



UNIVERSITÀ
DEGLI STUDI
DI TORINO

A hypothalamic lesion in a 46-year-old woman



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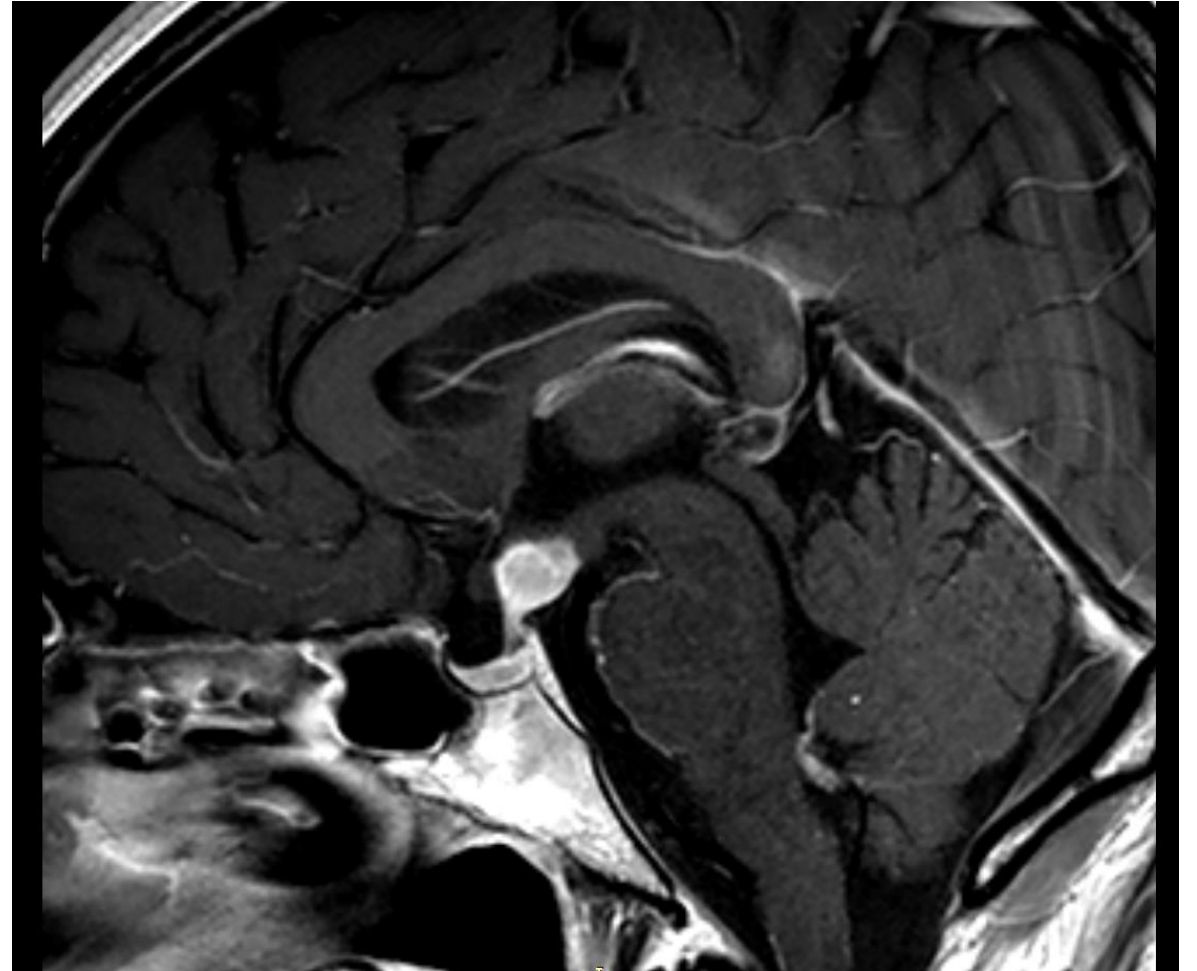
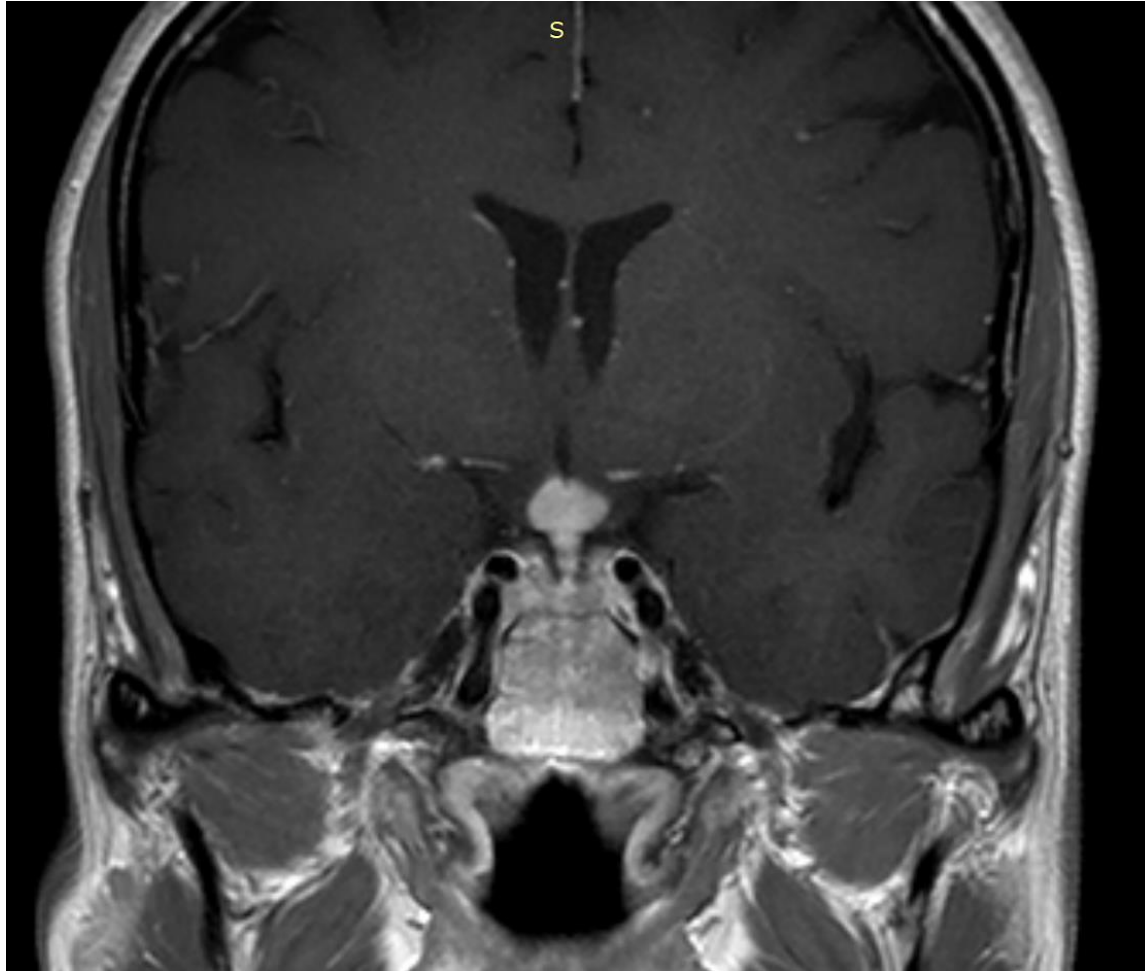
Dati clinici

