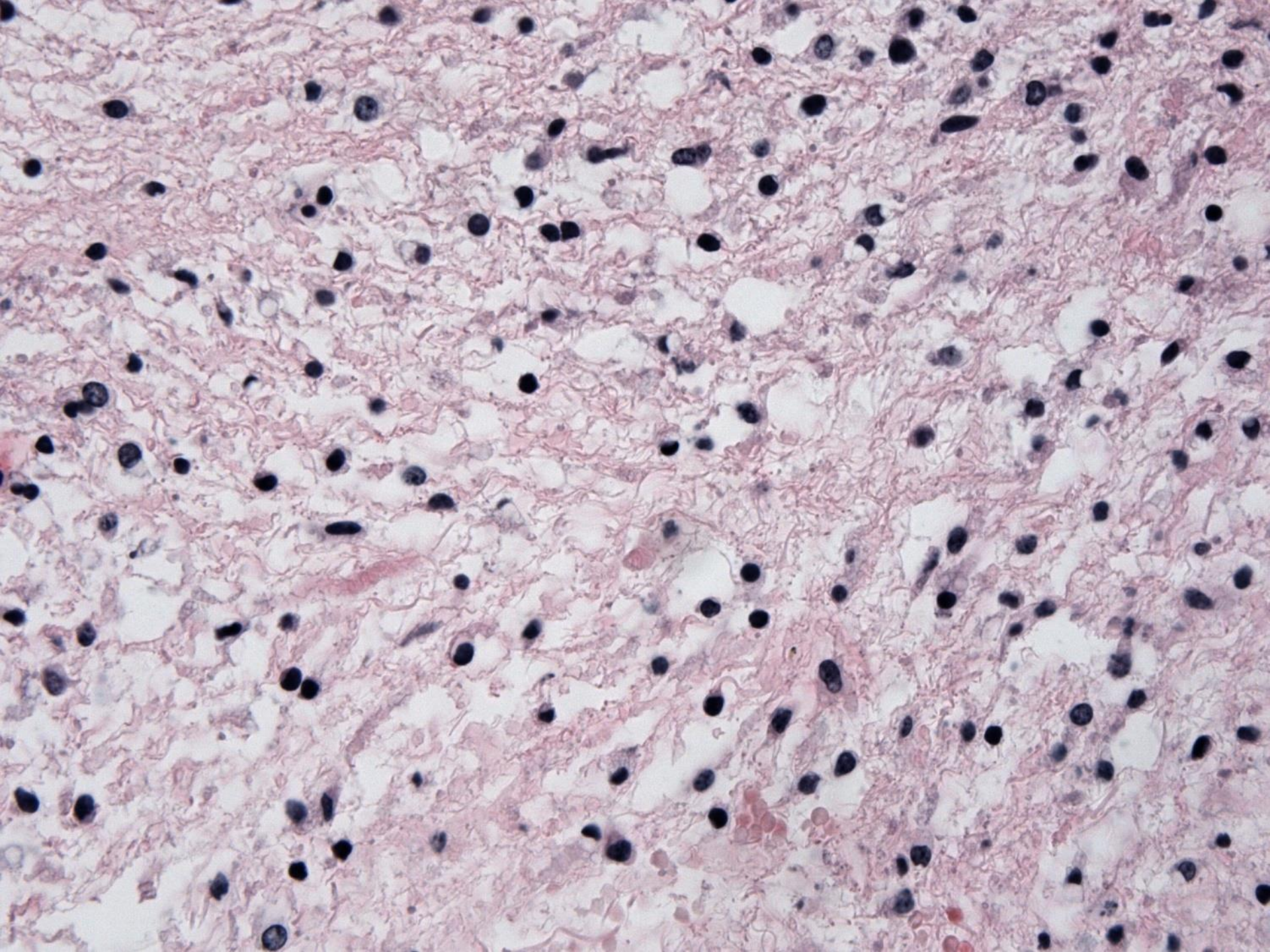


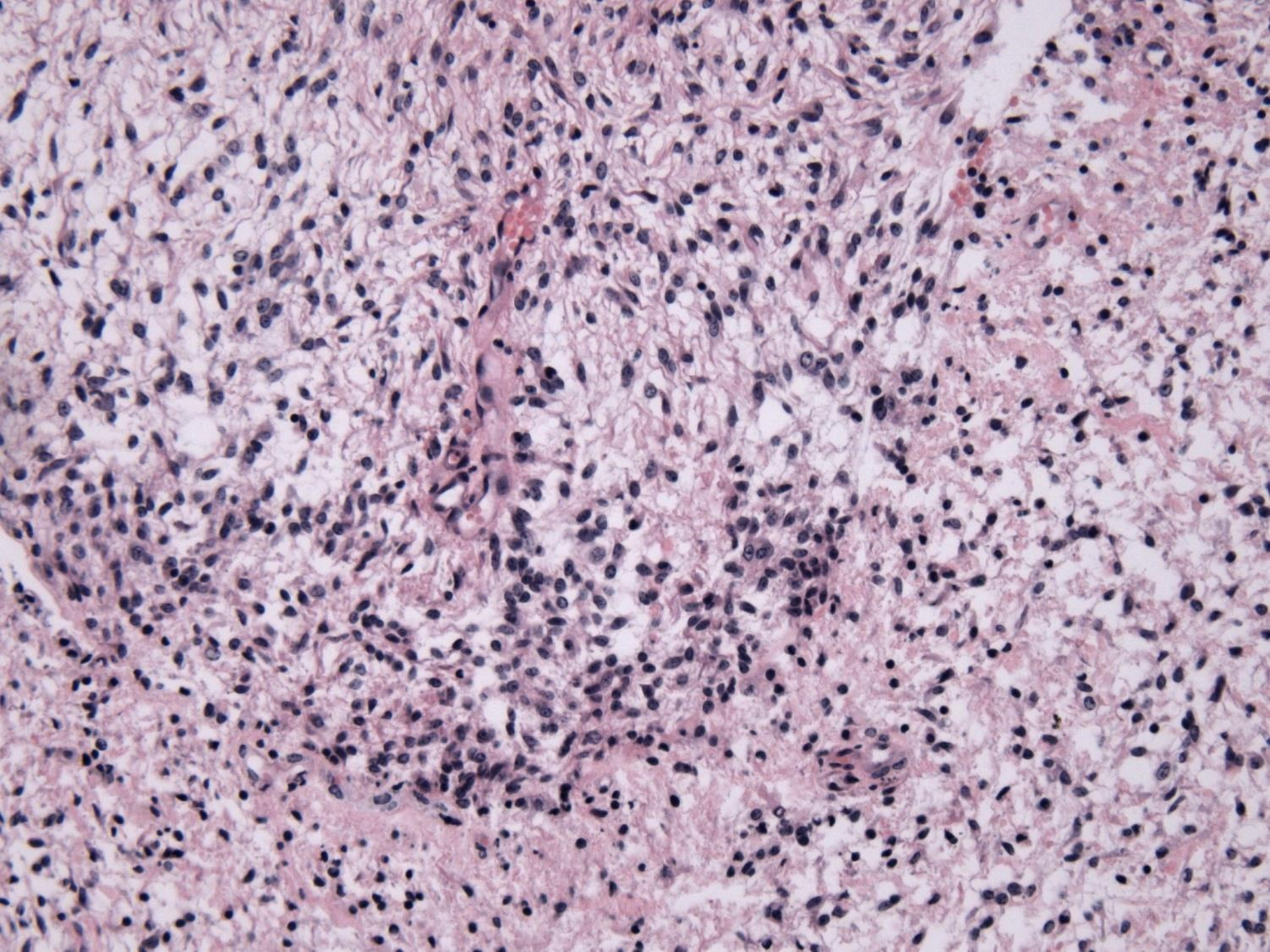


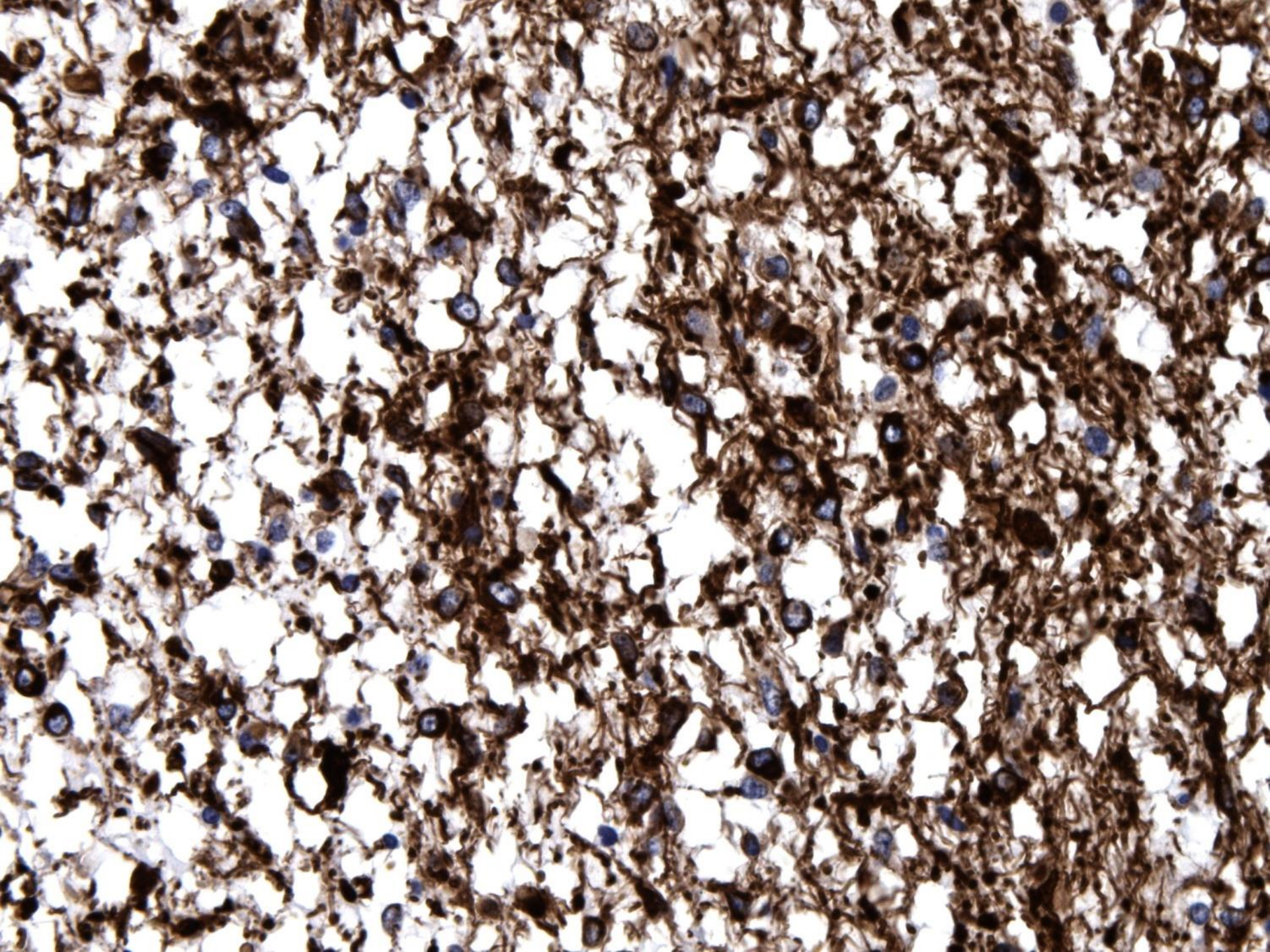




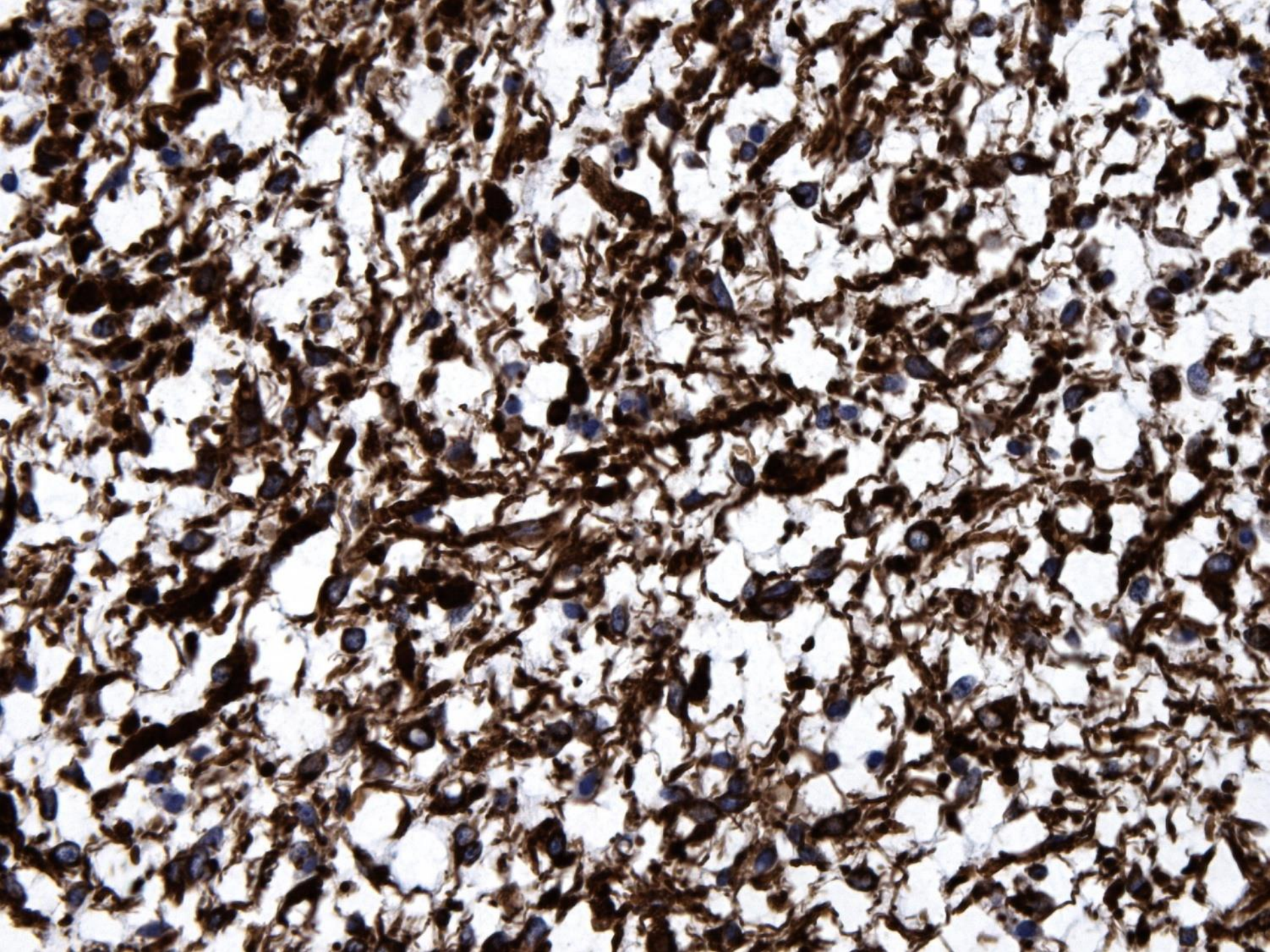


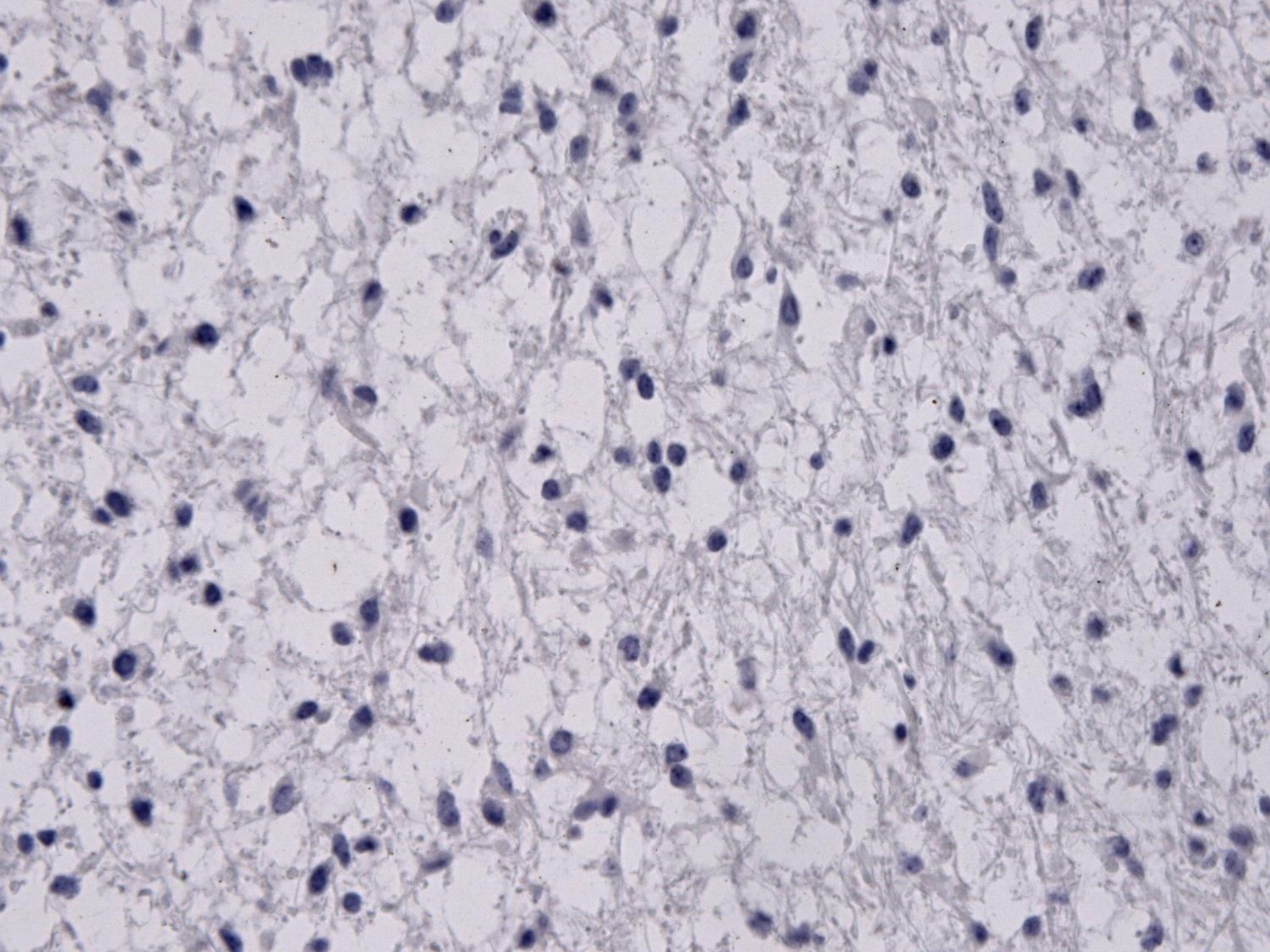


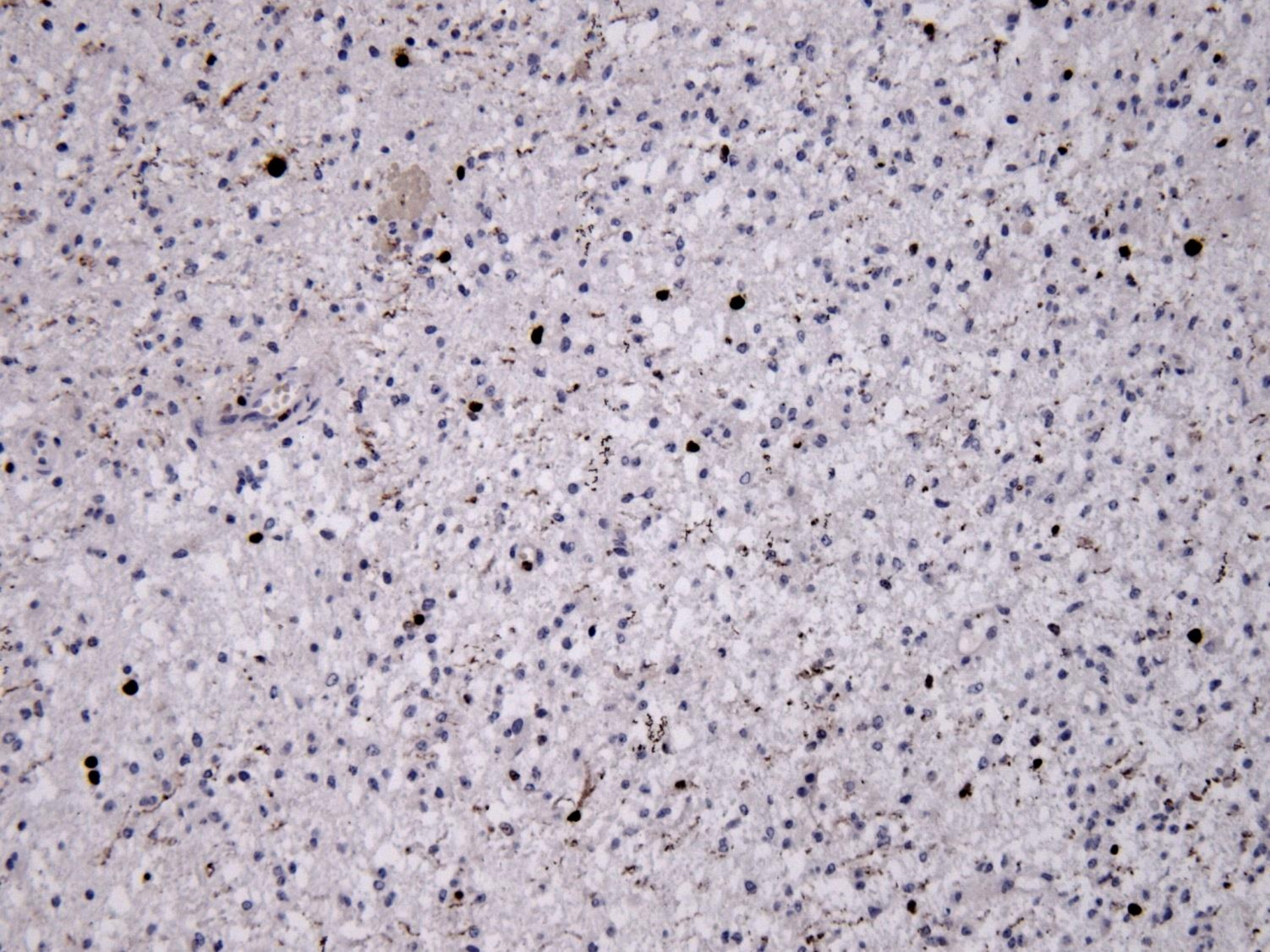












*Souhrn provedených molekulárně patologických vyšetření:*

