SOCIETAT CATALANA D'ANATOMIA PATOLÒGICA

Tumores del Sistema Nervioso Central

Sesión de Residentes

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Hospital Clínic de Barcelona

19/12/2013

Sant Joan de Déu HOSPITAL MATERNOINFANTIL - UNIVERSITAT DE BARCELONA

CLÍNIC BARCELONA Hospital Universitari

Historia Clínica

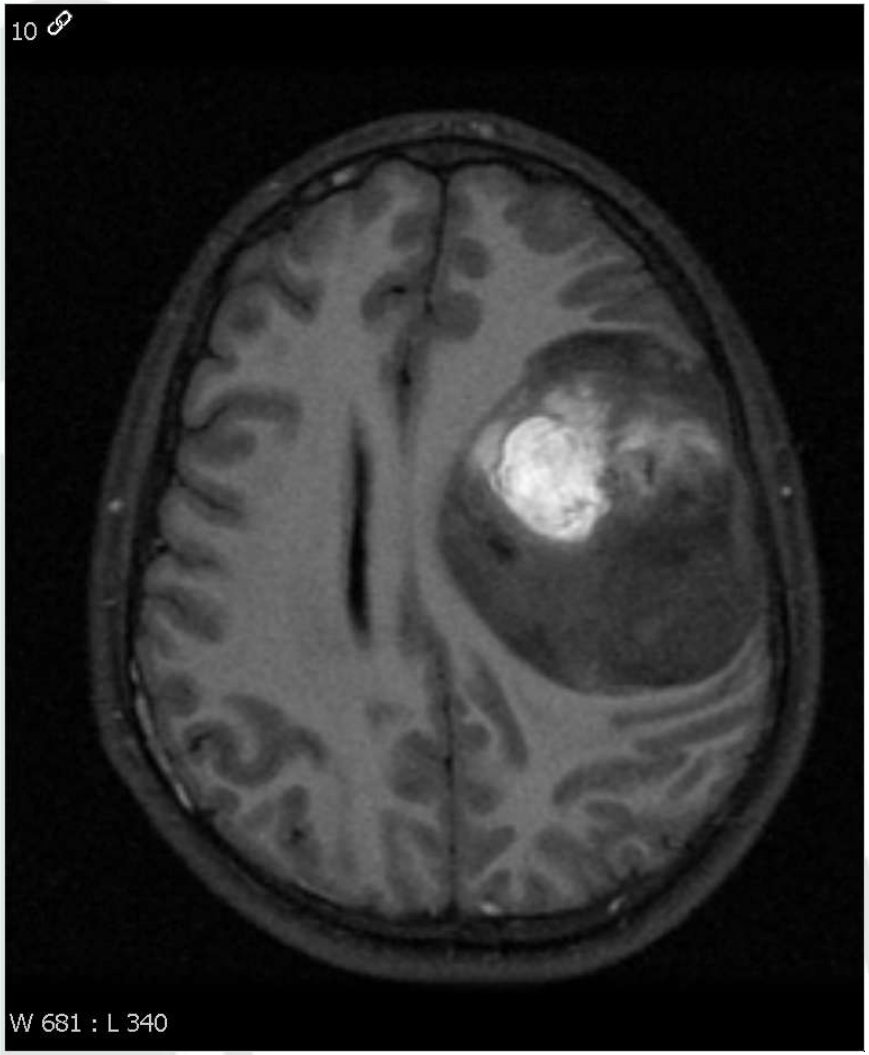
- Varón de 14a / 8m
- Antecedentes: Trastorno de conducta

Episodio actual

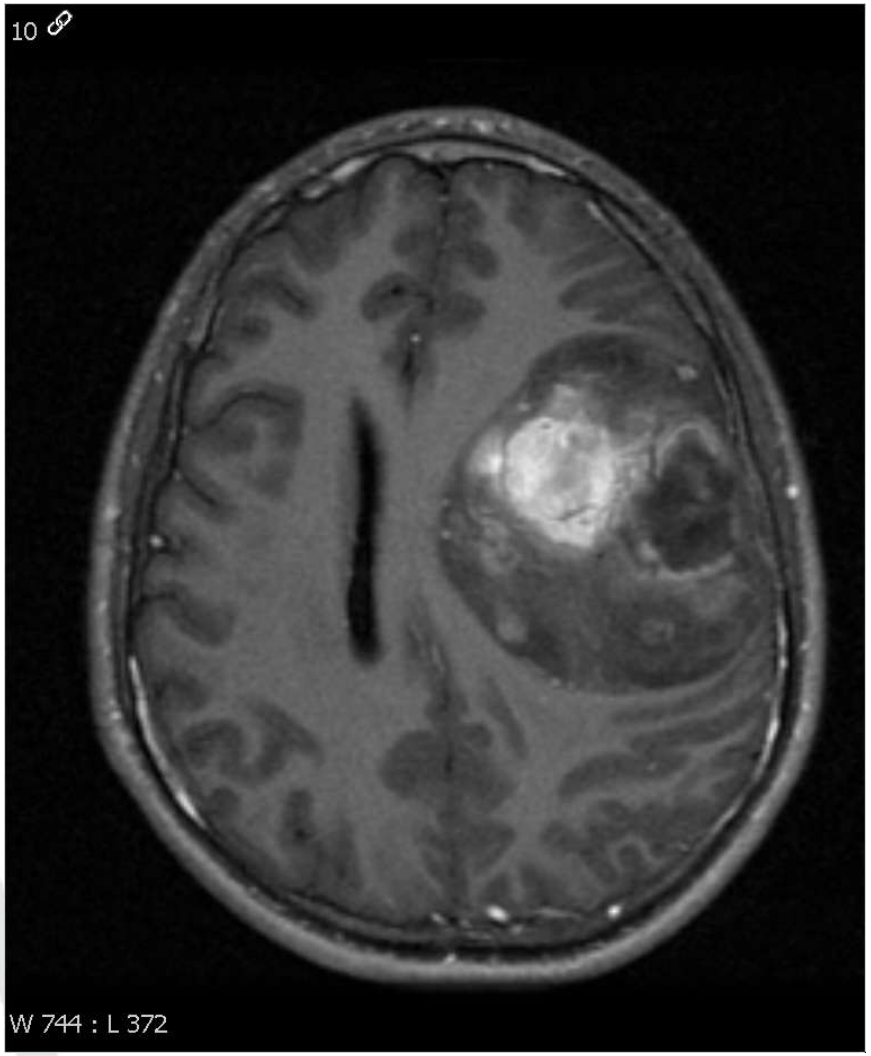
- Otro centro:
  - Cefalea y vómitos de 12h de evolución
  - *TC craneal*: masa en región parietal izquierda

- Al ingreso: focalidad neurológica, diplopia…

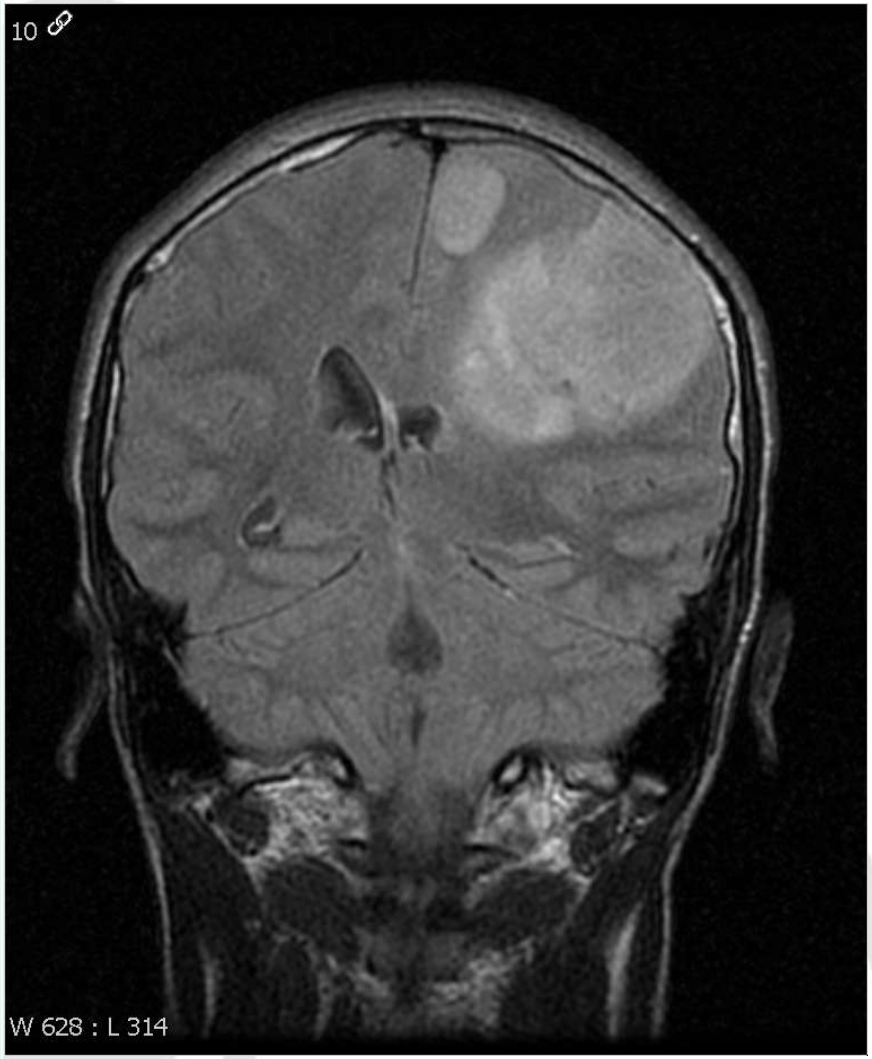
- UCI: 2º día: disminución del nivel de consciencia