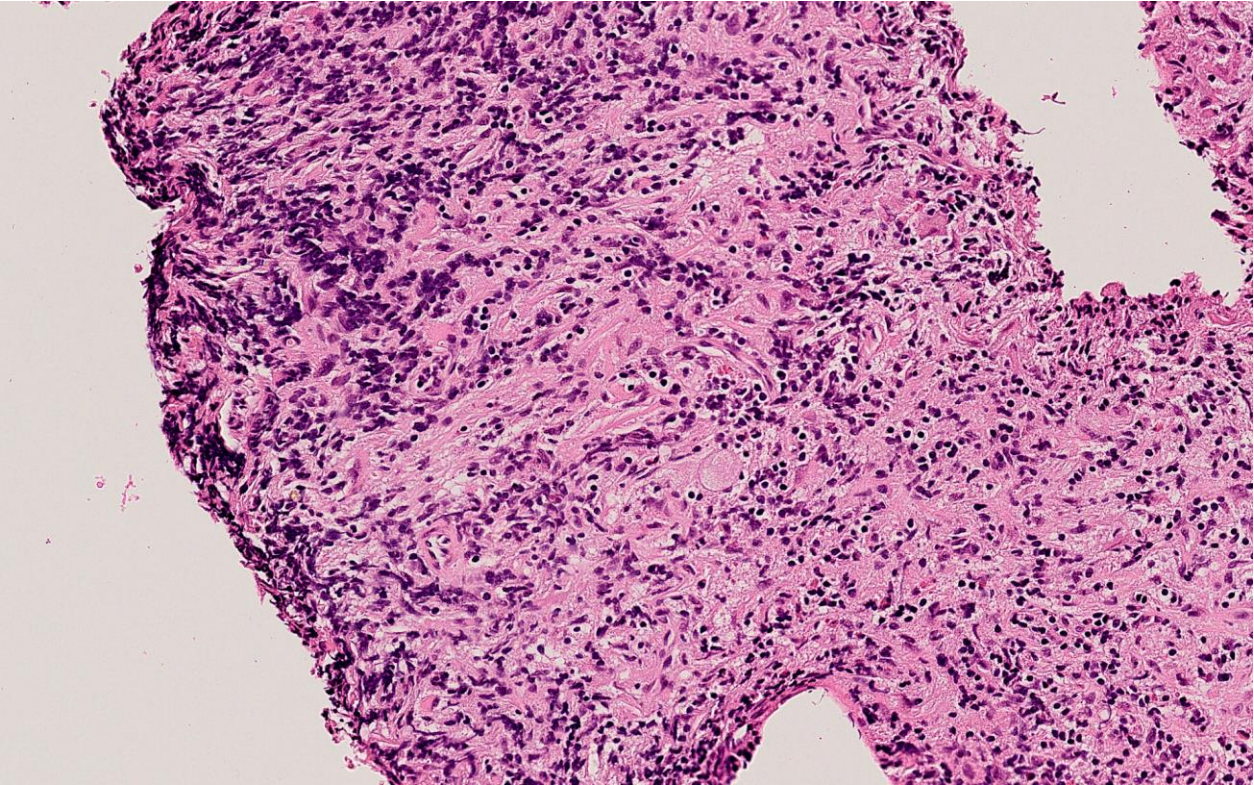
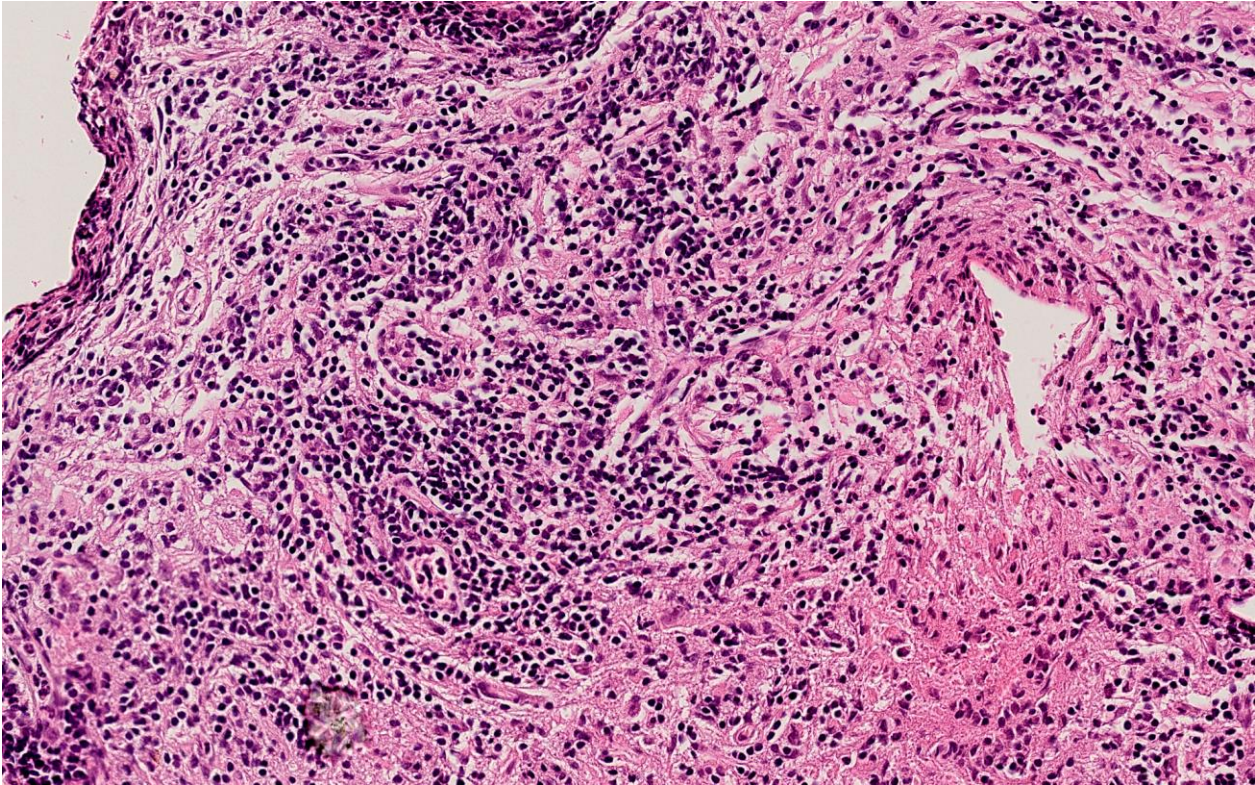
- F 46 anni
- APR: negativa