- Byla detekována přítomnost fúzního genu *KIAA1549/BRAF* - ***byla potvrzena fúze exonu 15 genu KIAA1549 s exonem 9 genu BRAF***
- Nebyla zjištěna přítomnost aktivační mutace genu *BRAF V600E*
- Nebyla detekována žádná z hot spot mutací genu *IDH1* a *IDH2*

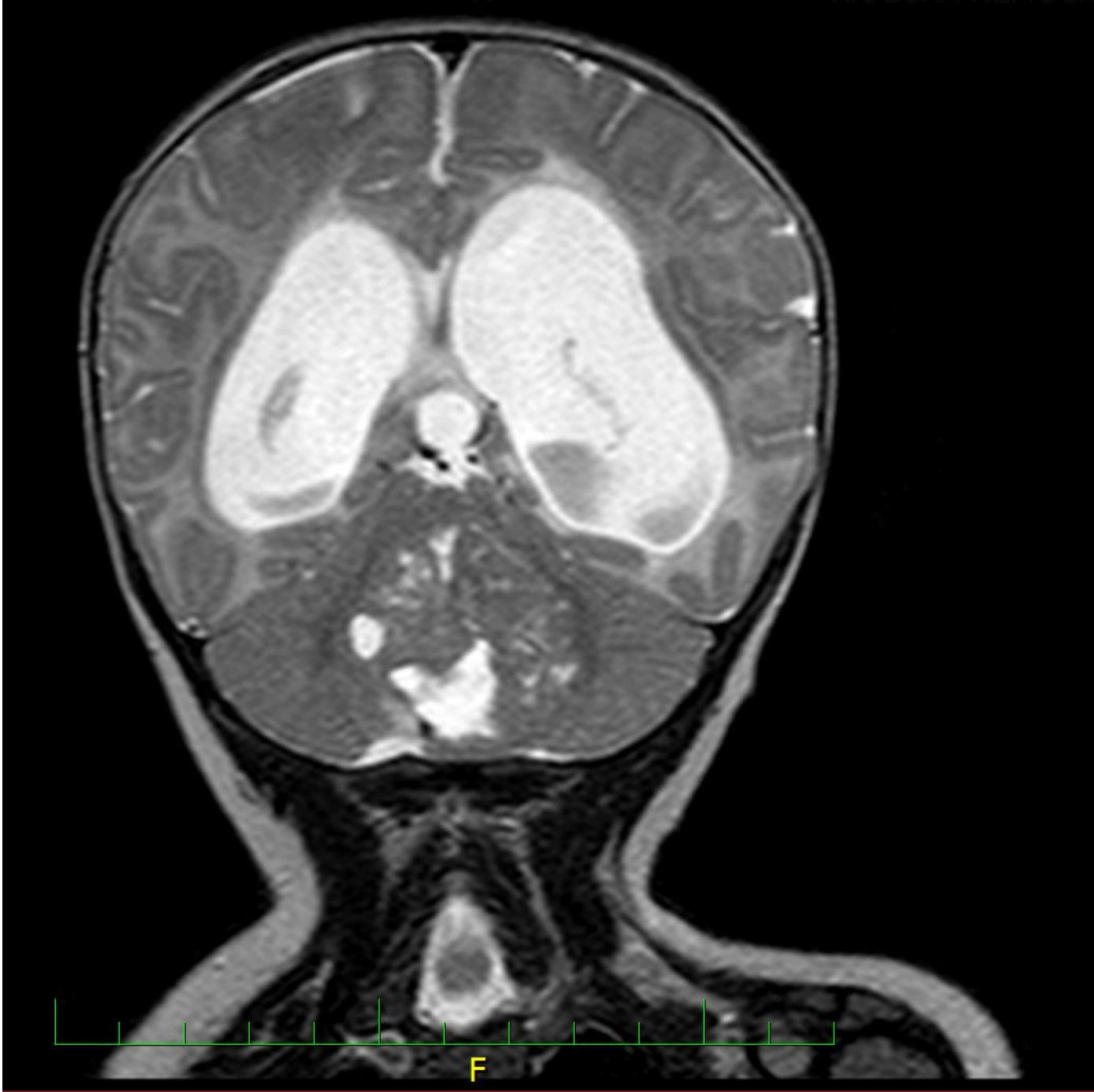
***pilocytární astrocytom, WHO grade I,***