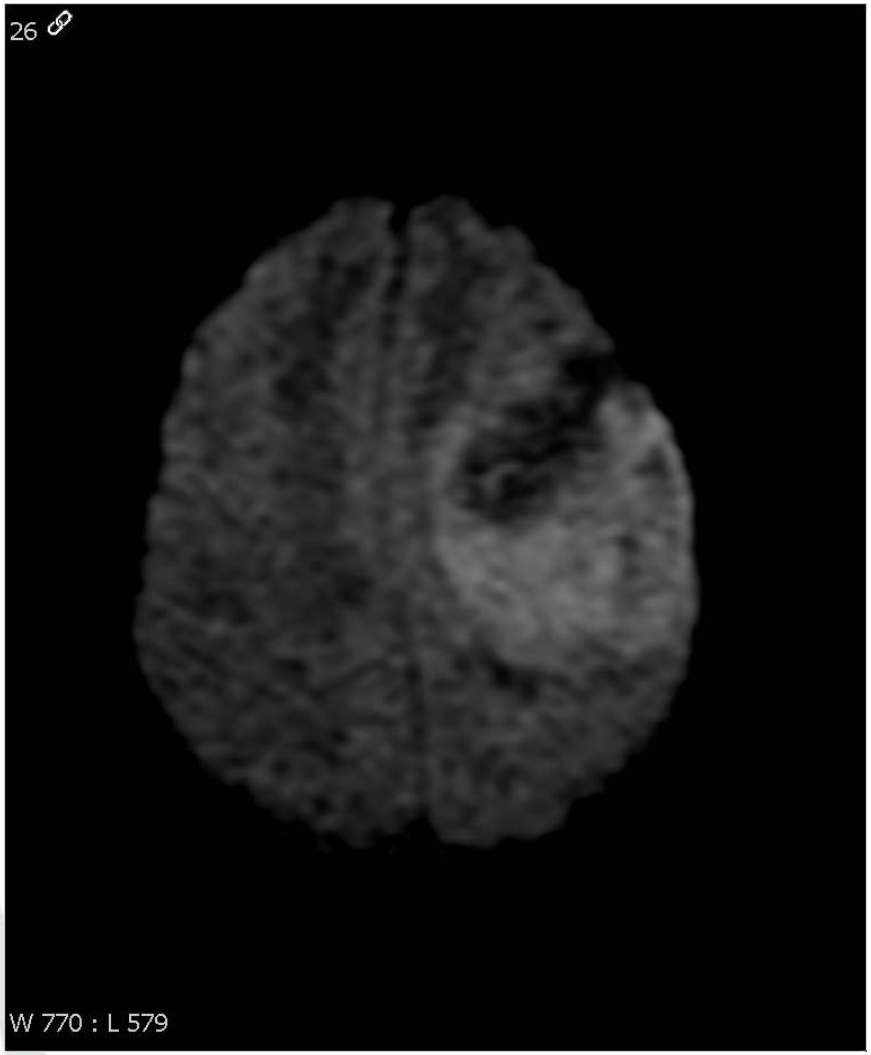
Axial FLAIR T1 fatsat



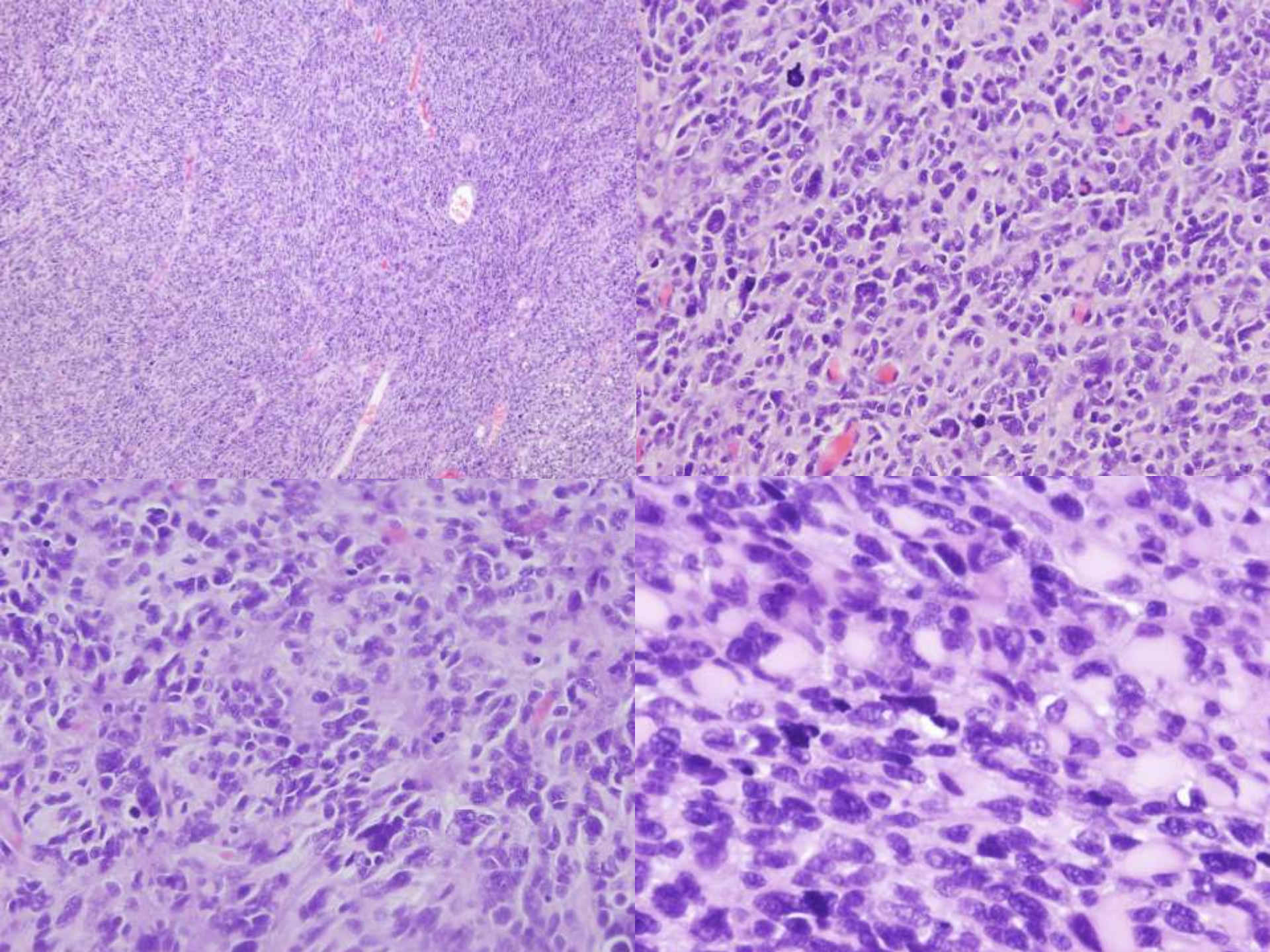
Axial FLAIR T1 fatsat + C

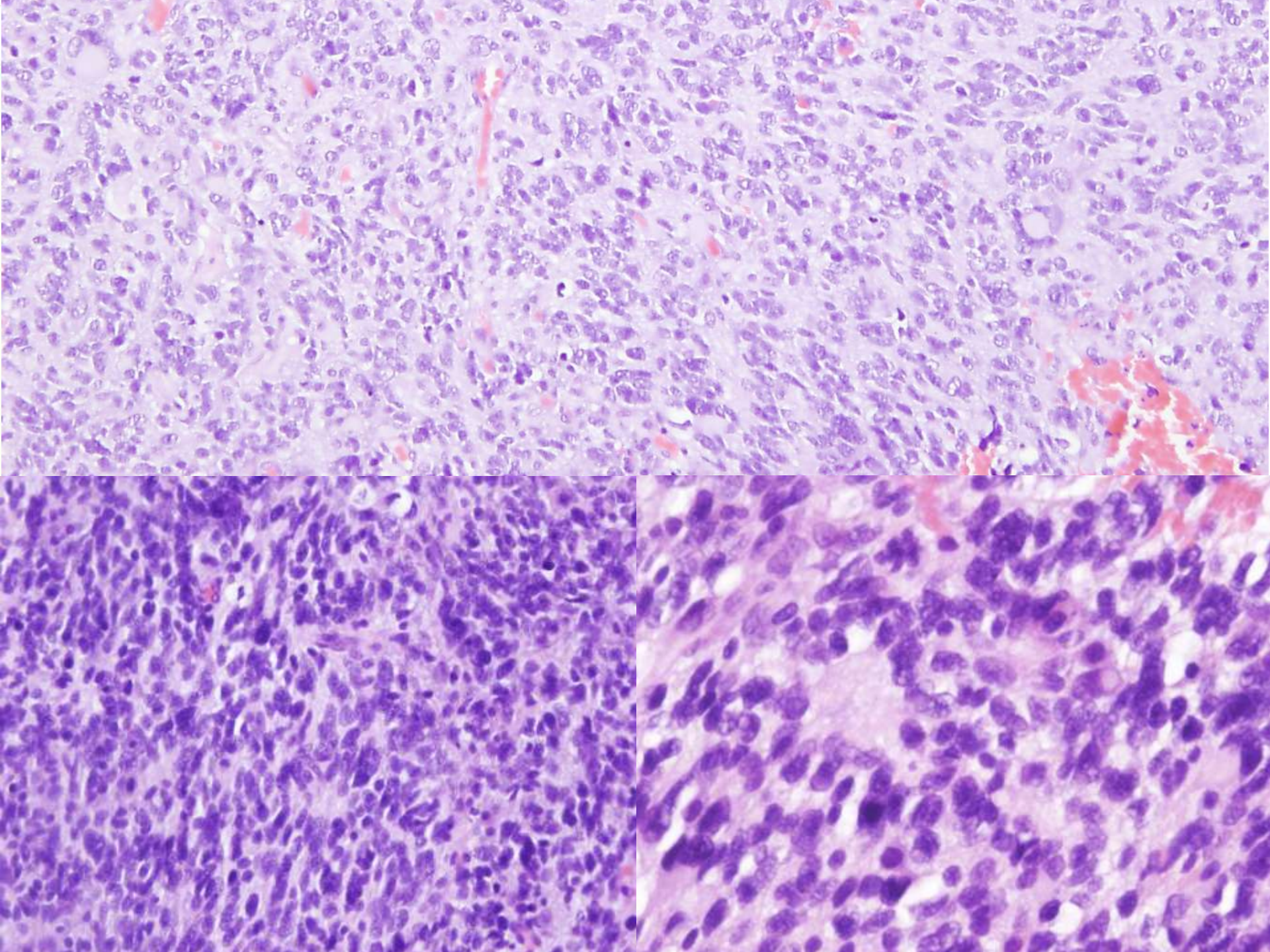


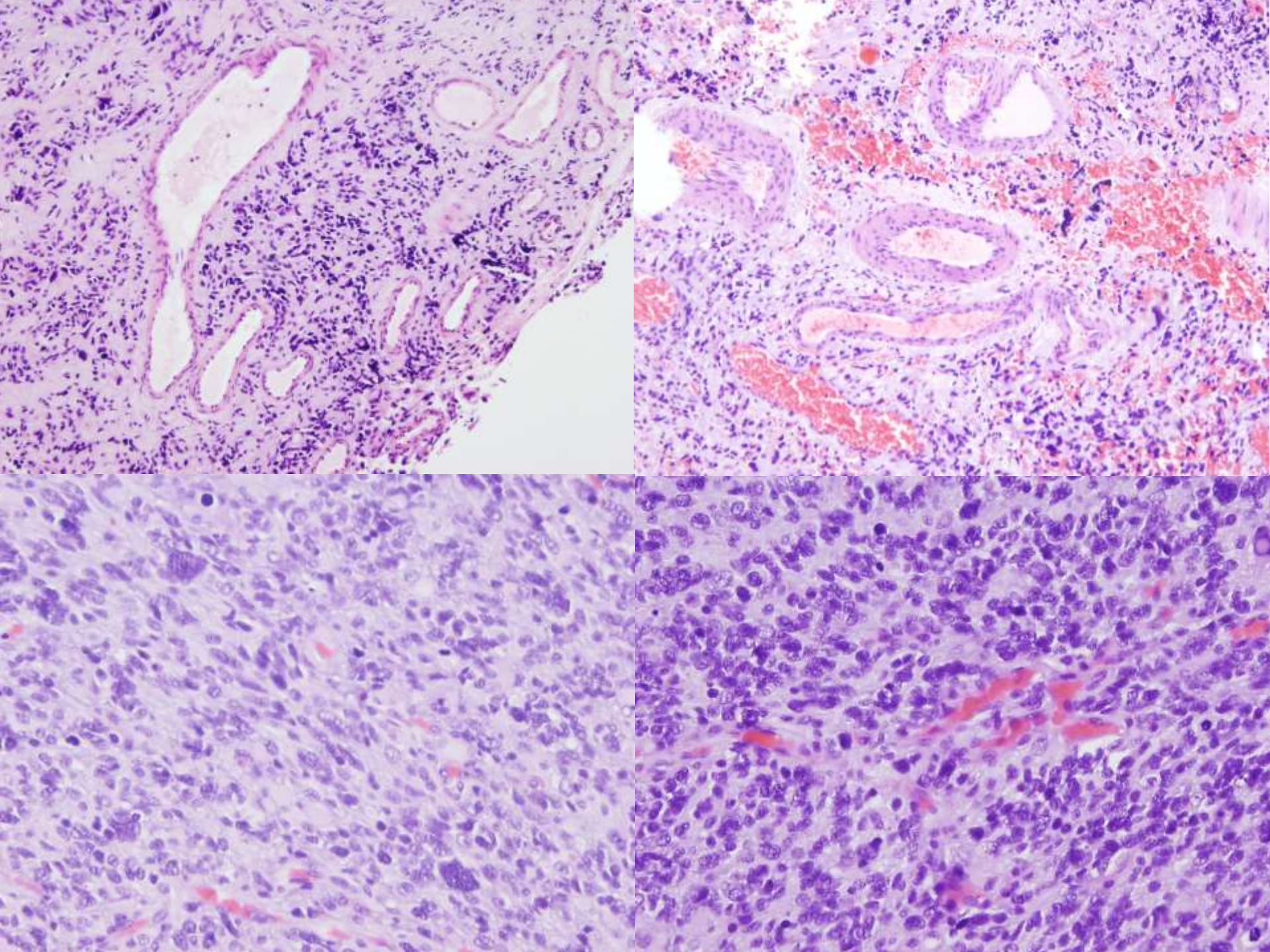