- APP: diabete insipido noto da circa 10 anni
- Lesione sovrasellare/ipotalamica con estensione chiasmatica

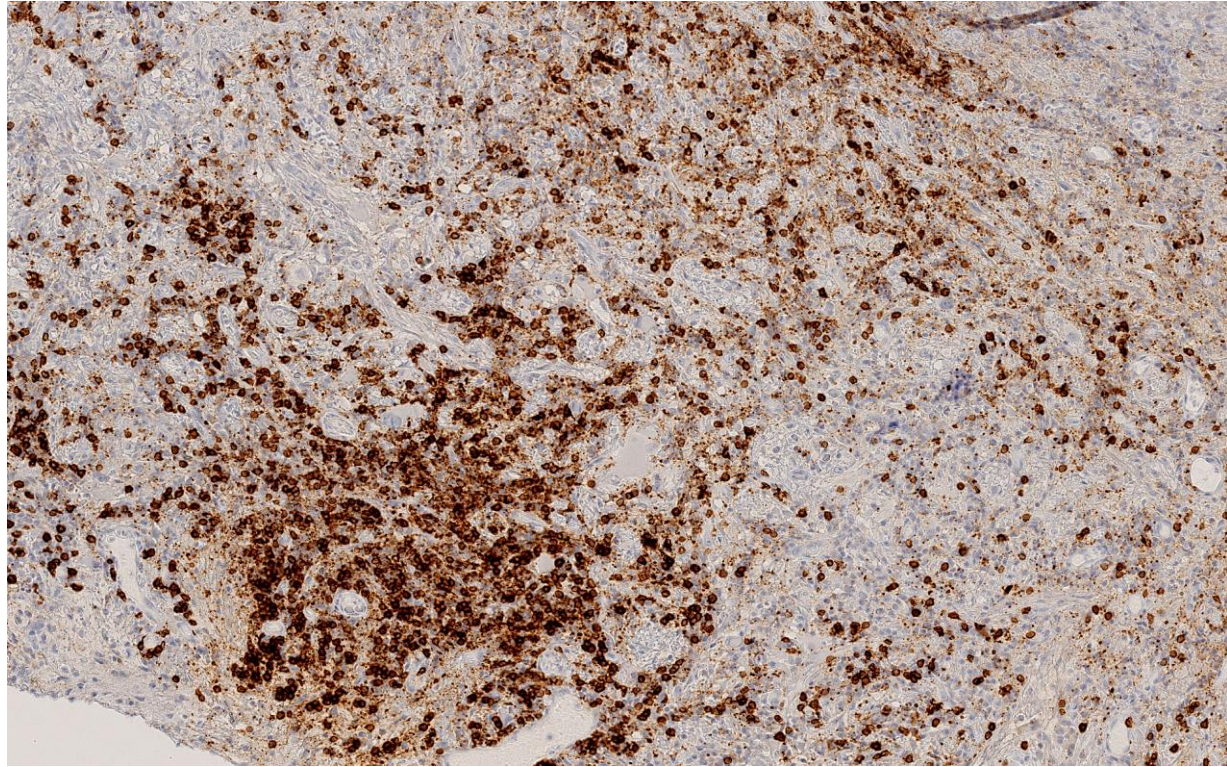
- Sospetto diagnostico: craniofaringioma? Pituicitoma?