***s přítomností fúzního genu KIAA1549/BRAF***



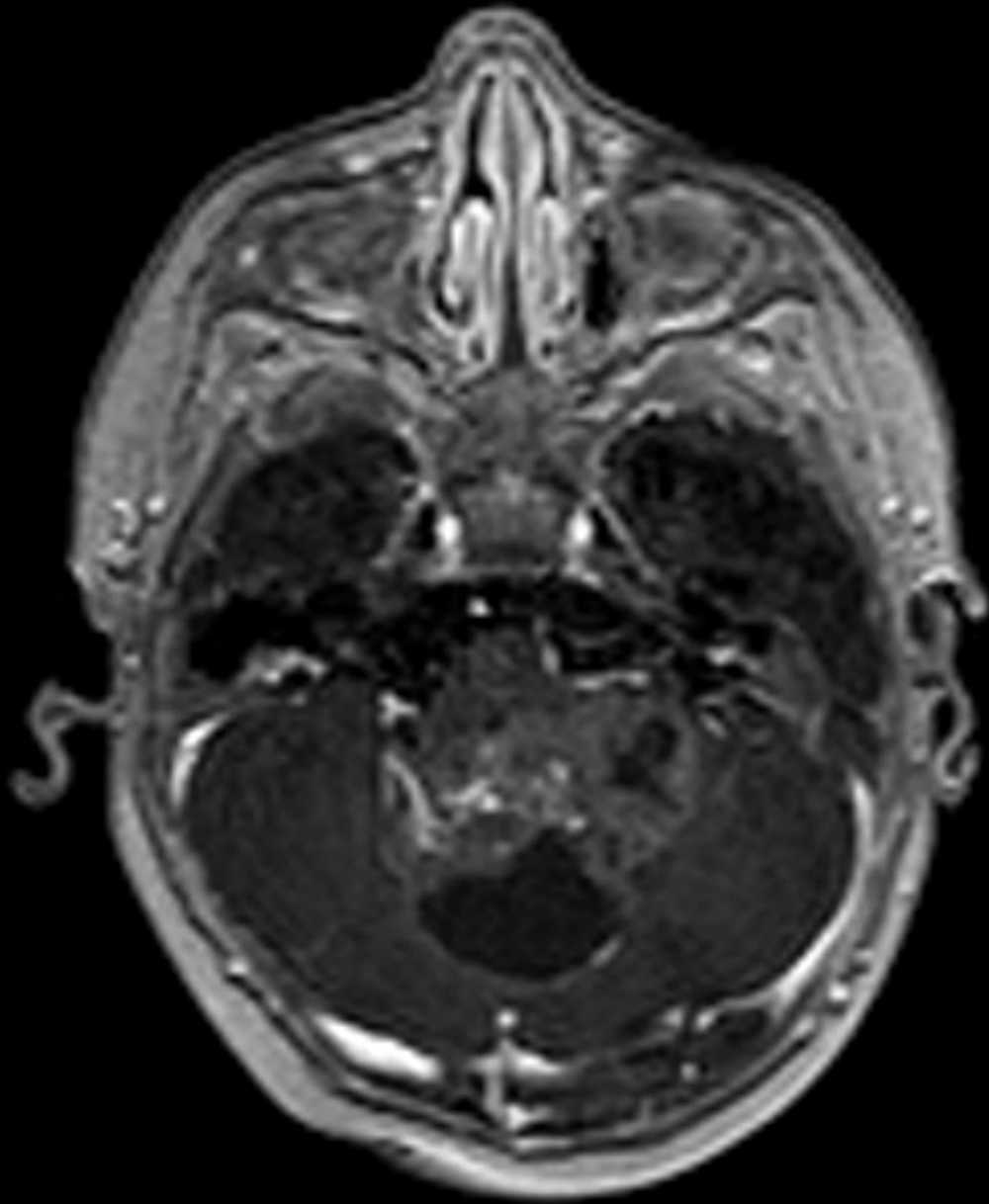
**3M dívka**

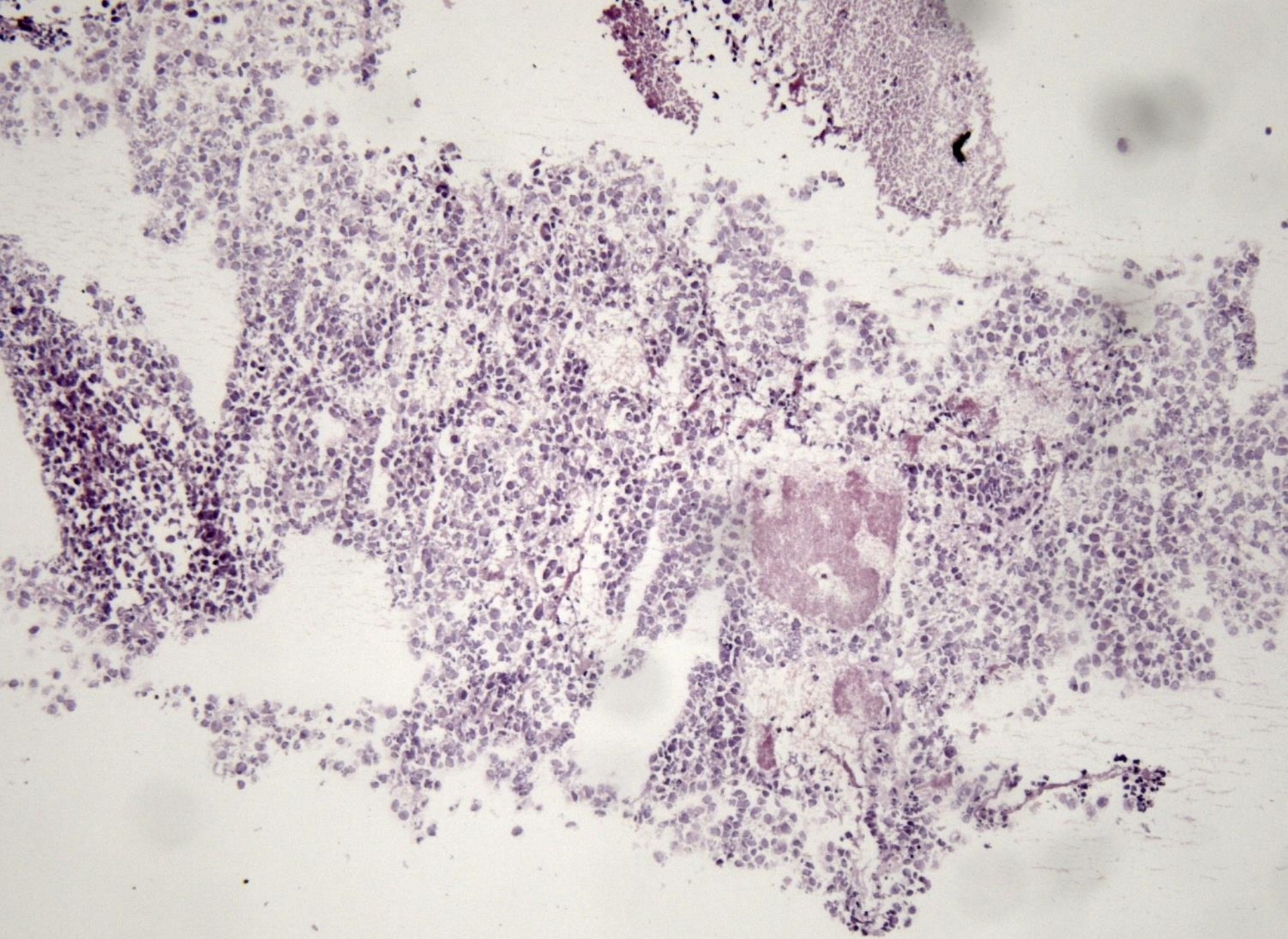
Tu expanze zadní jamy

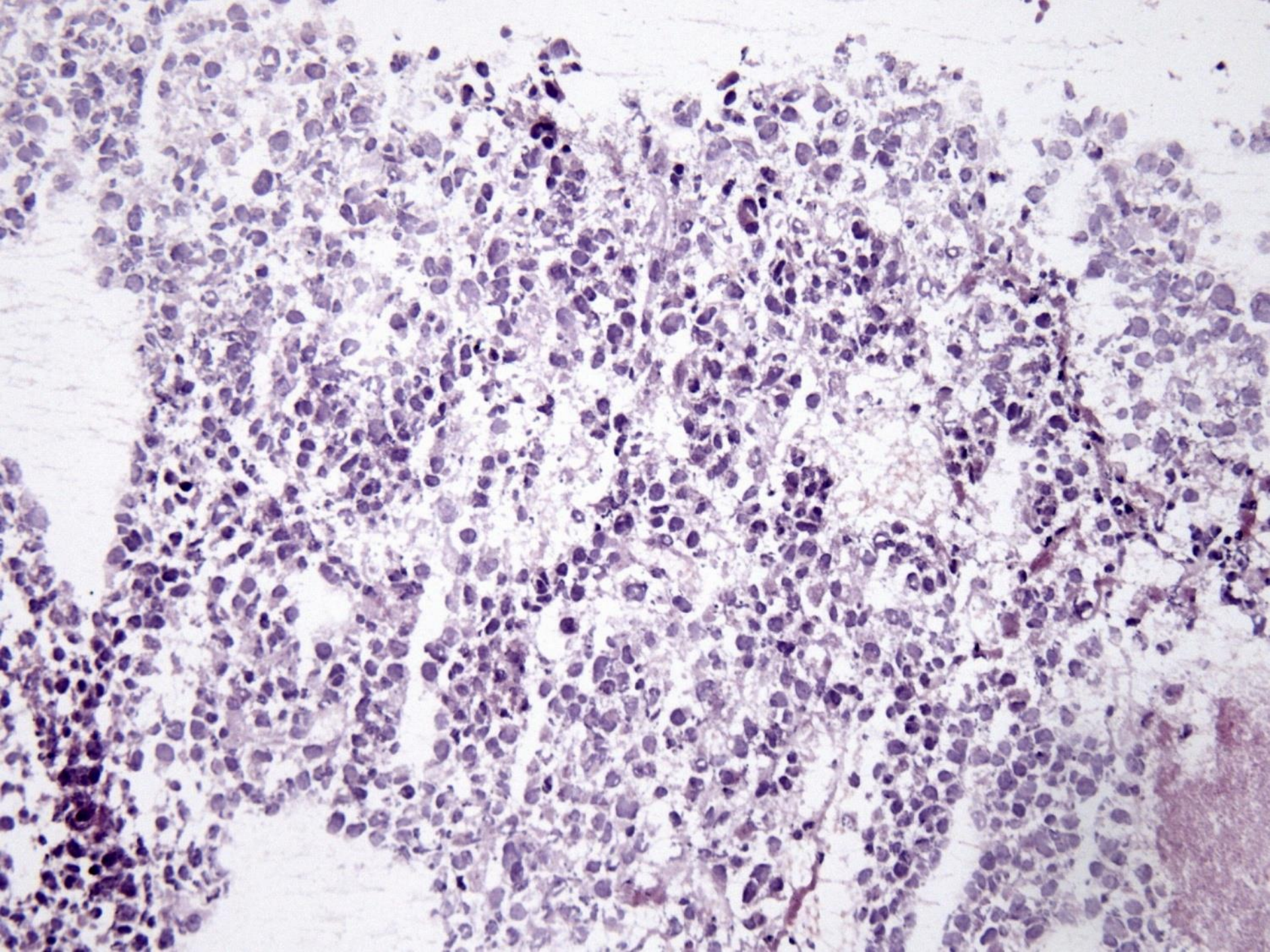


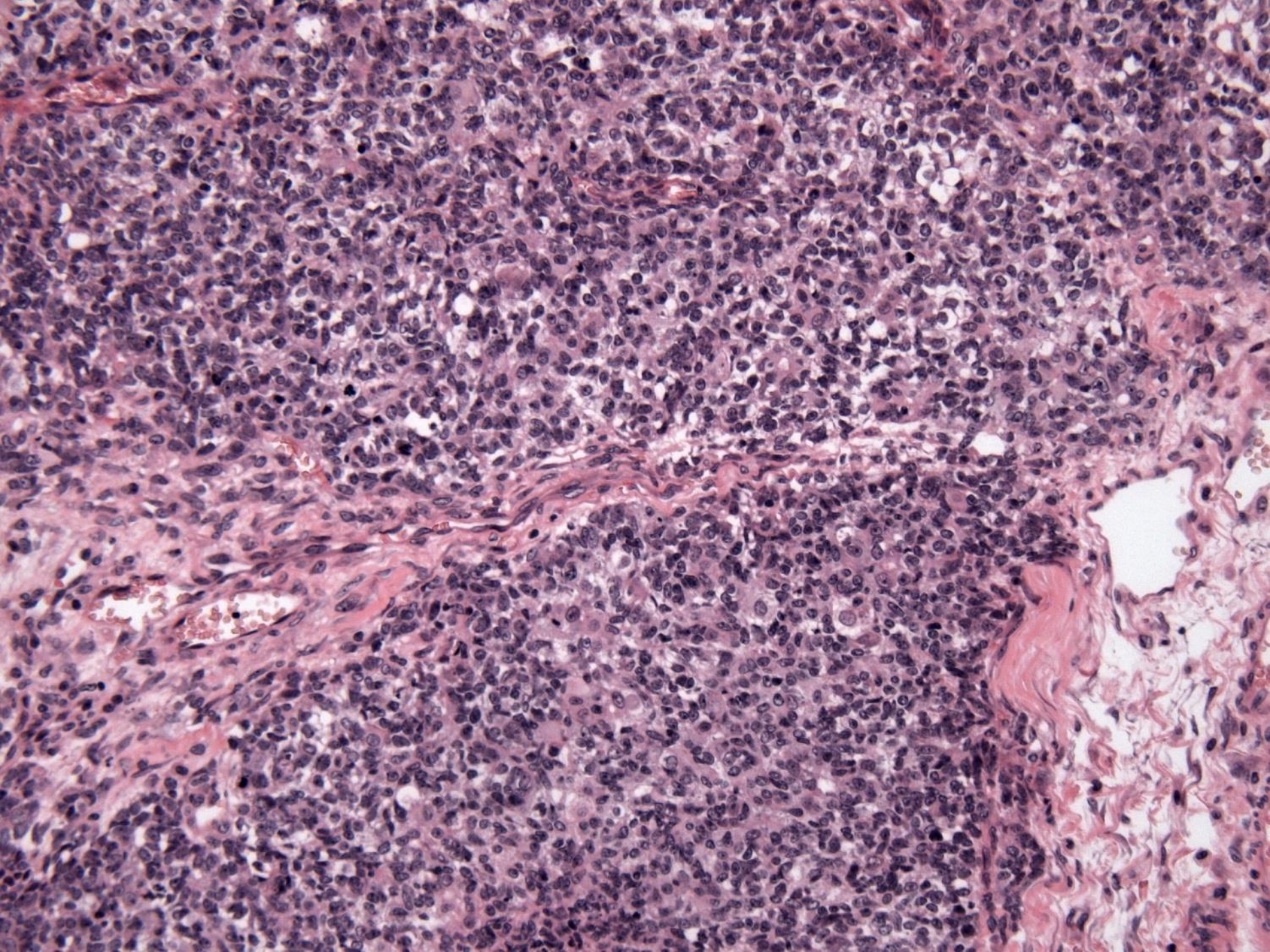


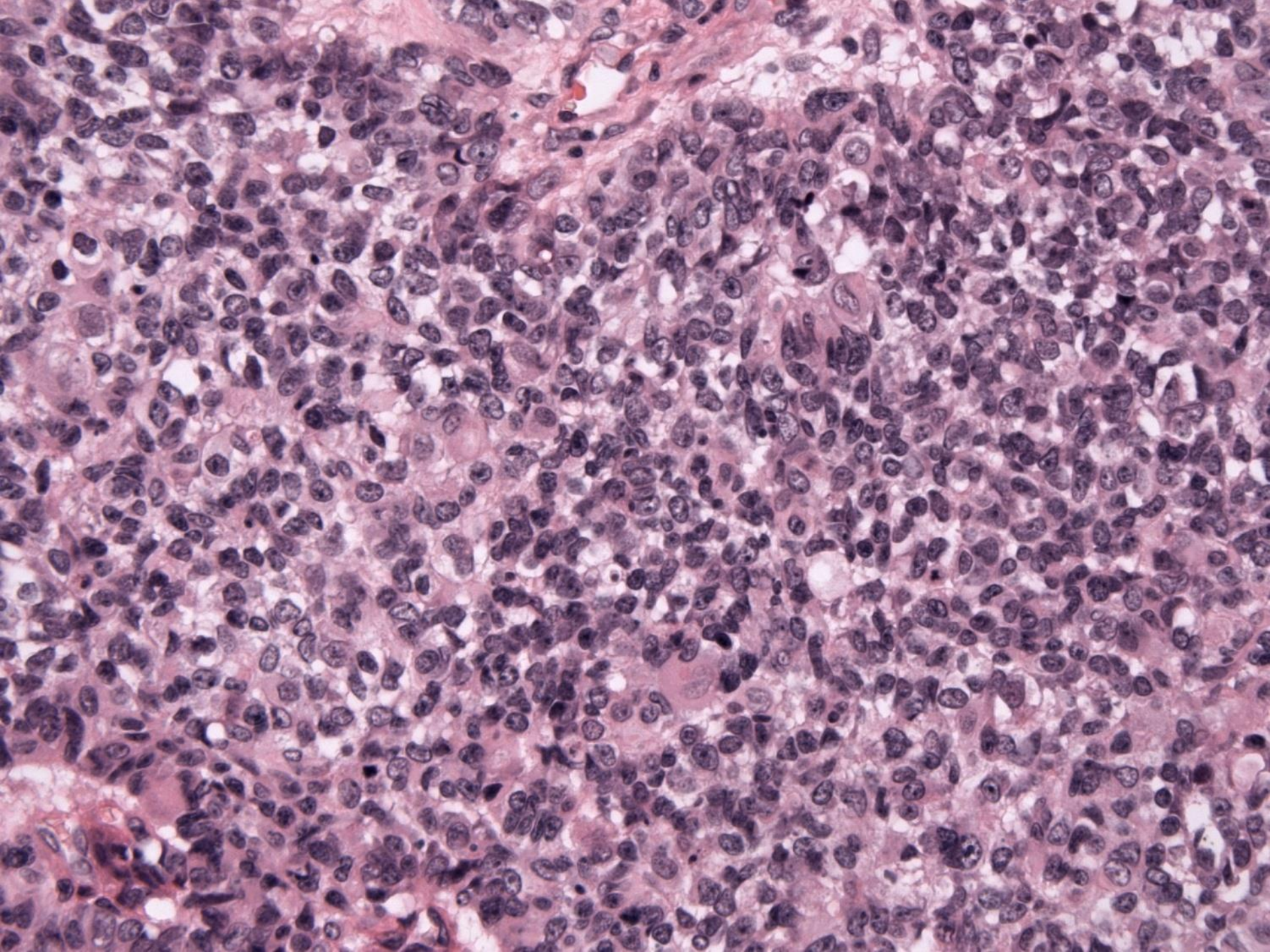


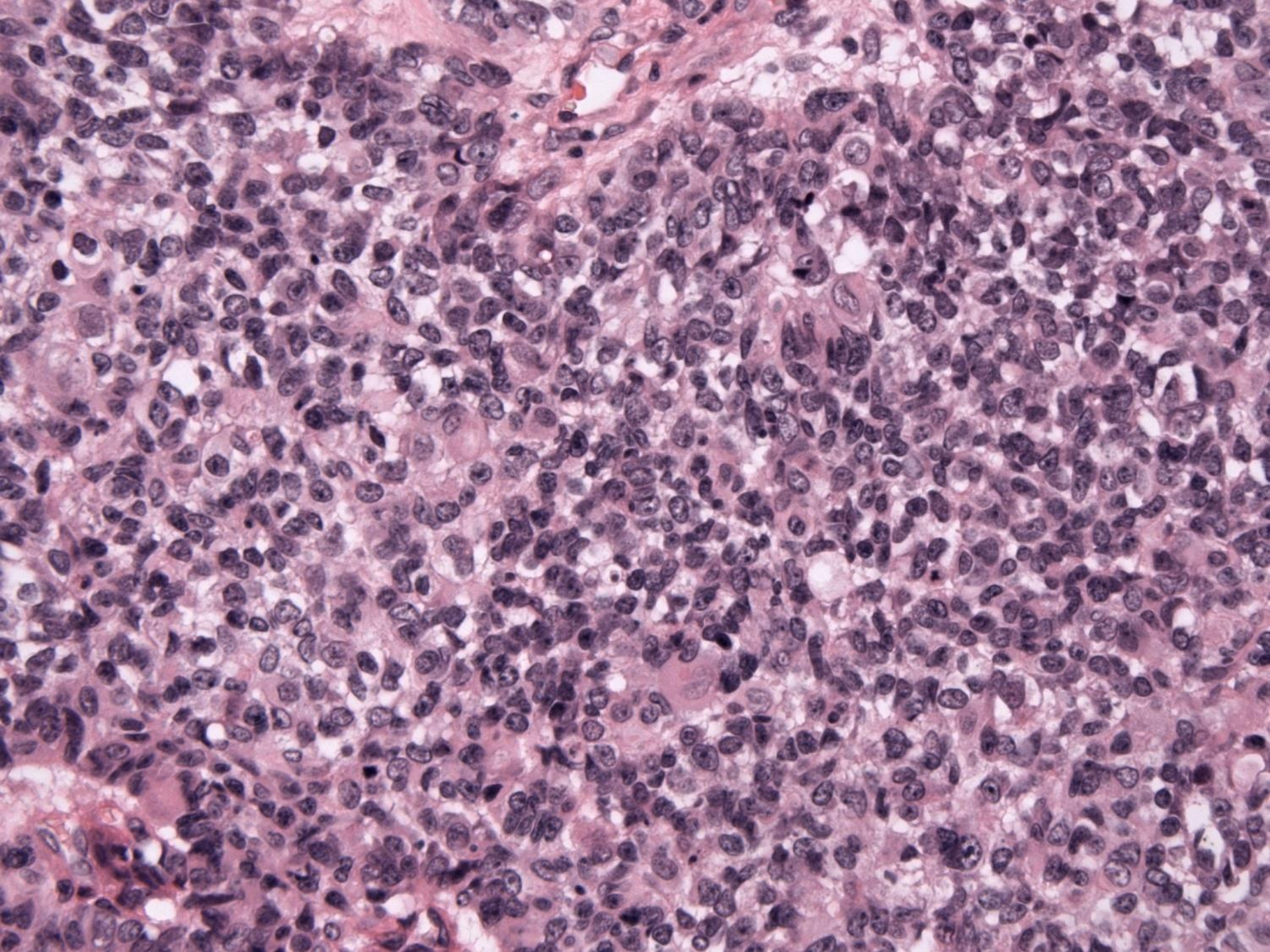