Coronal FLAIR T2

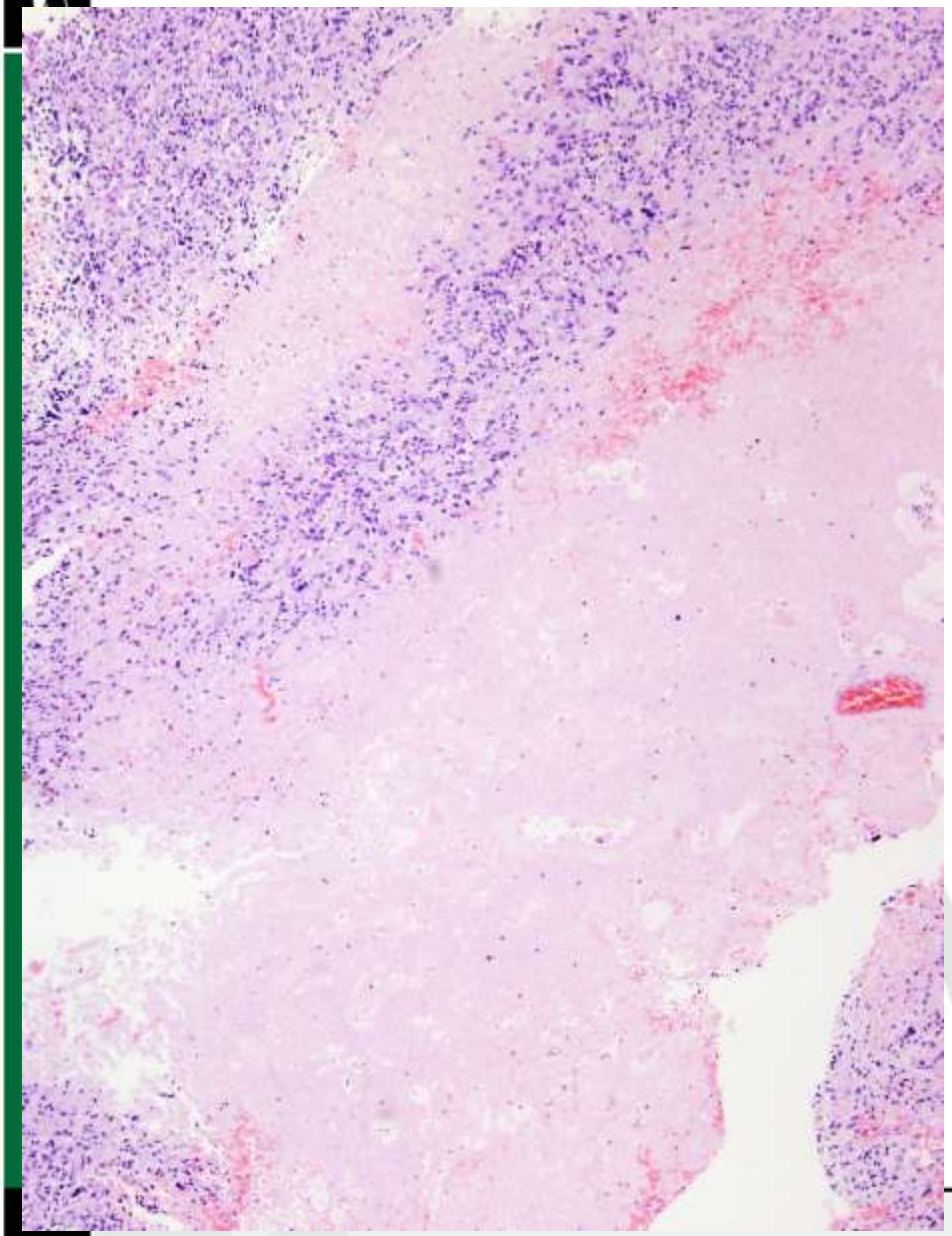


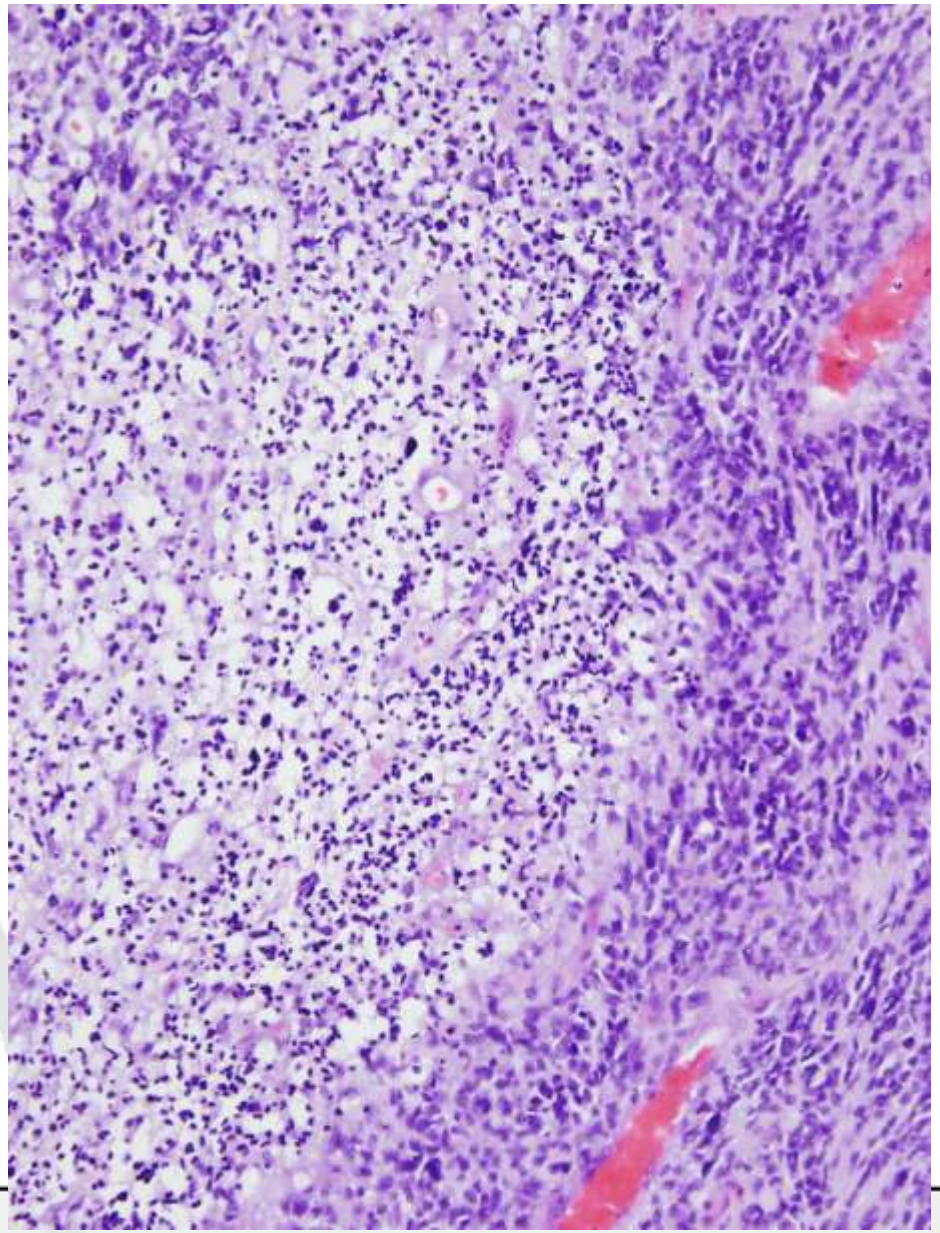
Axial DWI





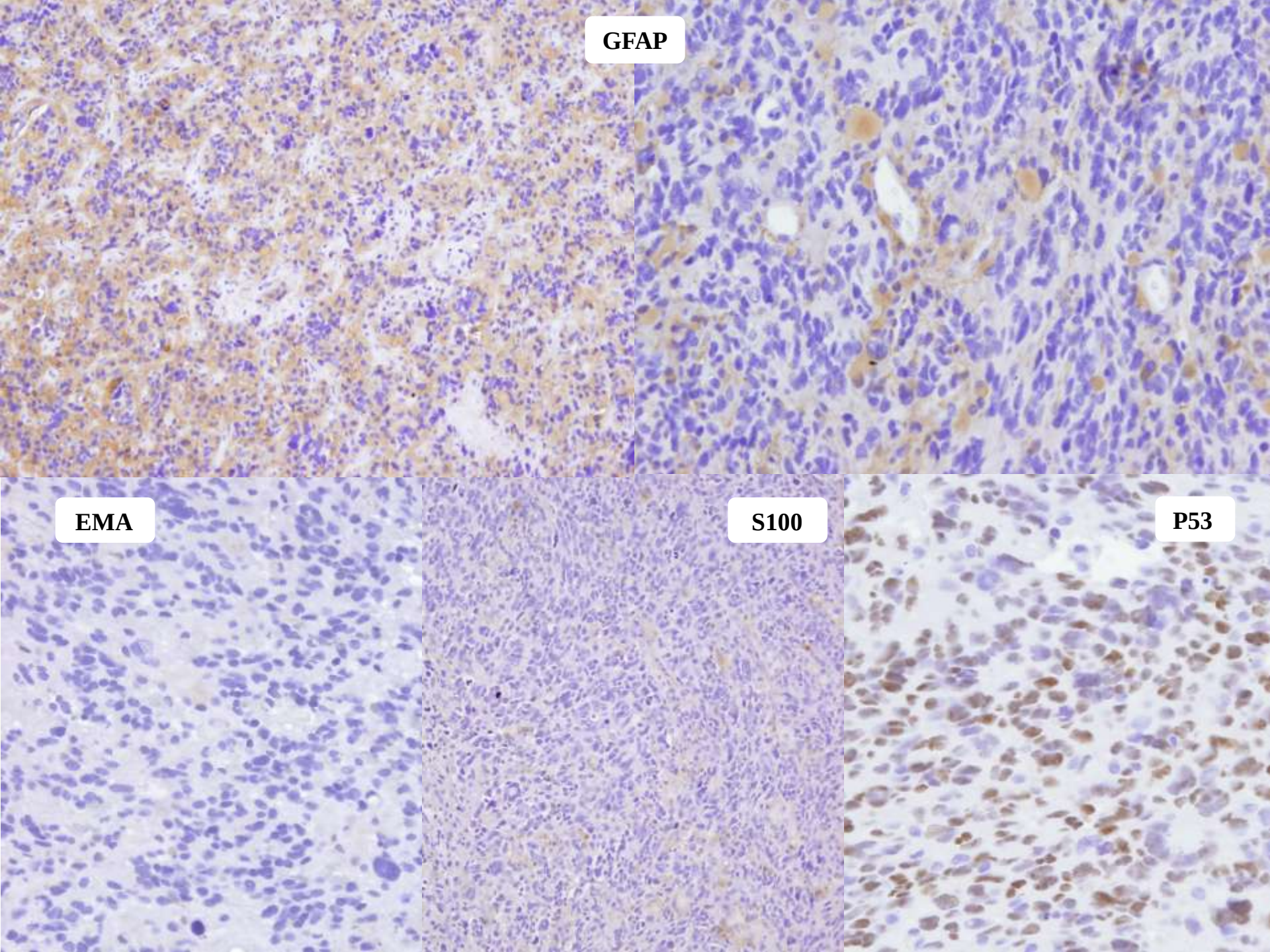