RM pre-operatoria (T1 con mdc)

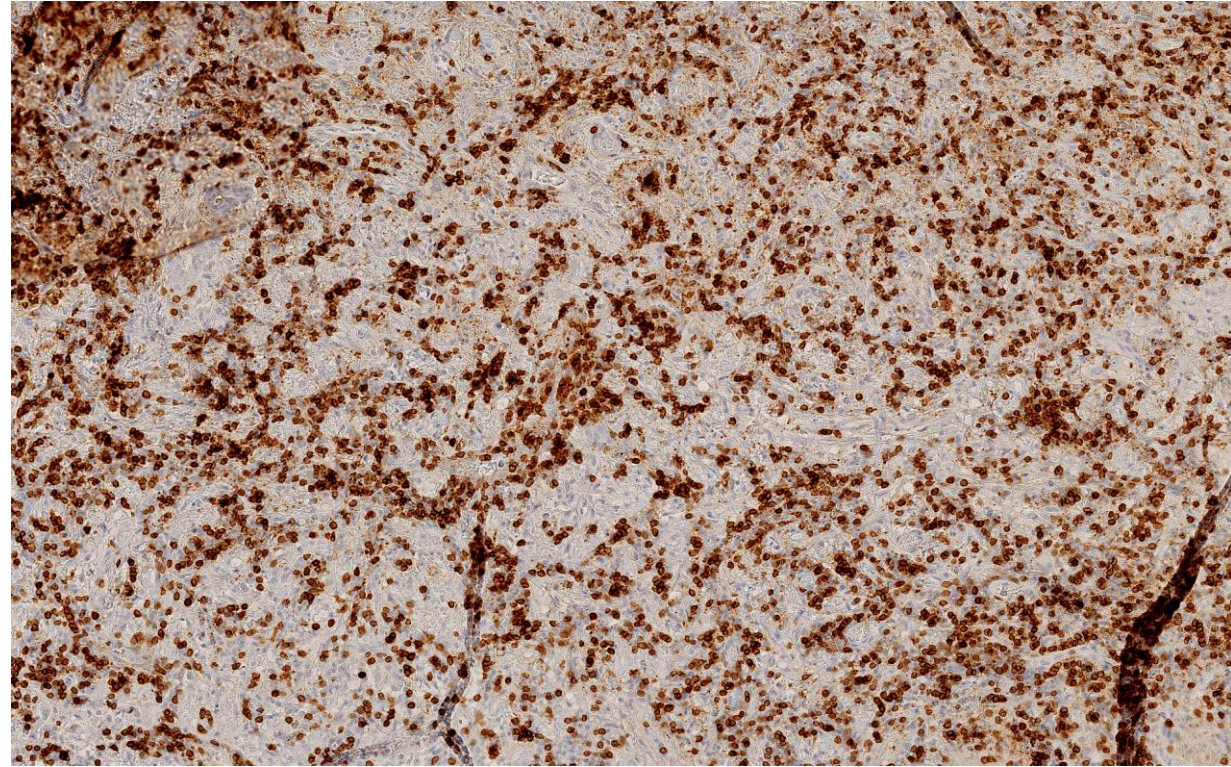




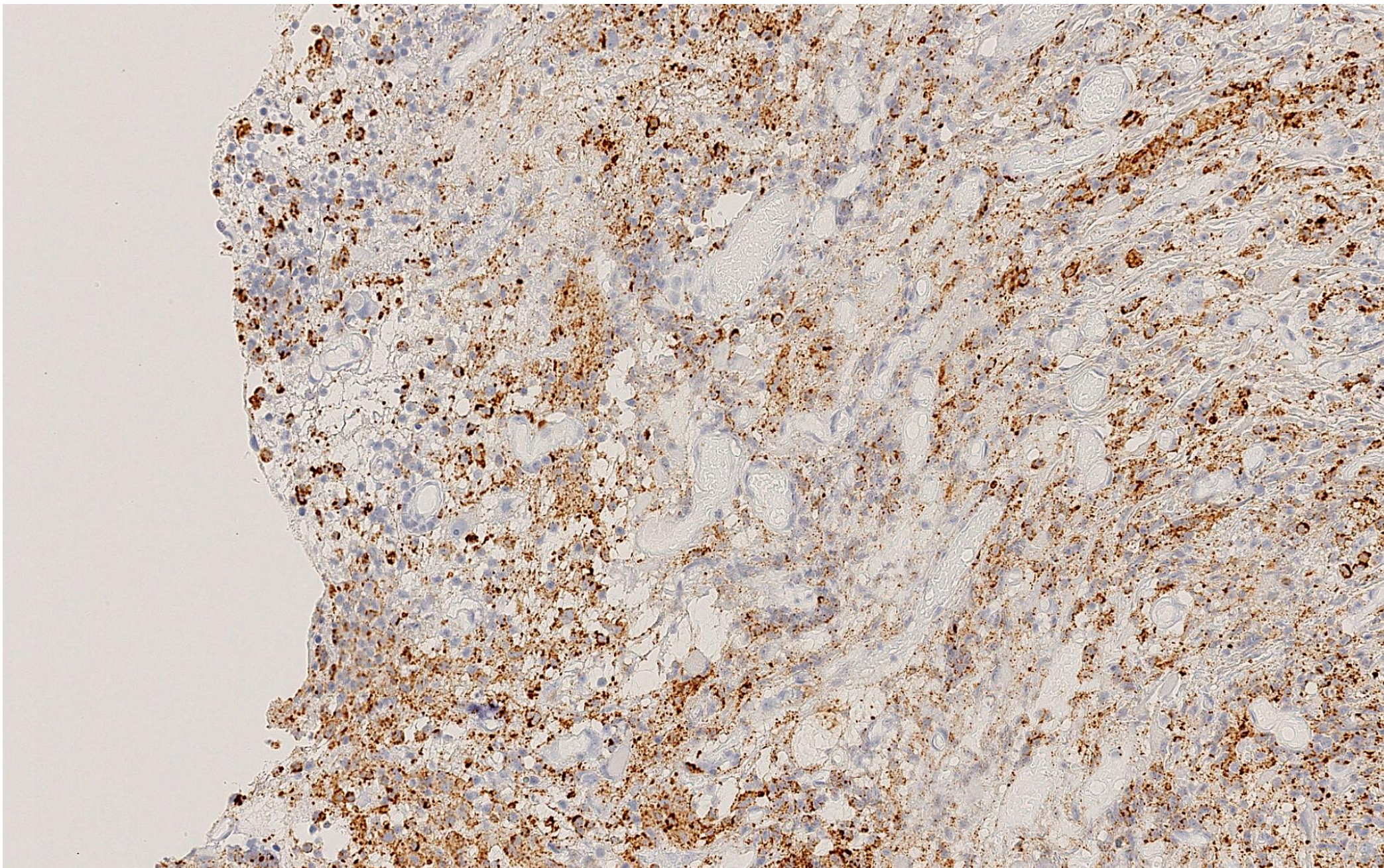
1° intervento - EE 200X