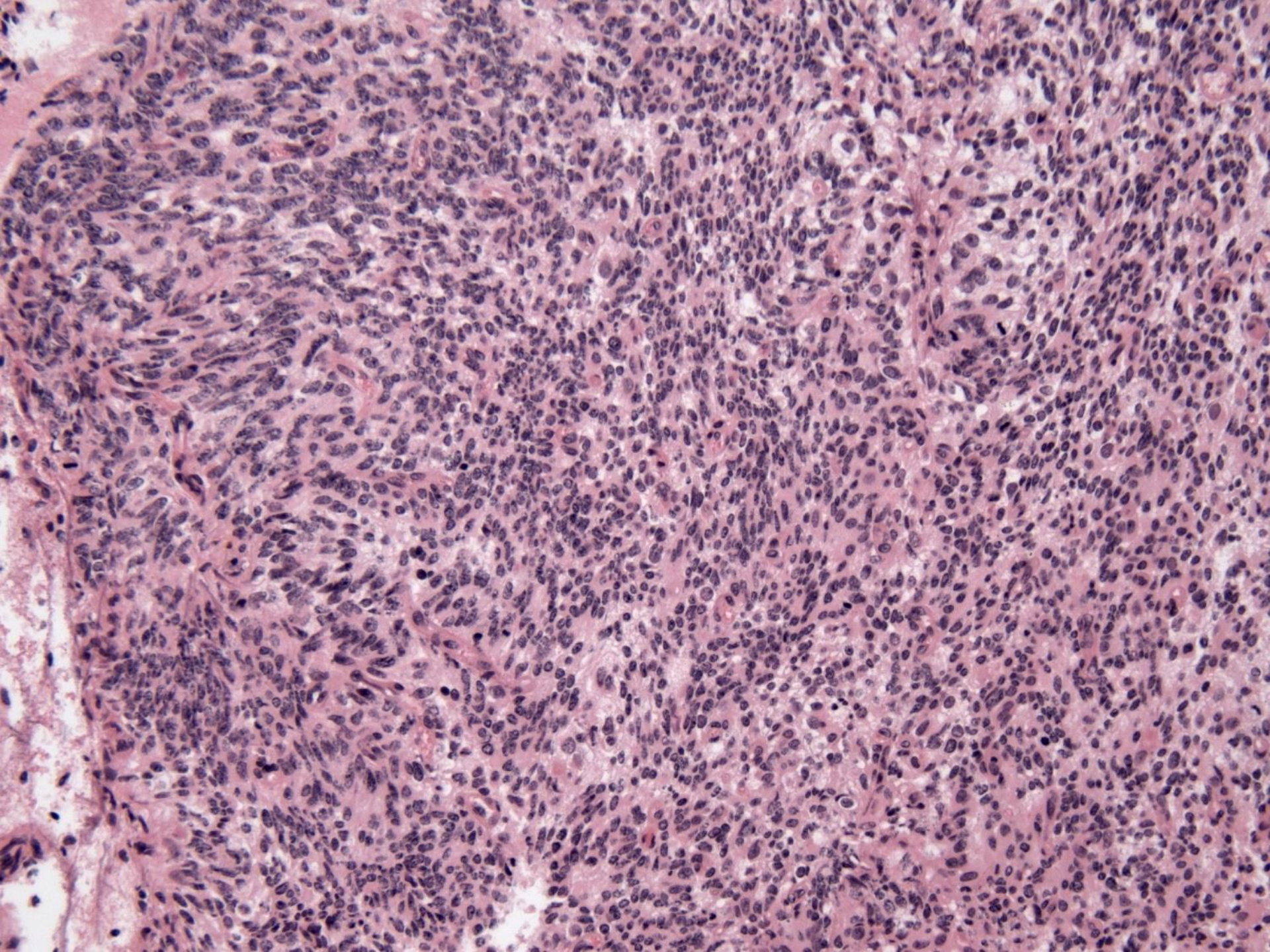


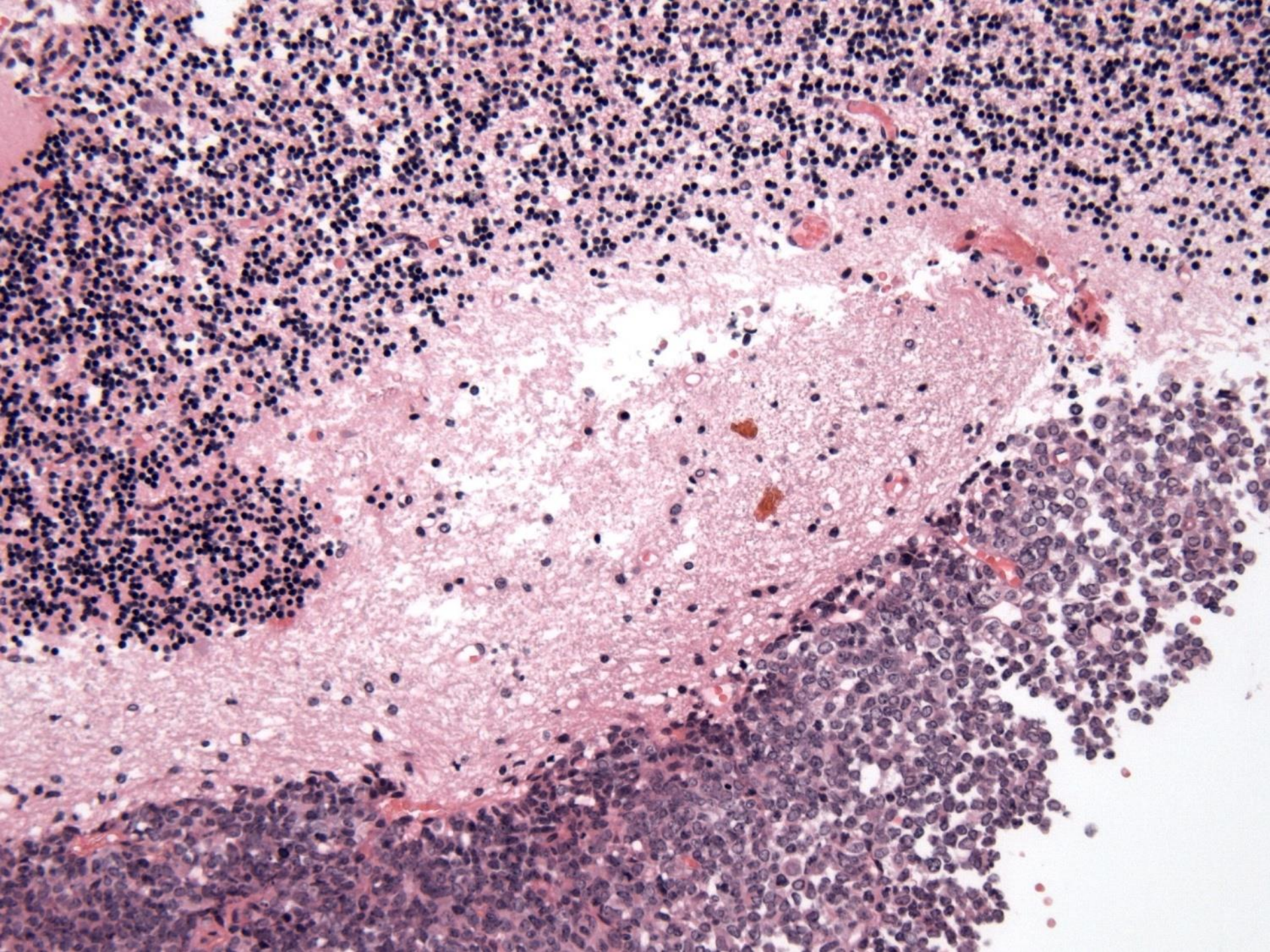




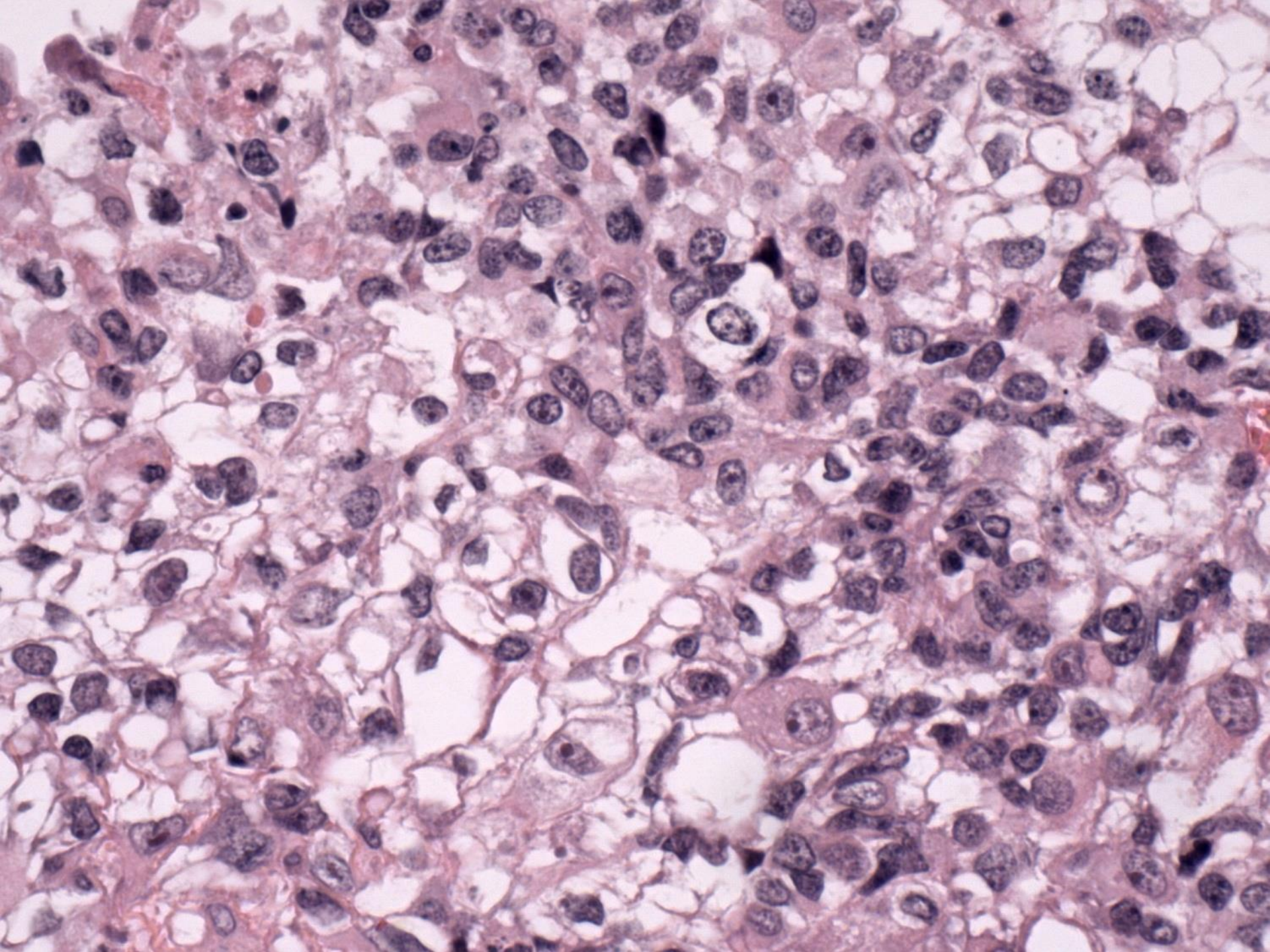


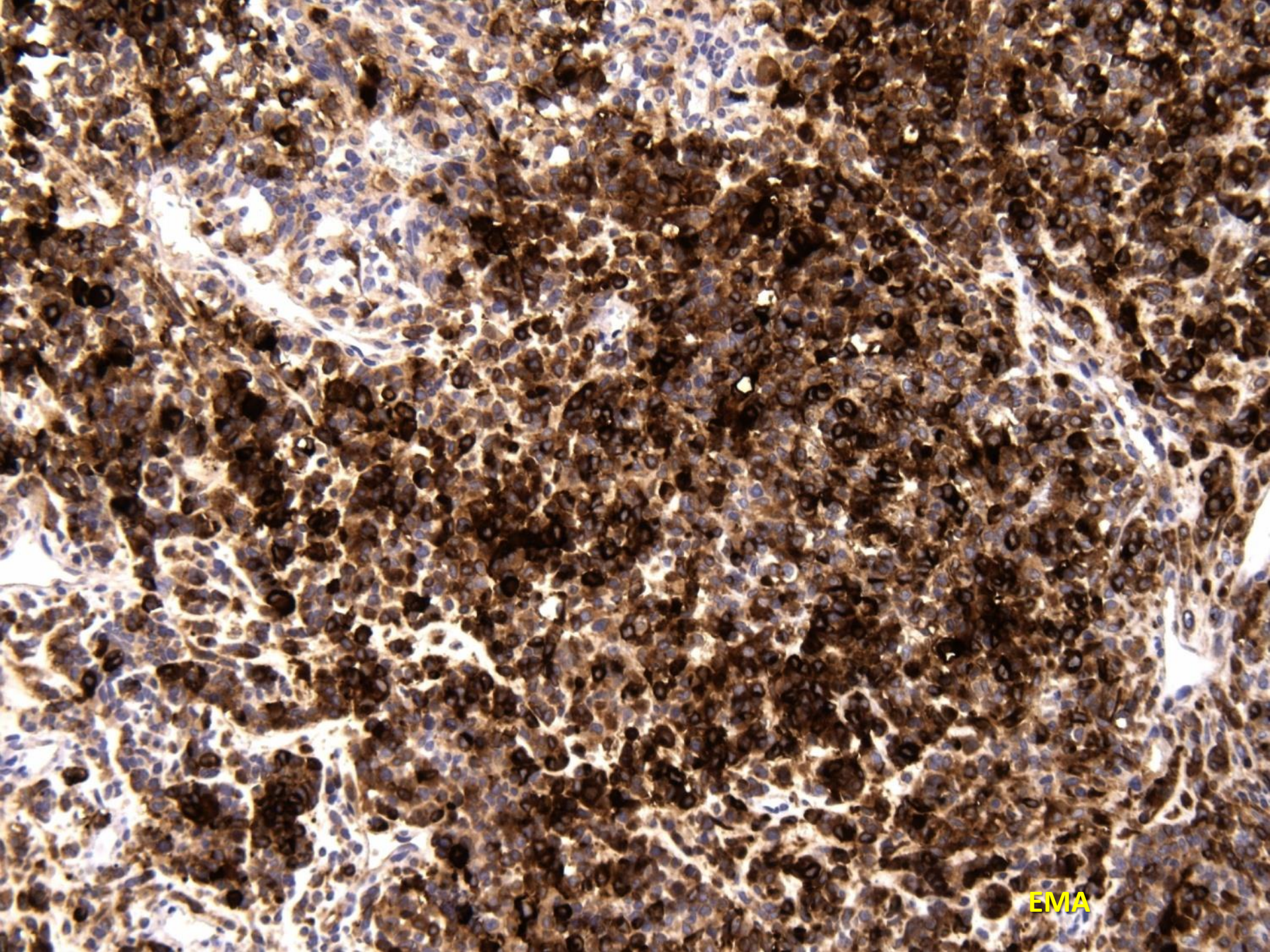




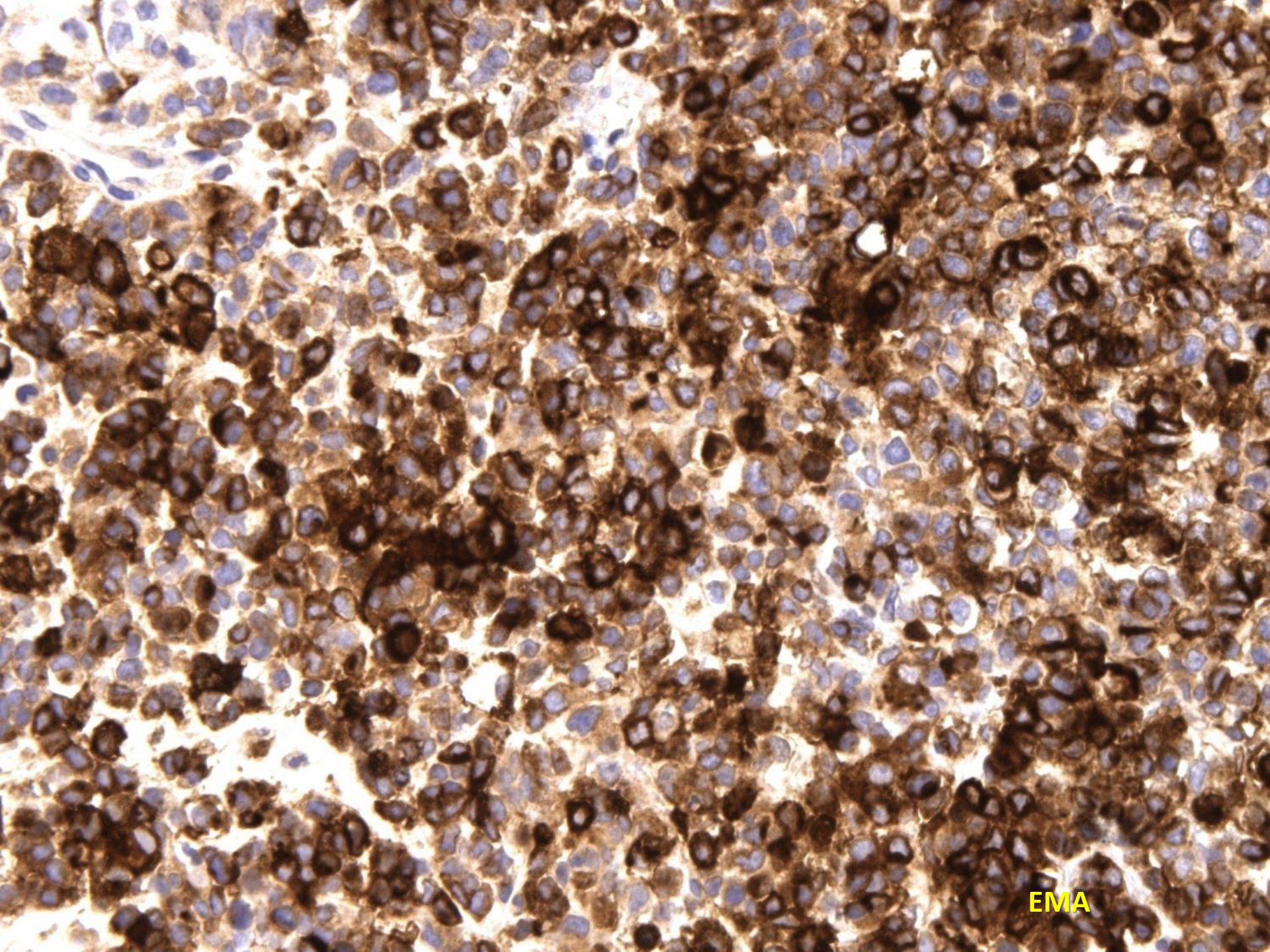




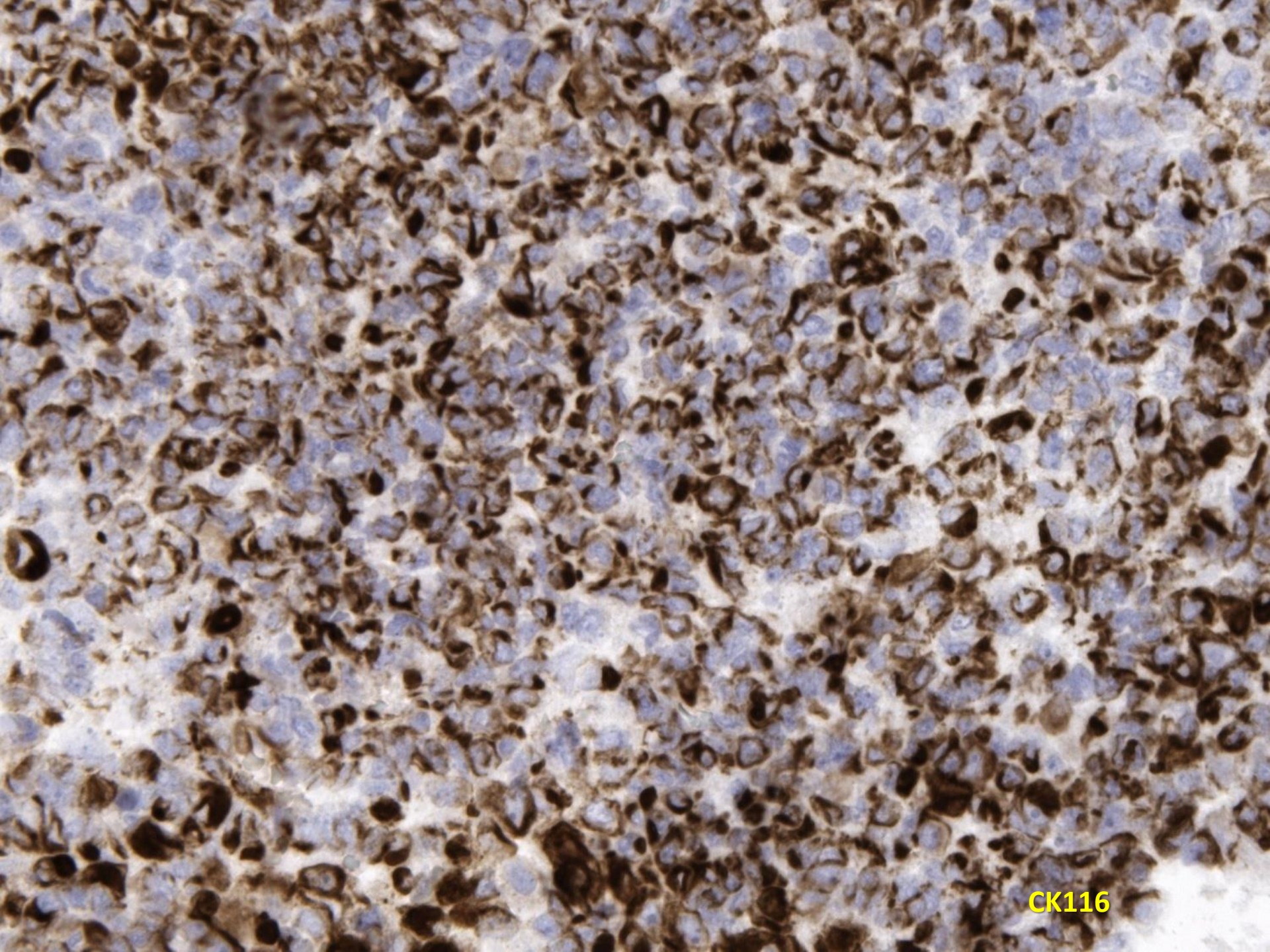




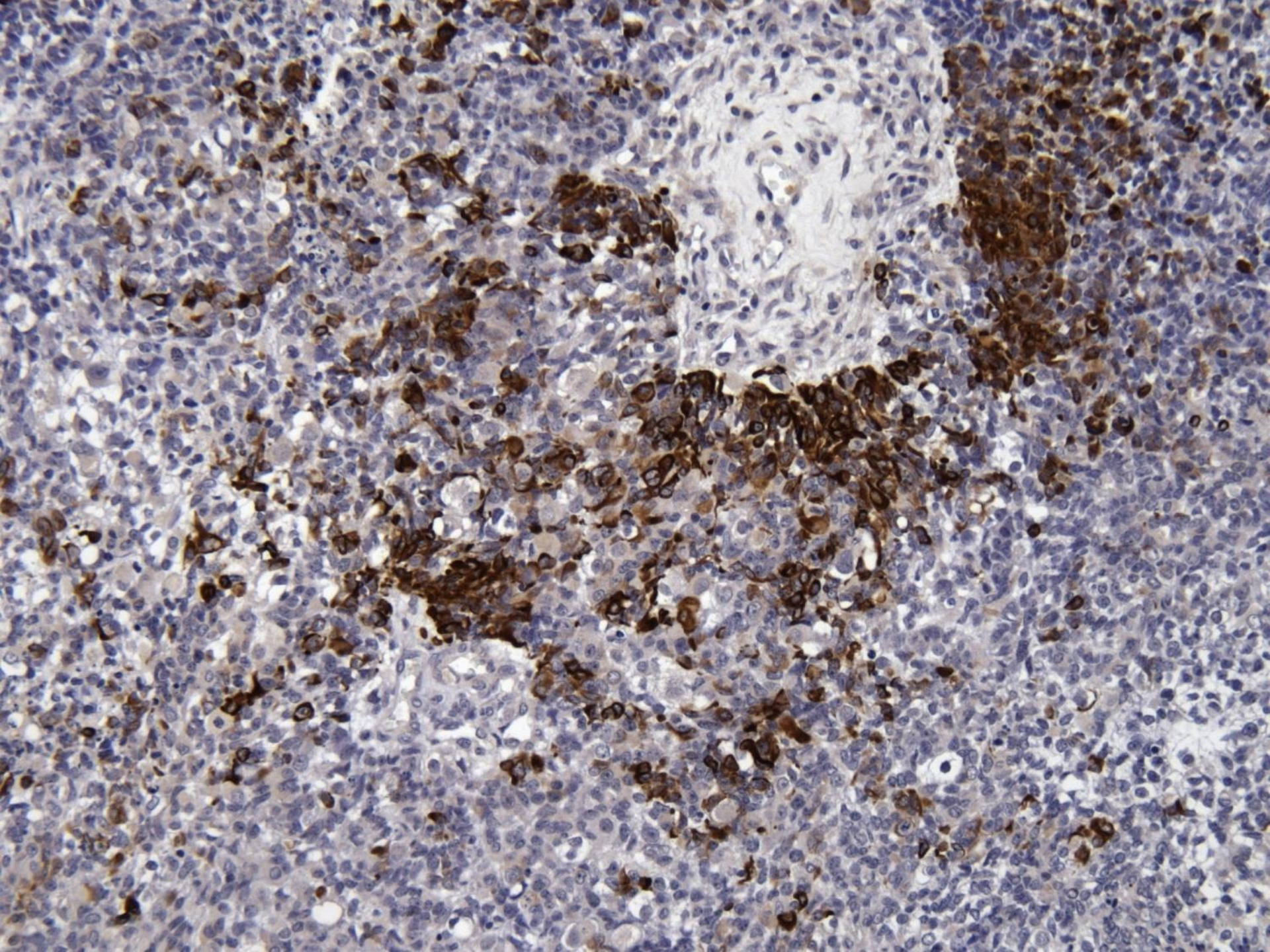
EMA