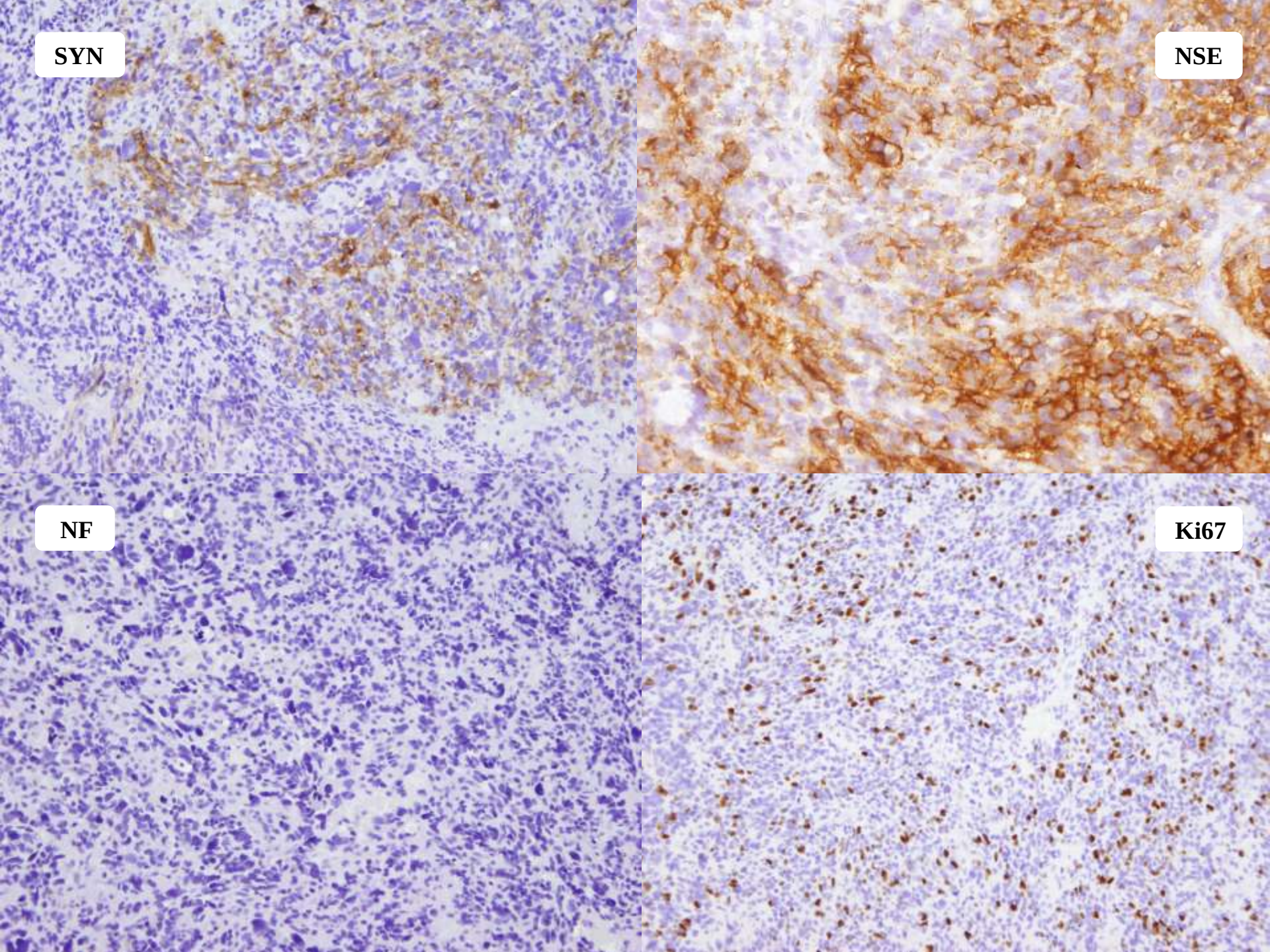


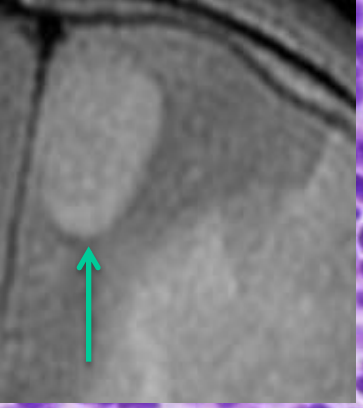


Diagnóstico diferencial

Niños	%	Adultos	%
Primarios	>99	Primarios	40-70
Astro I-II	45	Astro I-II	20
Astro III-IV	**5**	**Astro III-IV**	**55**
Ependimoma	16	Ependimoma	6
Oligodendroglioma	<1	Oligodendroglioma	10
Meningioma	2	Meningioma	20
MDB/PNETc	**21**	**MDB/PNETc**	**raro**
AT/RT	2	AT/RT	0
Metástasis	raro	Metástasis	30-60