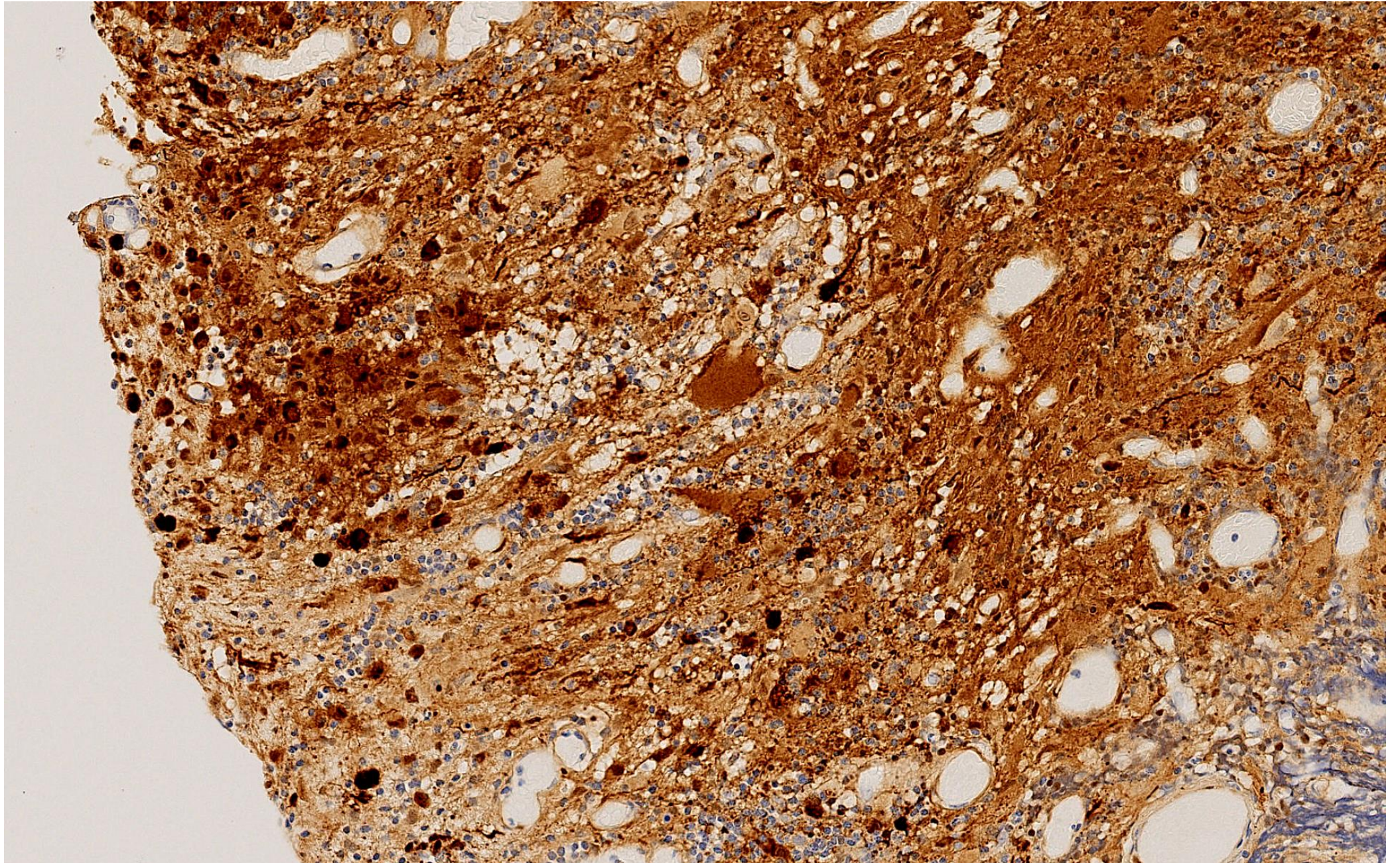
CD20 200X



CD3 200X



CD68 (PG-M1)
200X



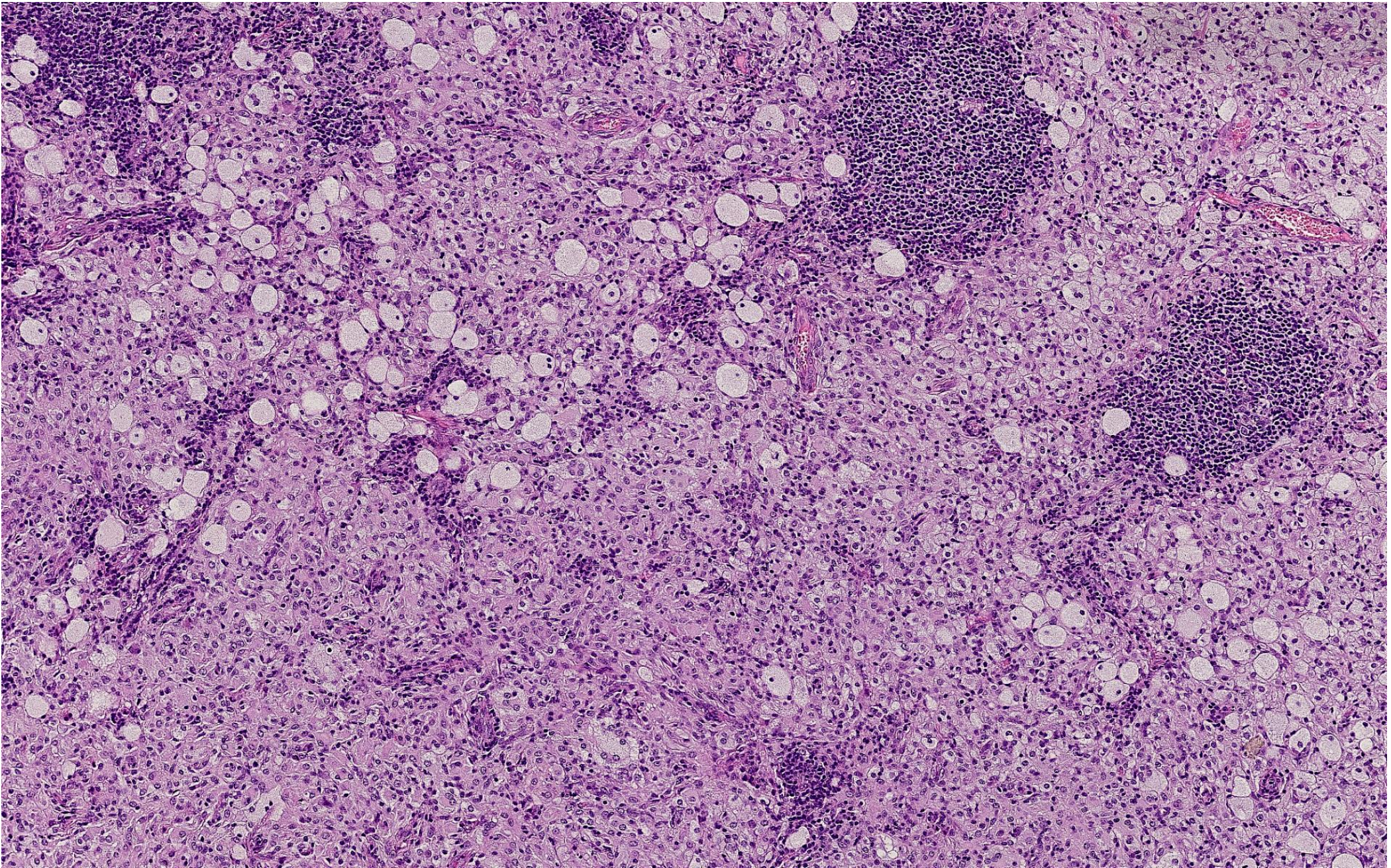
S100 200X

Diagnosi?