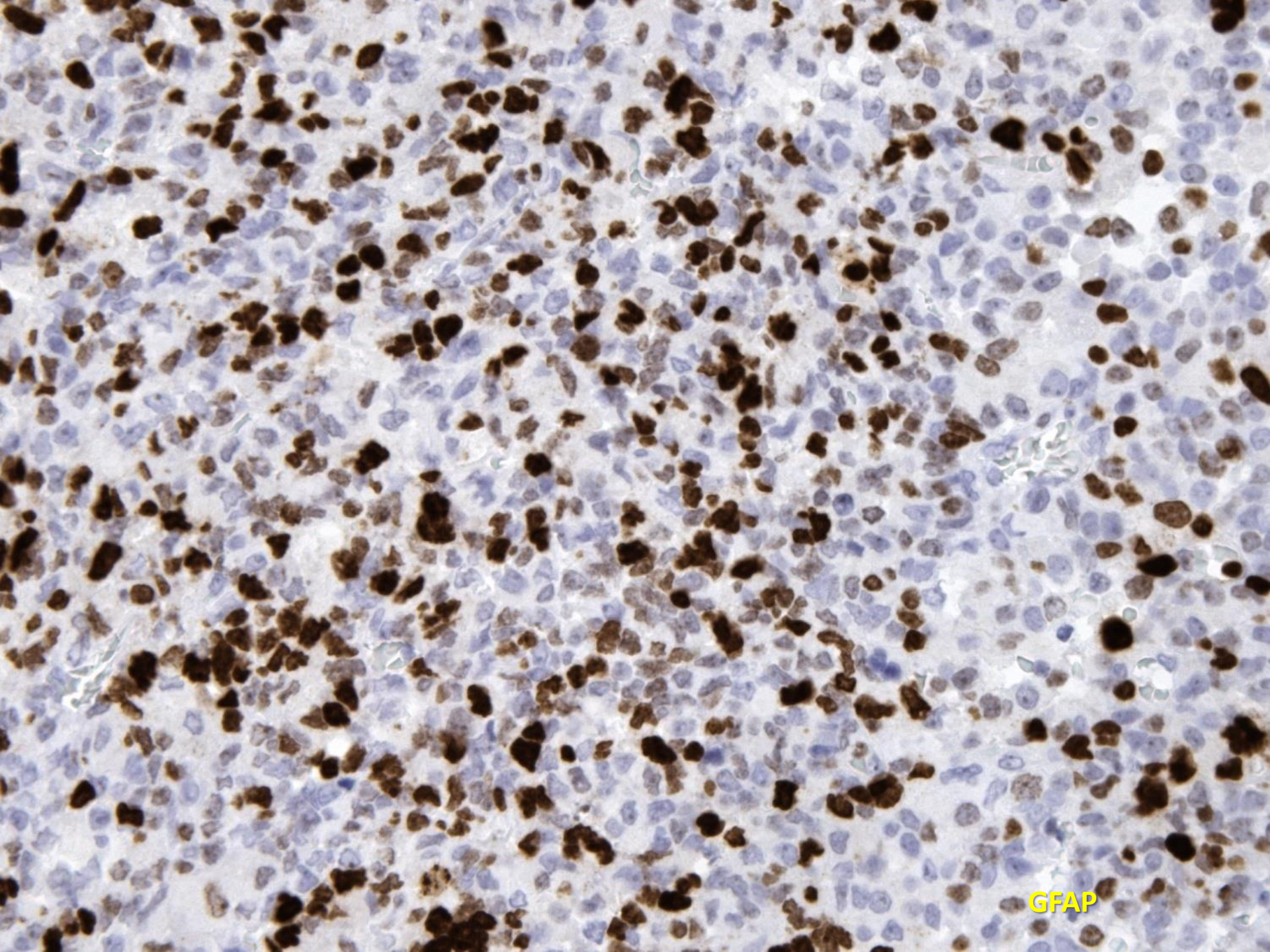


EMA

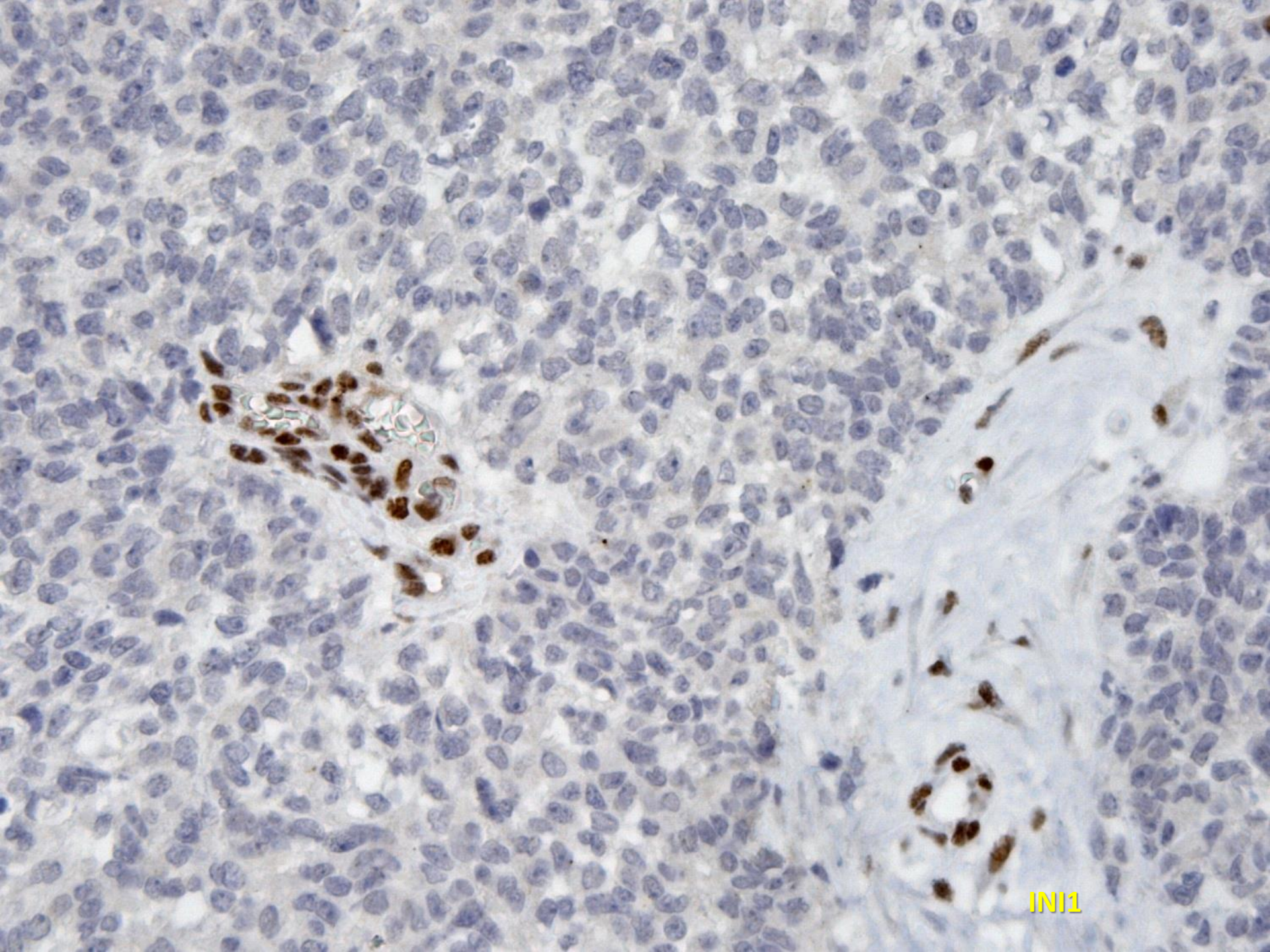


CK116





GFAP



INI1

gen *SMARCB1* (*INI1*):

Mutační analýza: přítomna inserce v intronu 7-8, mutace v exonu 9 p?/c1158+116 insG

***atypický teratoidní/rhabdoidní tumor (AT/RT),  
WHO grade IV.***





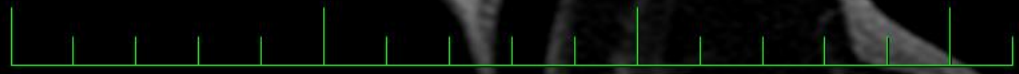