GFAP
EMA
S100
P53


SYN
NSE
NF
Ki67



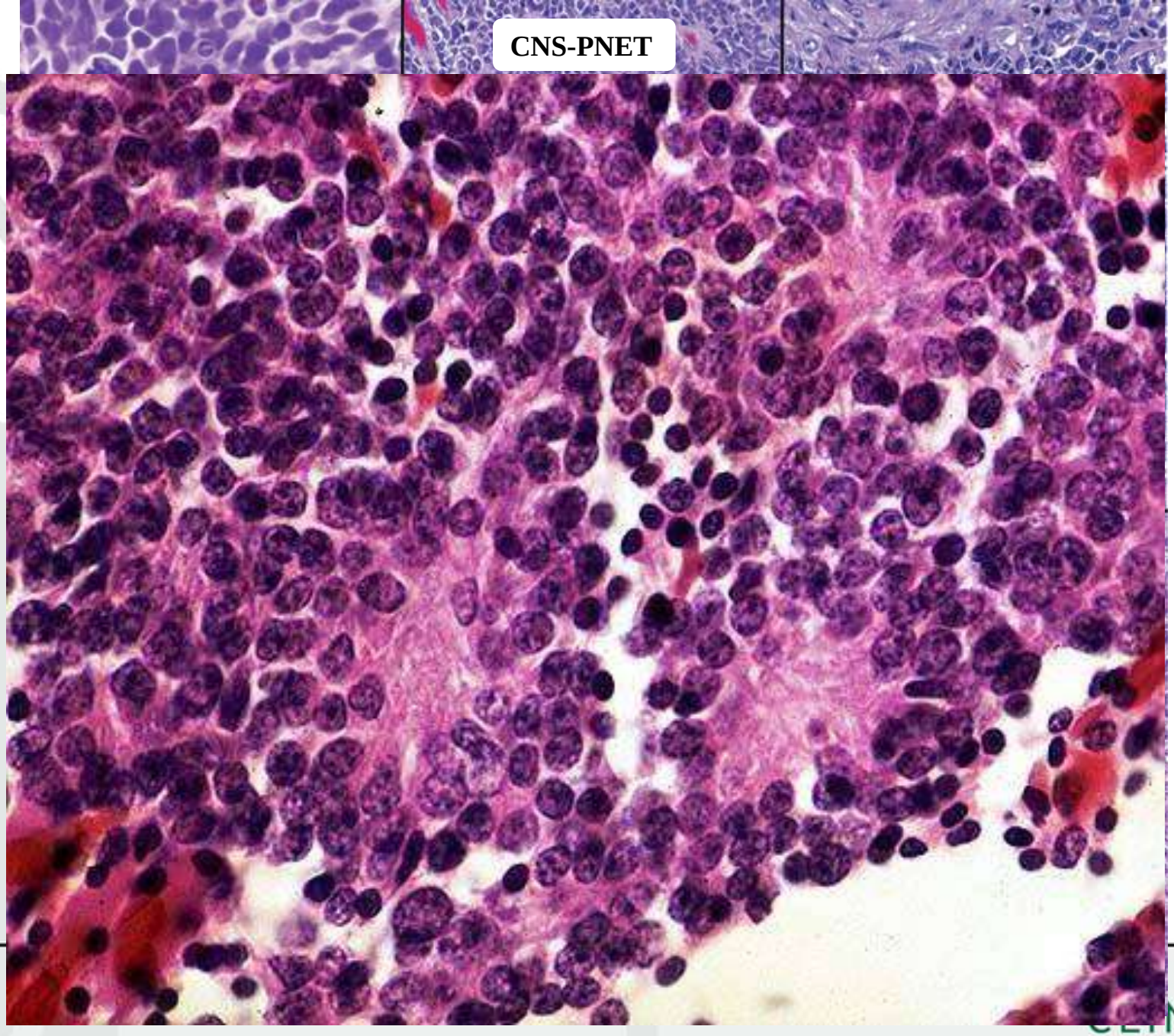
Diagnóstico

- CNS-PNET
- Glioblastoma Multiforme

Tumores embrionarios (WHO, 2007)

PNET (IV)

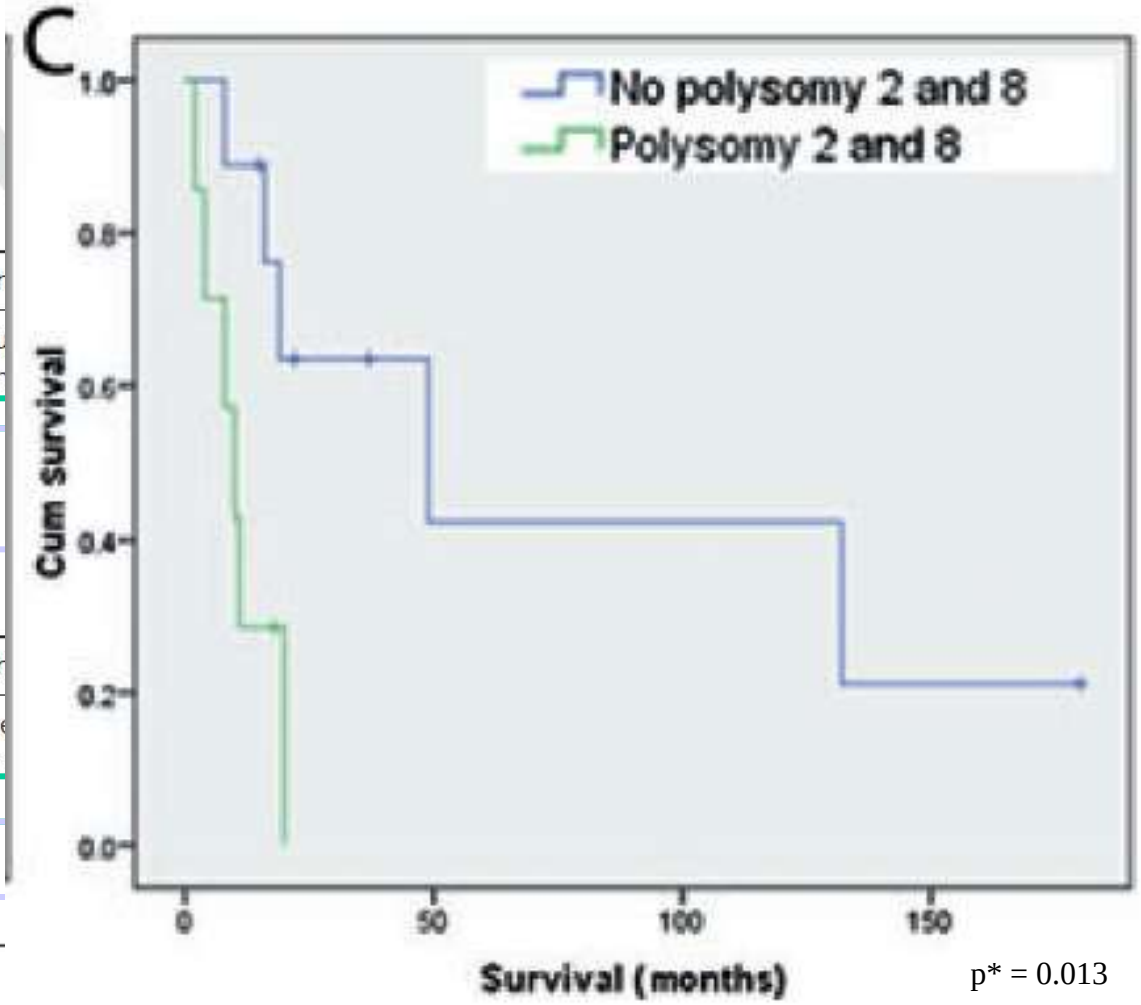
Medulloblastoma	9470/3
Desmoplastic/nodular medulloblastoma	9471/3
Medulloblastoma with extensive nodularity	*9471/3*
Anaplastic medulloblastoma	*9474/3*
Large cell medulloblastoma	9474/3
CNS primitive neuroectodermal tumour	9473/3
CNS Neuroblastoma	9500/3
CNS Ganglioneuroblastoma	9490/3
Medulloepithelioma	9501/3
Ependymoblastoma	9392/3
Atypical teratoid / rhabdoid tumour	9508/3



Behdad A,Perry A. Central Nervous System Primitive Neuroectodermal Tumors: A Clinicopathologic and Genetic Study of 33 Cases. Brain Pathology 20 (2010) 441–450

Histologic features/protein	Pediatric (%)
Anaplastic/Large cell featu	41
Homer Wright Rosettes (r	9.5
GFAP ($n = 26$)	0
Synaptophysin ($n = 23$)	43
Neurofilament ($n = 10$)	52
Neu-N ($n = 14$)	5
CD99 ($n = 11$)	
CAM 5.2 ($n = 9$)	