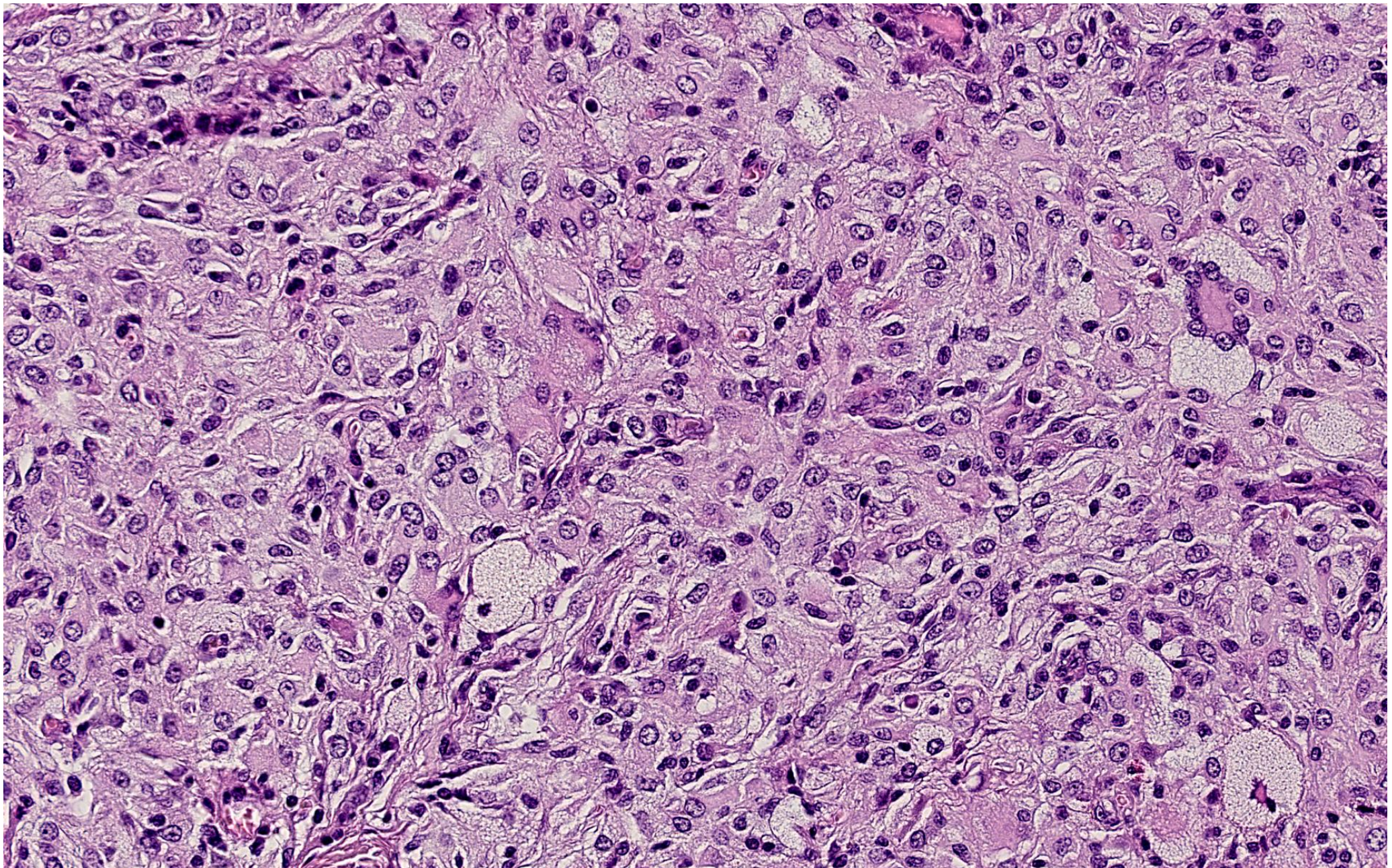
Diagnosi: Lesione infiammatoria con aspetti granulomatosi, potenzialmente compatibile con istiocitosi a cellule di Langerhans