**7 let, chlapec**

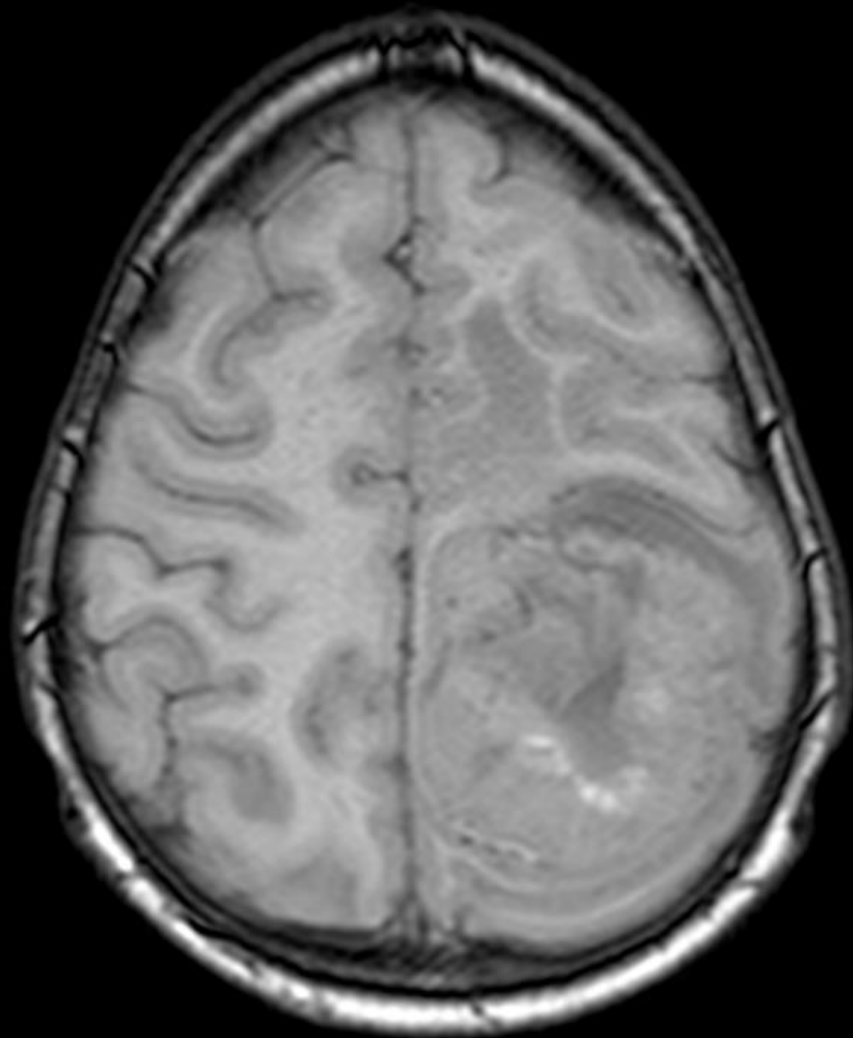
MR

Vlevo parietalne v bile i sede hmote je okrouhle nehomogenni lozisko s centralnim rozpadem rozmeru asi 63 x 53 x 45 mm.

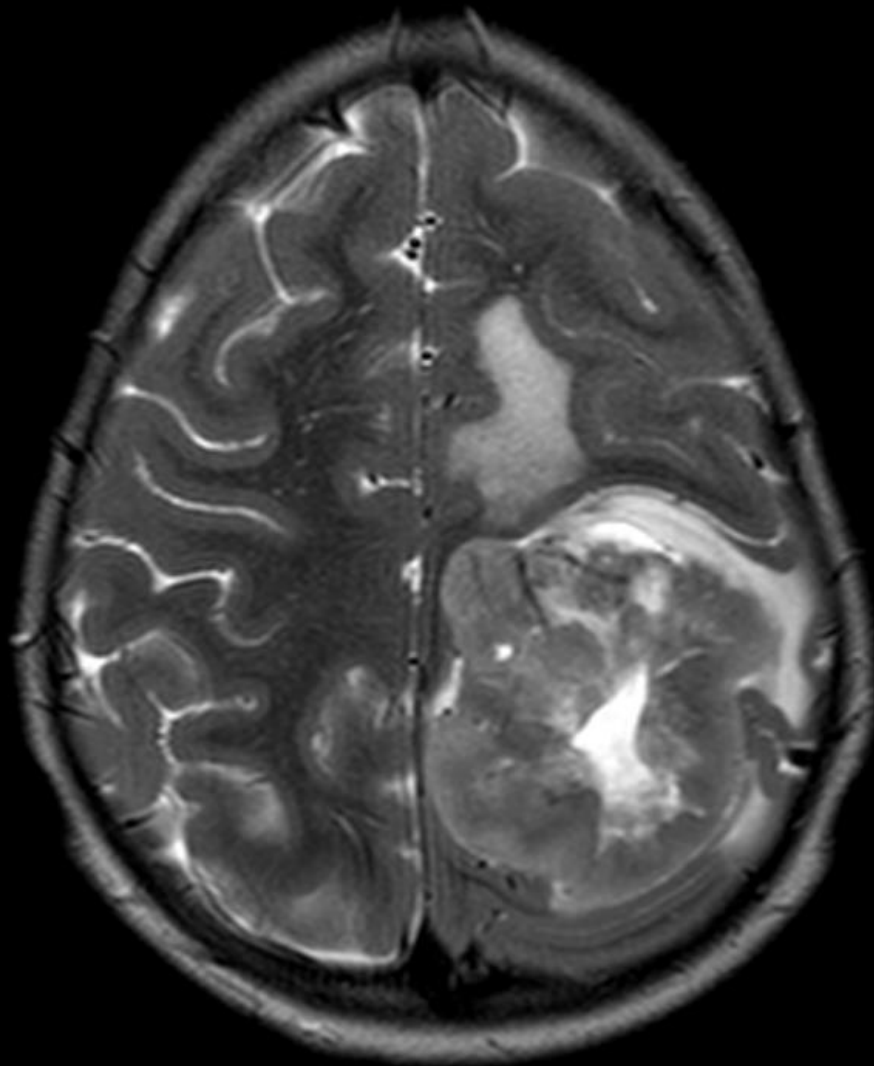
V okoli loziska je pomerne rozsahly perifokalni edem.

Rostralne dosahuje az ke kalve.