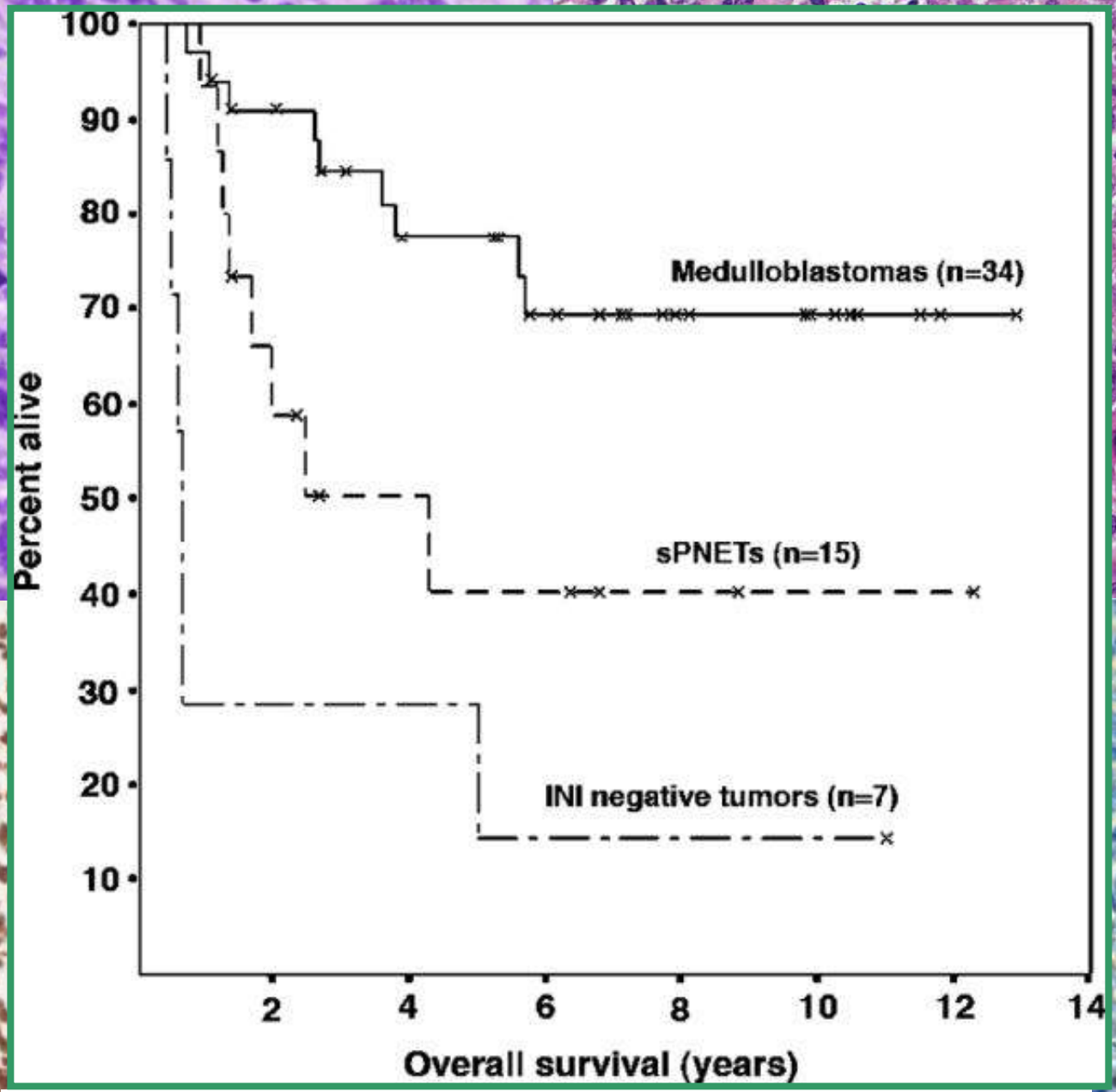
Histologic features/protein	Adults (%)
Anaplasia/large cell feature	0
Homer Wright rosettes (n	16
GFAP ($n = 5$)	0
Synaptophysin ($n = 6$)	100
Neurofilament ($n = 5$)	100
	33



p* = 0.013

Behdad A,Perry A. Central Nervous System Primitive Neuroectodermal Tumors: A Clinicopathologic and Genetic Study of 33 Cases. Brain Pathology 20 (2010) 441–450

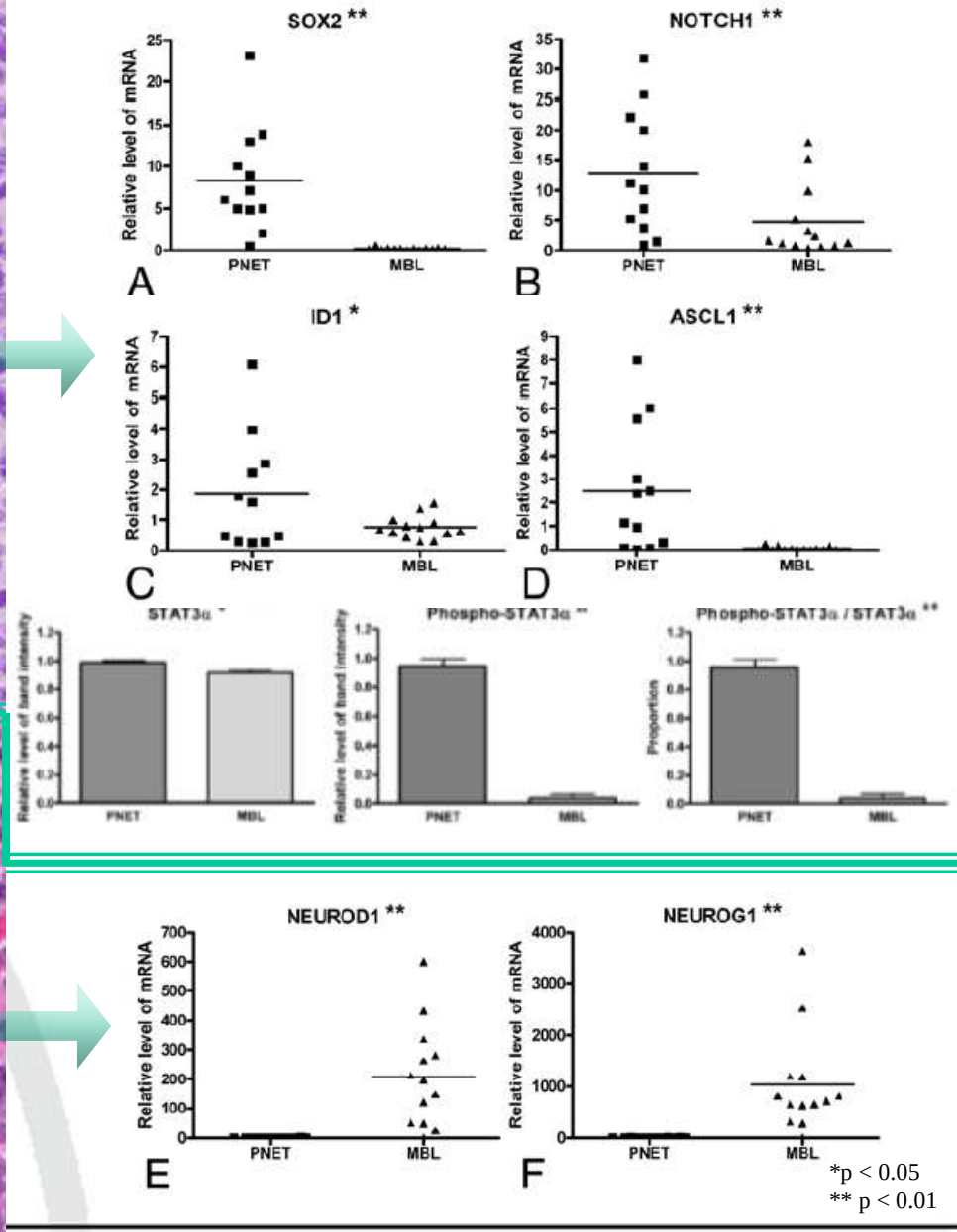
CNS-PNET



Haberler C et al. Immunohistochemical Analysis of INI1 Protein in Malignant Pediatric CNS Tumors: Lack of INI1 in Atypical Teratoid/ Rhabdoid Tumors and in a Fraction of Primitive Neuroectodermal Tumors without Rhabdoid Phenotype. American Journal of Surgical Pathology 2006;30:1462-1468