Dati clinici

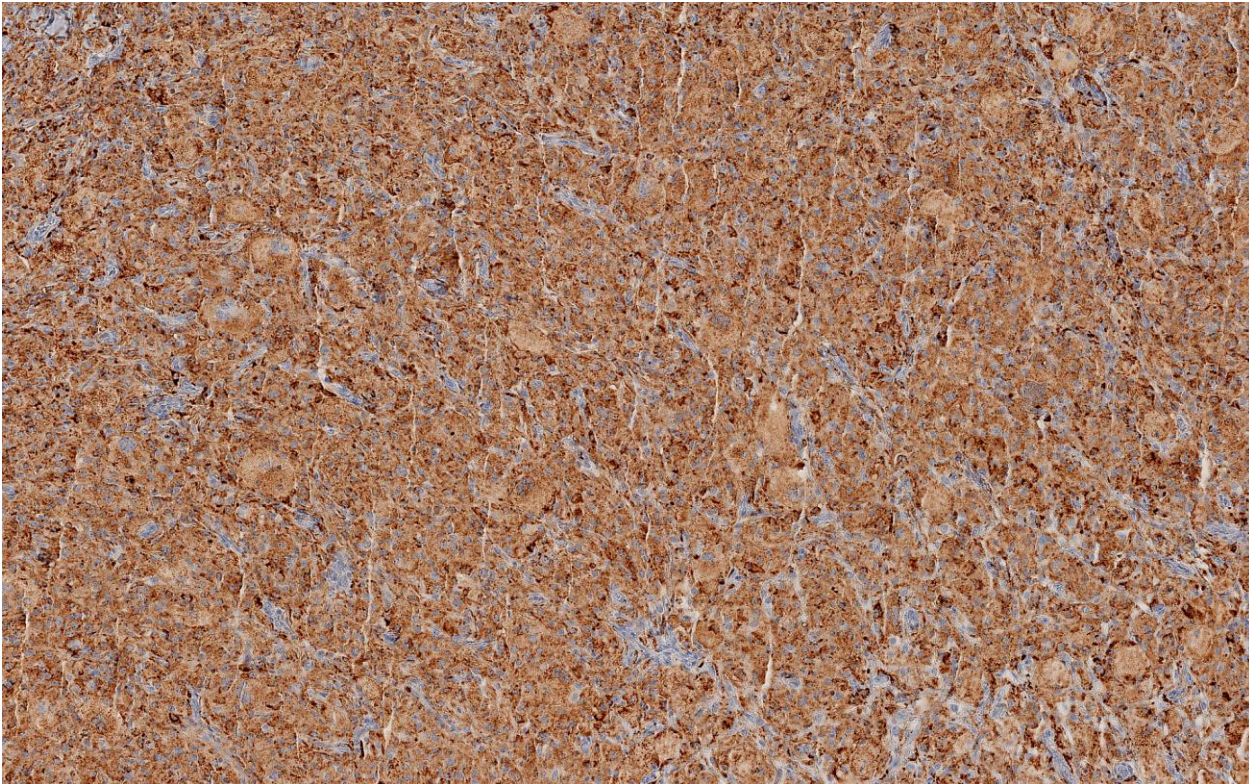
- RM encefalo post-operatoria: assenza di residuo di malattia
- Staging sistemico: negativo
- Non eseguite terapie adiuvanti con stabilità quadro clinico-radiologico
- Dopo 3 anni insorgenza di nuova lesione in sede meningeo frontale destra