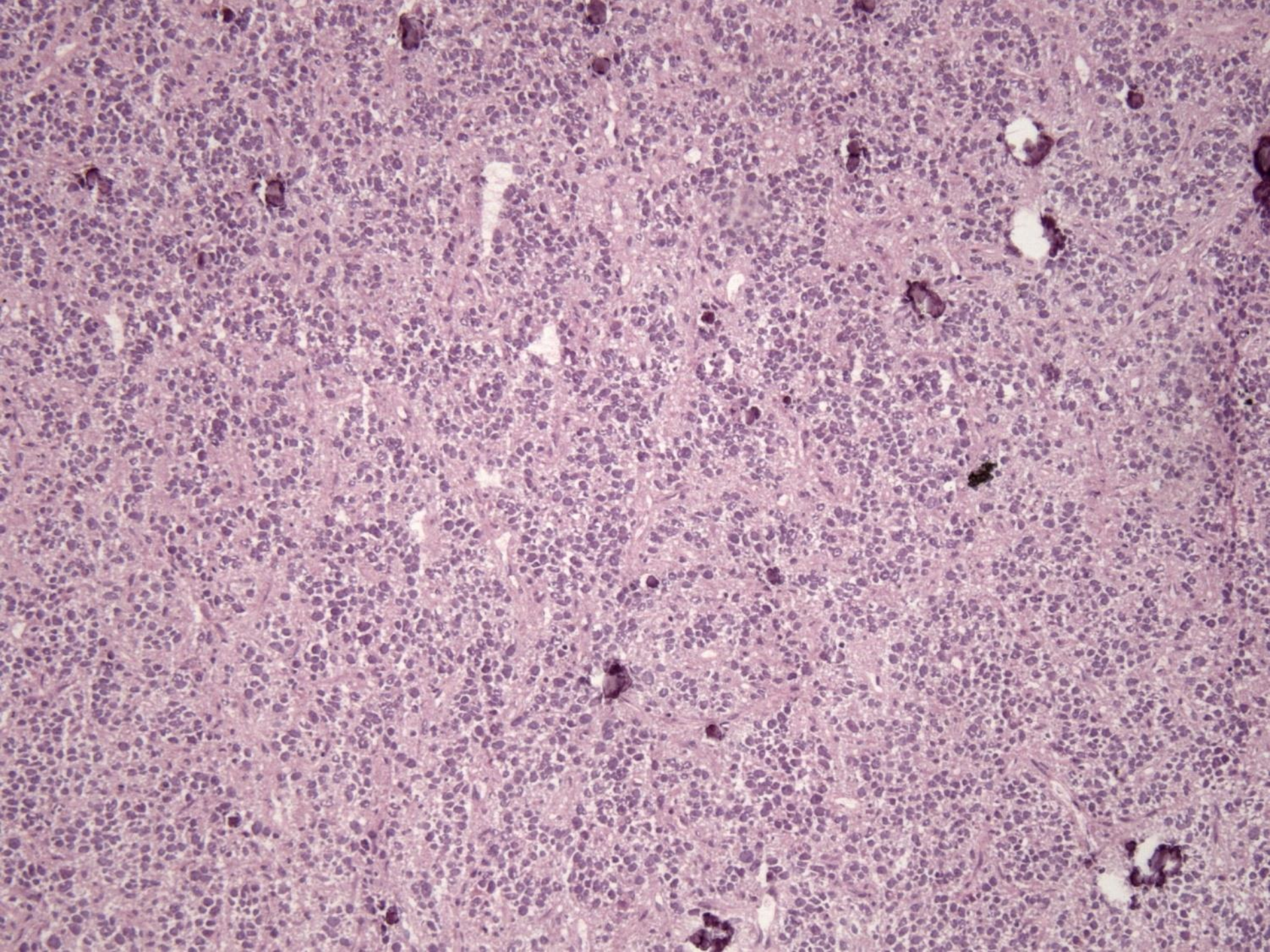


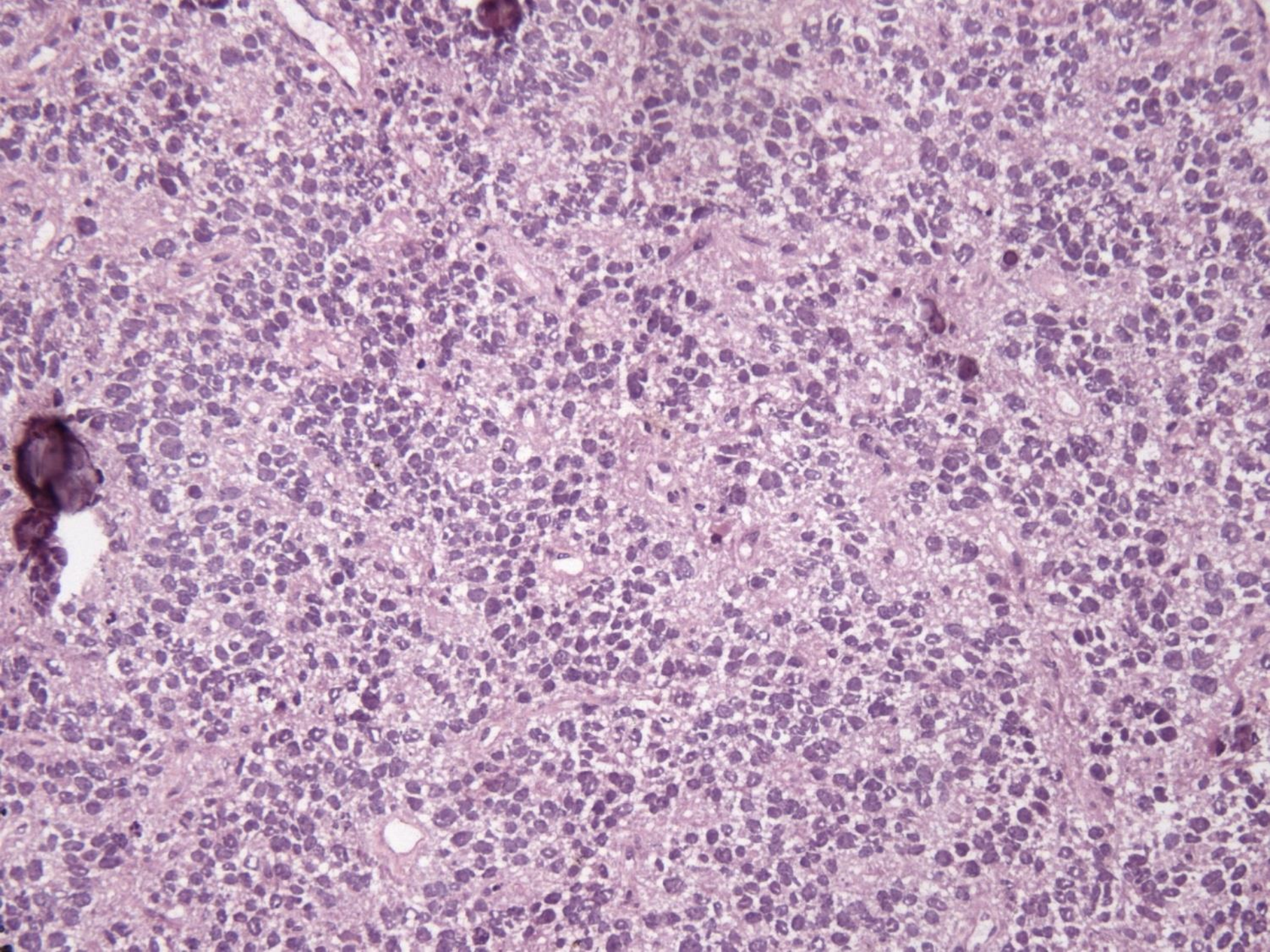


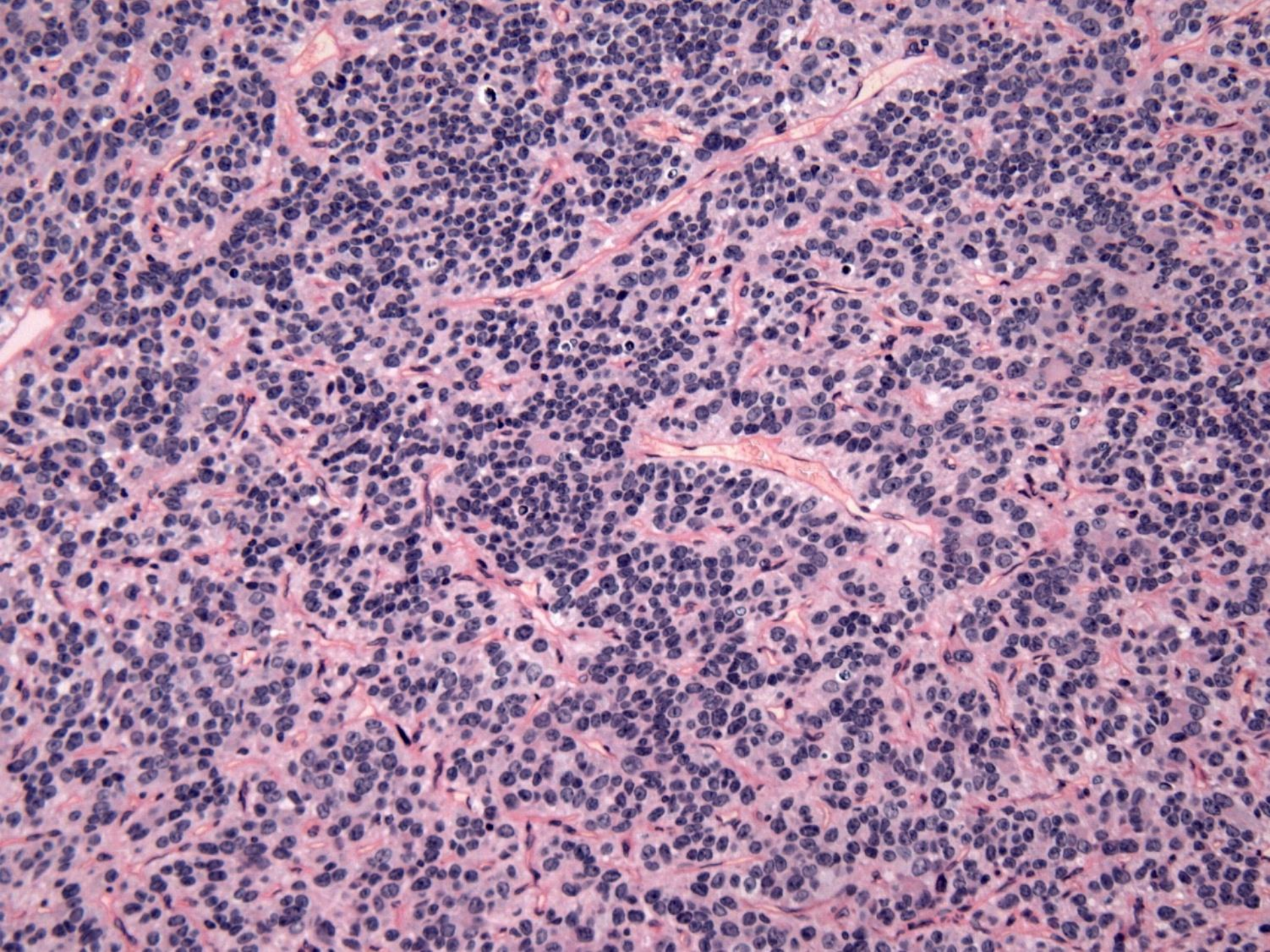
P



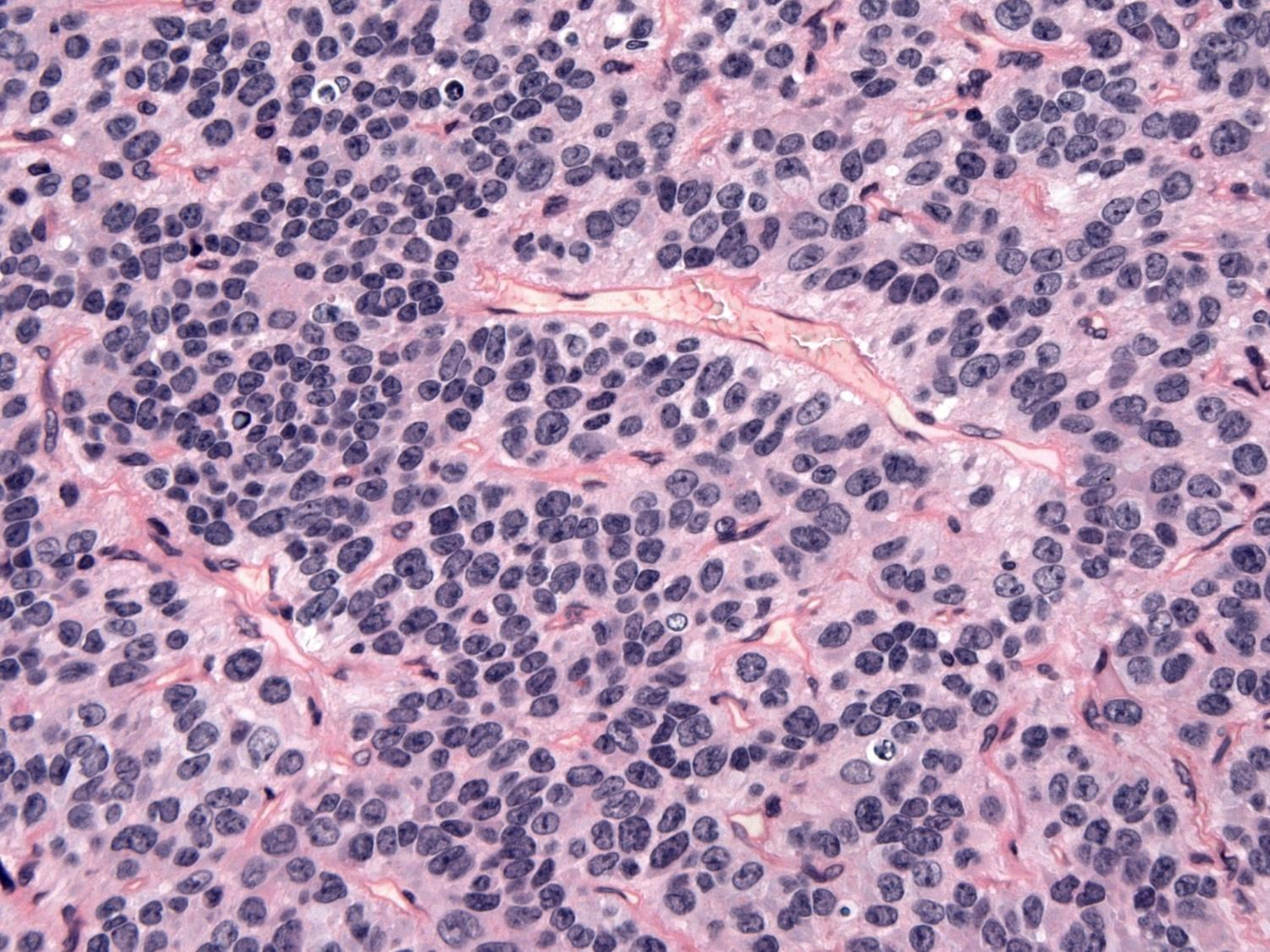
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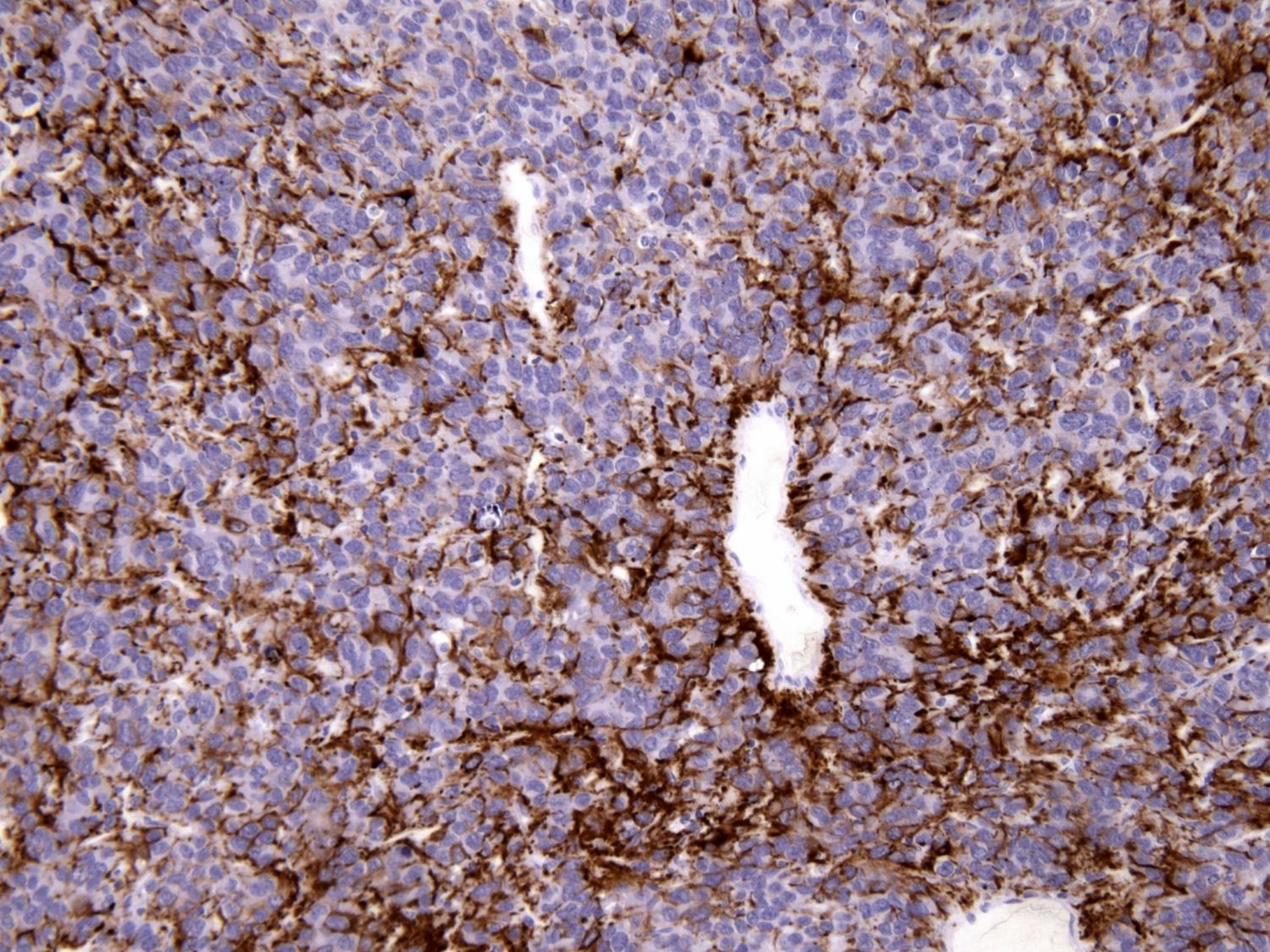