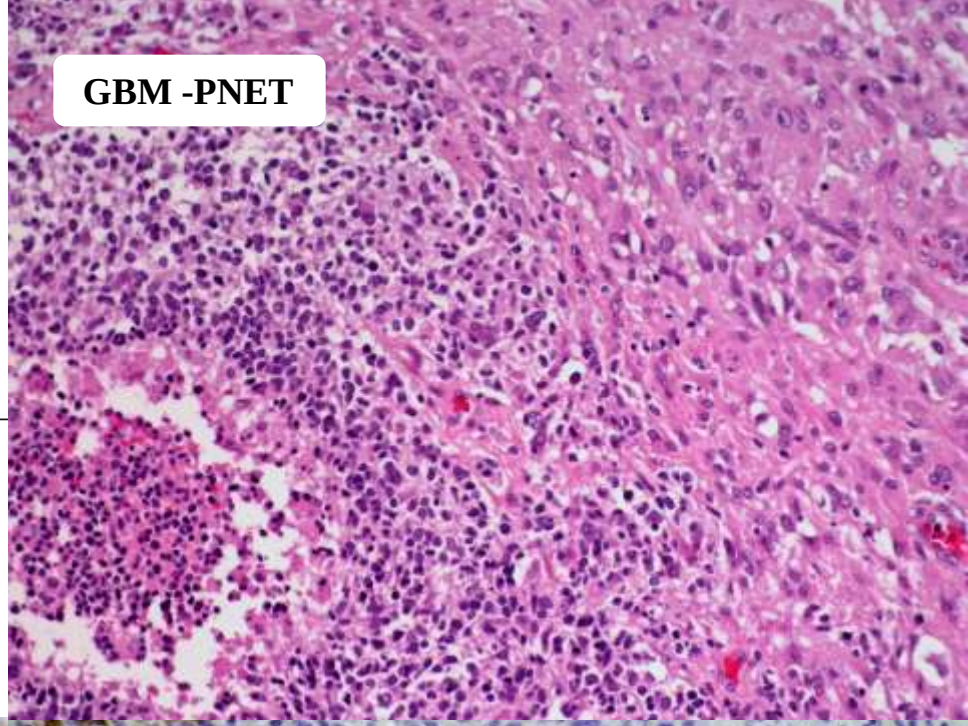
CNS-PNET

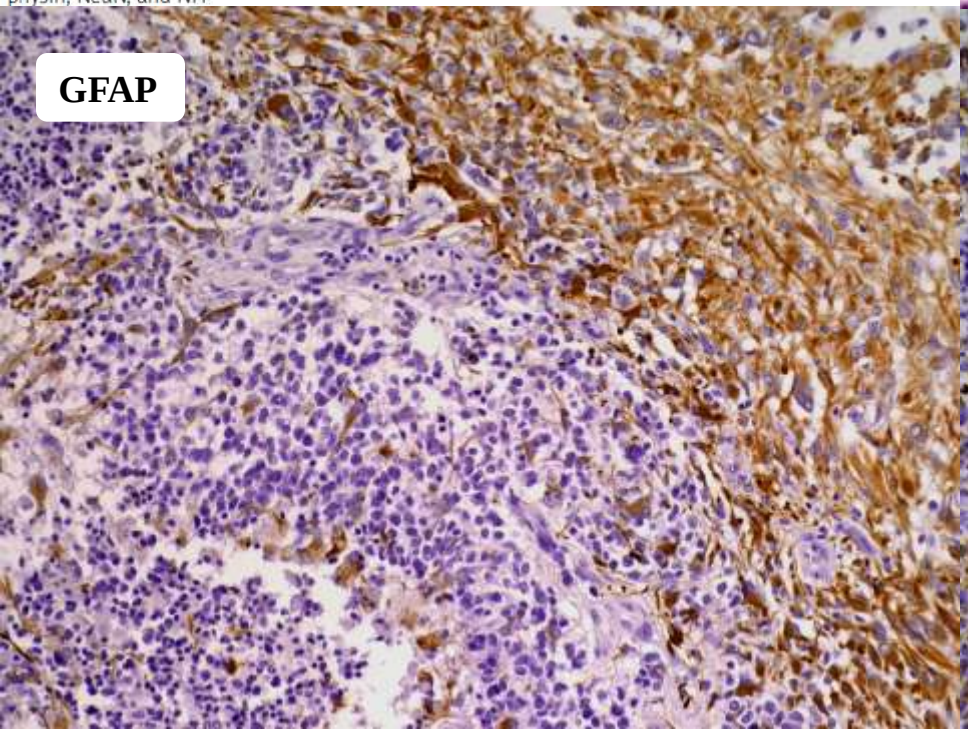
Medulloblastoma

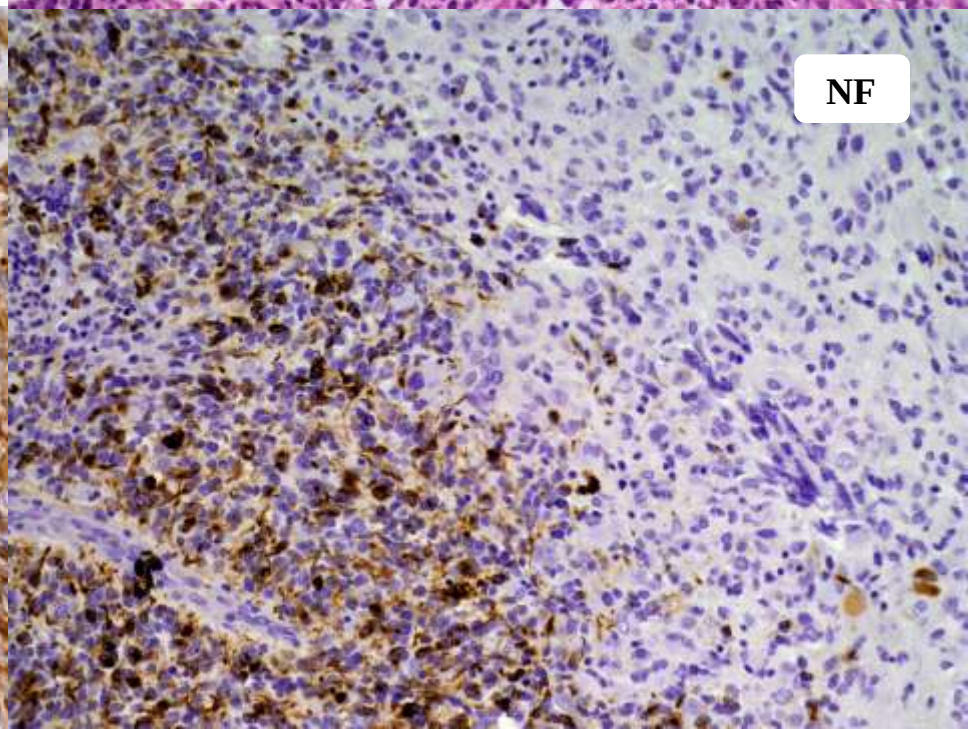


Ji Hoon Phi et al. Upregulation of *SOX2*, *NOTCH1*, and *ID1* in supratentorial primitive neuroectodermal tumors: a distinct differentiation pattern from that of medulloblastomas. J Neurosurg Pediatrics 5:000–000, 2010.

GBM variant and morphological features	Molecular alterations	Prognostic markers	Survival
Classic GBM Infiltrating, pleomorphic, hyperchromatic cells with glassy, astrocytic cytoplasm. Frequent presence of pseudopalisading necrosis, neoepithelialization, mitotic figures, and hypercellularity	EGFR, EGFRvIII, $p16^{INK4A}$, PTEN, p53, MGMT, PI3K/AKT, DH1; Loss chromosome: 1p, 10, 19q **Genomic subtypes:** **Proneural:** PDGF, IDH1/IDH2, p53, PI3KCA, PI3KR1 **Mesenchymal:** NF1, p53, PTEN **Proliferative/classical:** EGFR, EGFRvIII, PTEN, $p16^{INK4A}$ **Neural:** nonspecific	EGFRvIII, MGMT, IDH1, PTEN, p53, CD133, proneural subtype	5 year survival: 9.8% Median PFS: 5.3-10.3 months Median OS: 12.7-21.7 months
GBM with primitive neuroectodermal tumour (GBM-PNET) Features of GBM along with PNET-like areas showing hypercellularity, minimal fibrillary background, small undifferentiated cells with scant cytoplasm, oval-round hyperchromatic nuclei, and Homer Wright neuroblastic rosettes staining for S-100, synaptophysin, NeuN, and NFP	N-myc, C-myc, IDH1; Loss chromosome 10q	IDH1	Mean survival: 44 months







CNS PNET con diferenciación glial