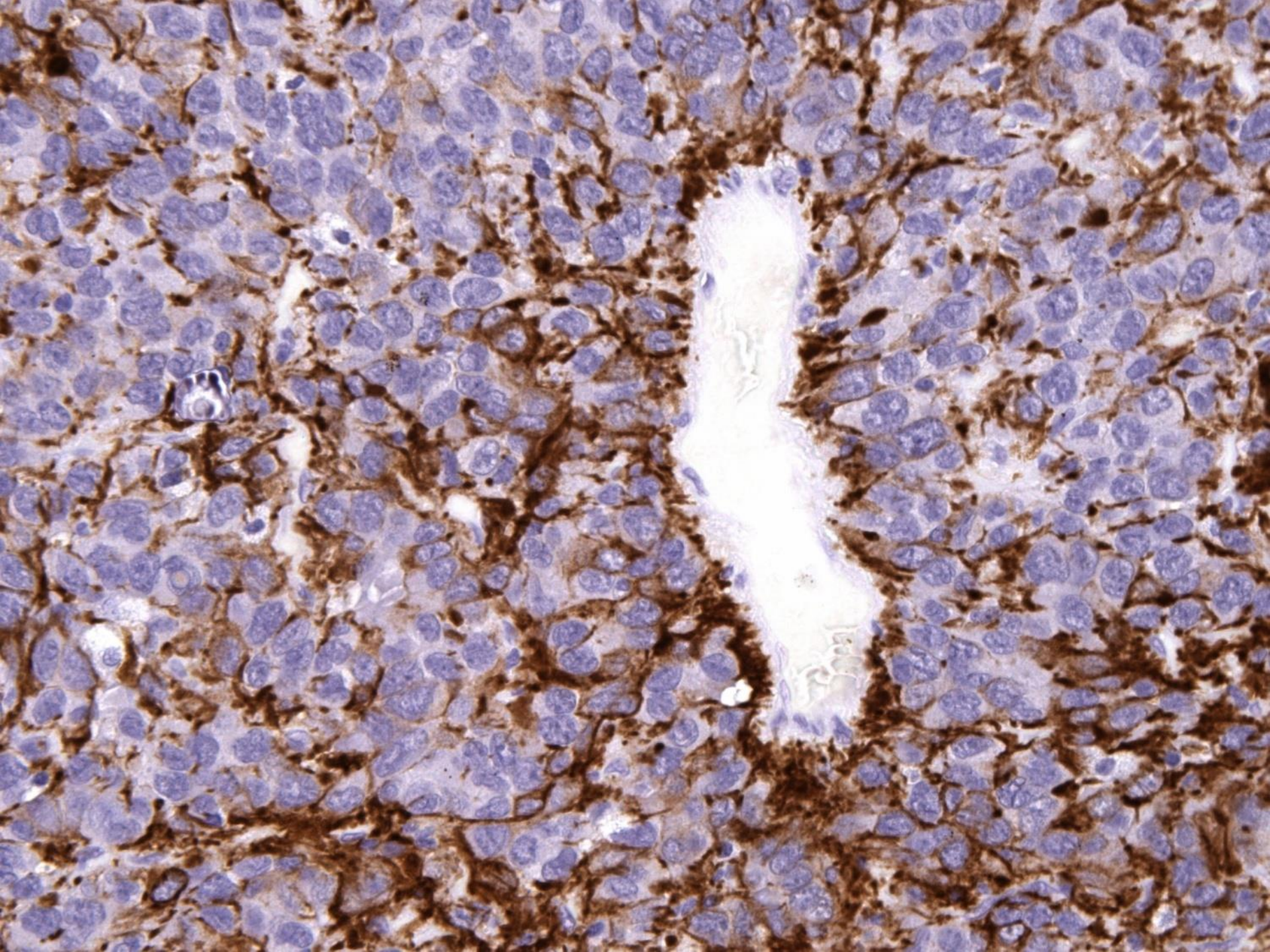


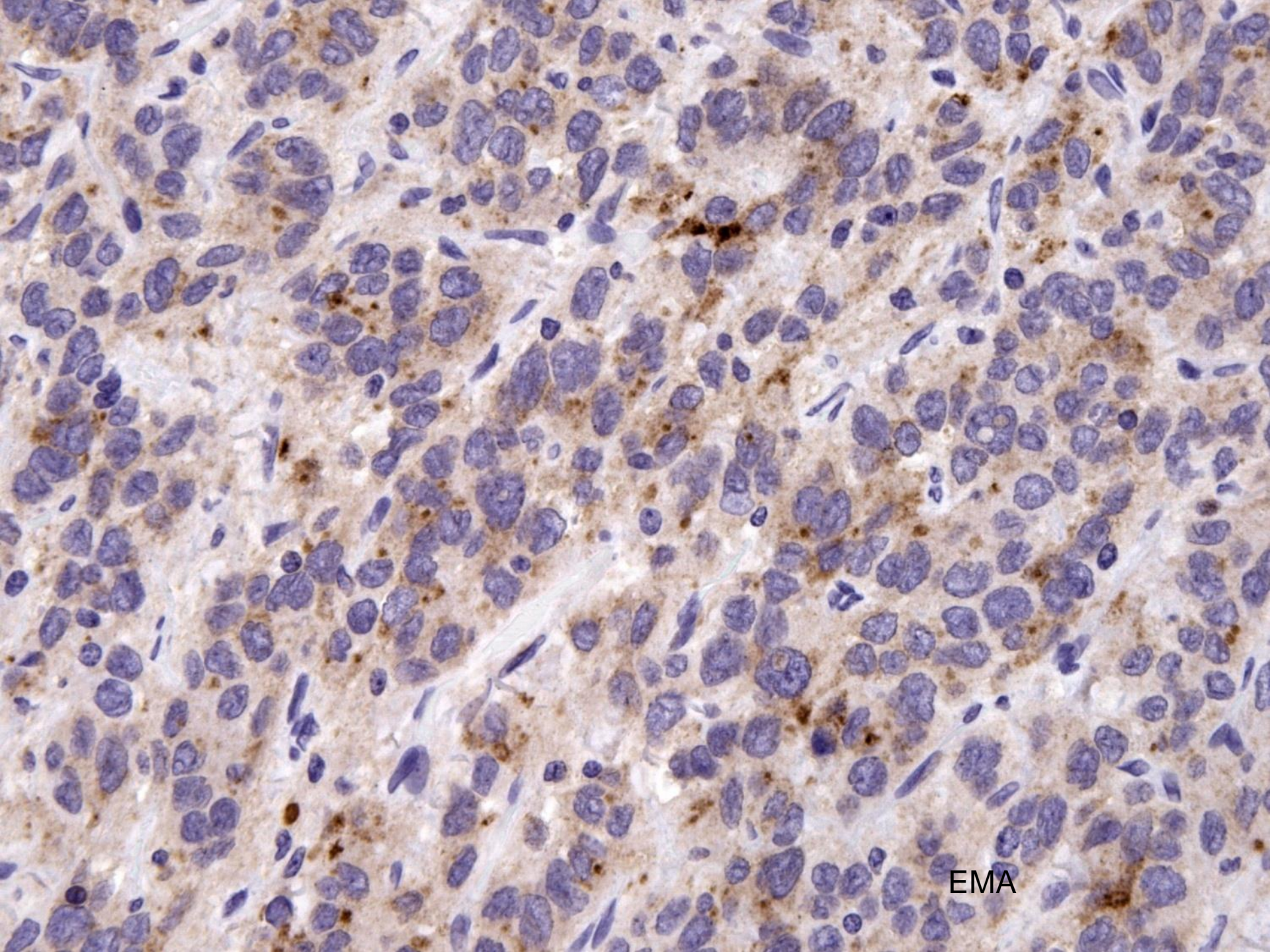




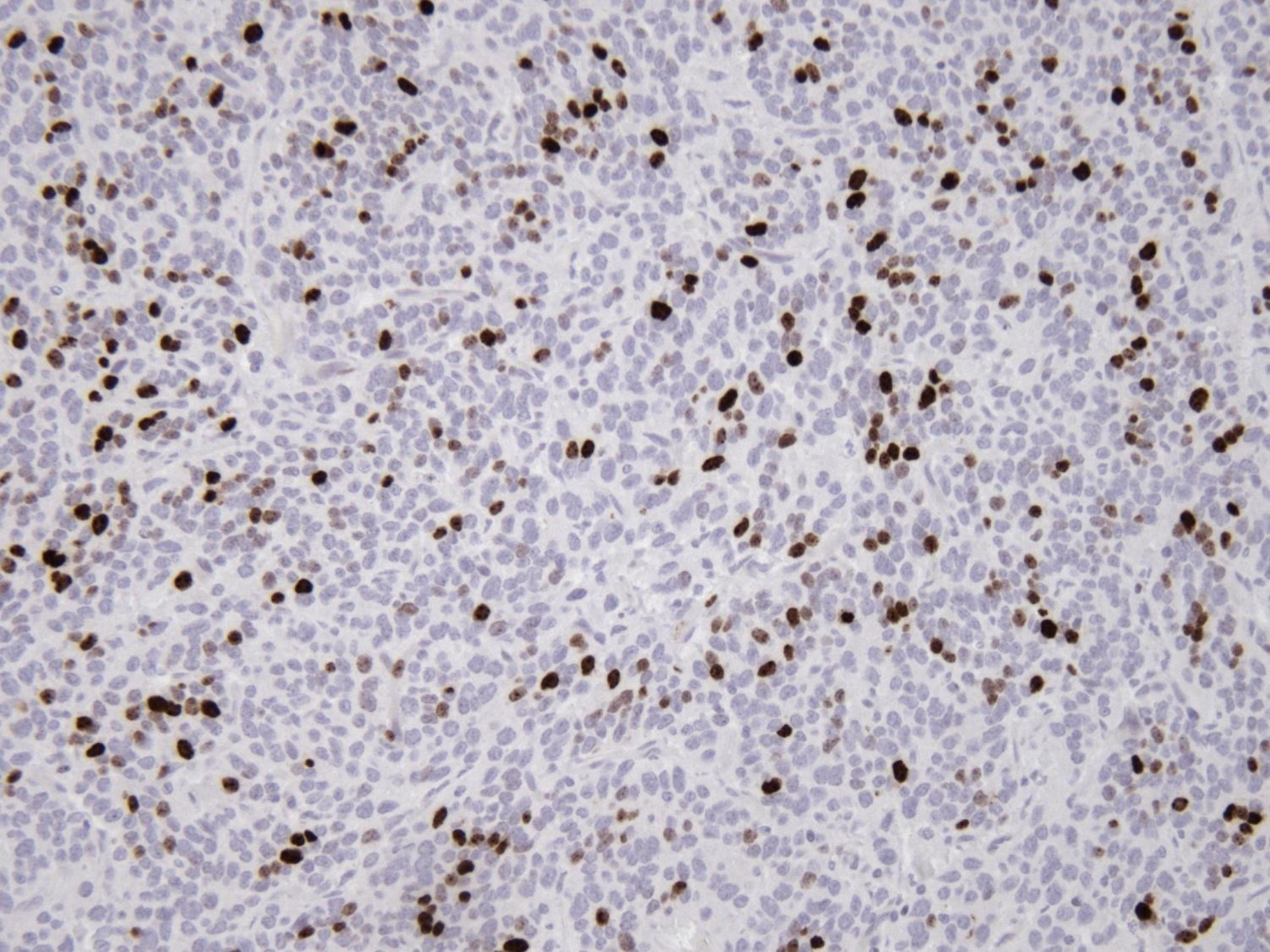








EMA



*Souhrn provedených molekulárně patologických vyšetření:*

Ve vyšetřovaném vzorku jsme metodou RT-PCR detekovali přítomnost fúzního genu ***C11orf95/RELA***

***Anaplastický ependymom, WHO grade III,  
RELA-fúze pozitivní.***