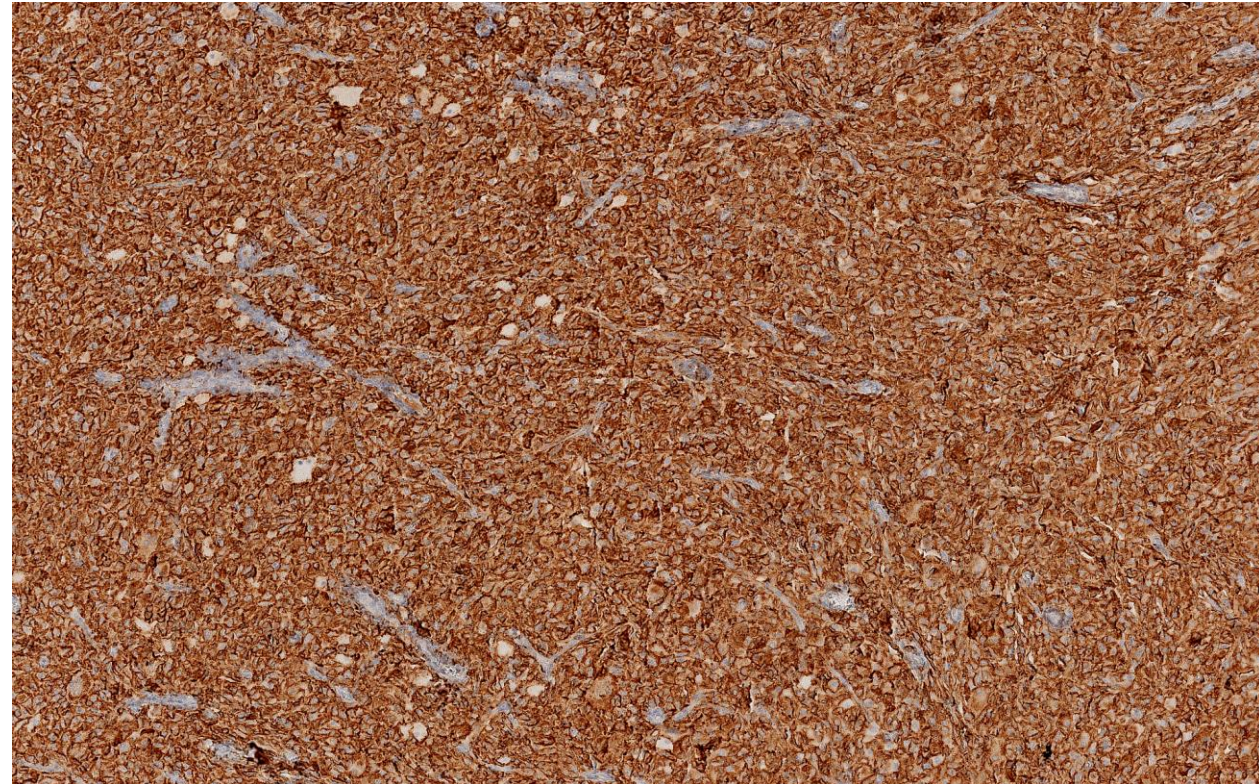
2° intervento - EE 100X



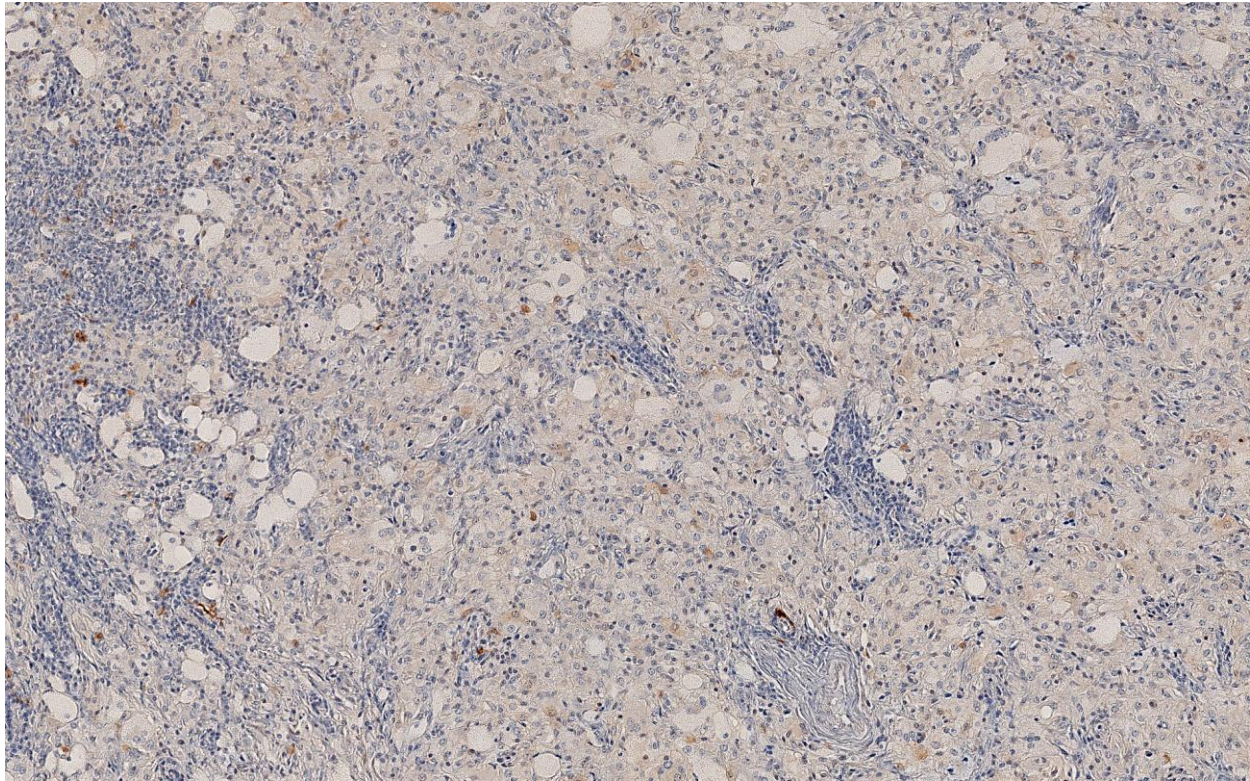
2° intervento - EE 300X



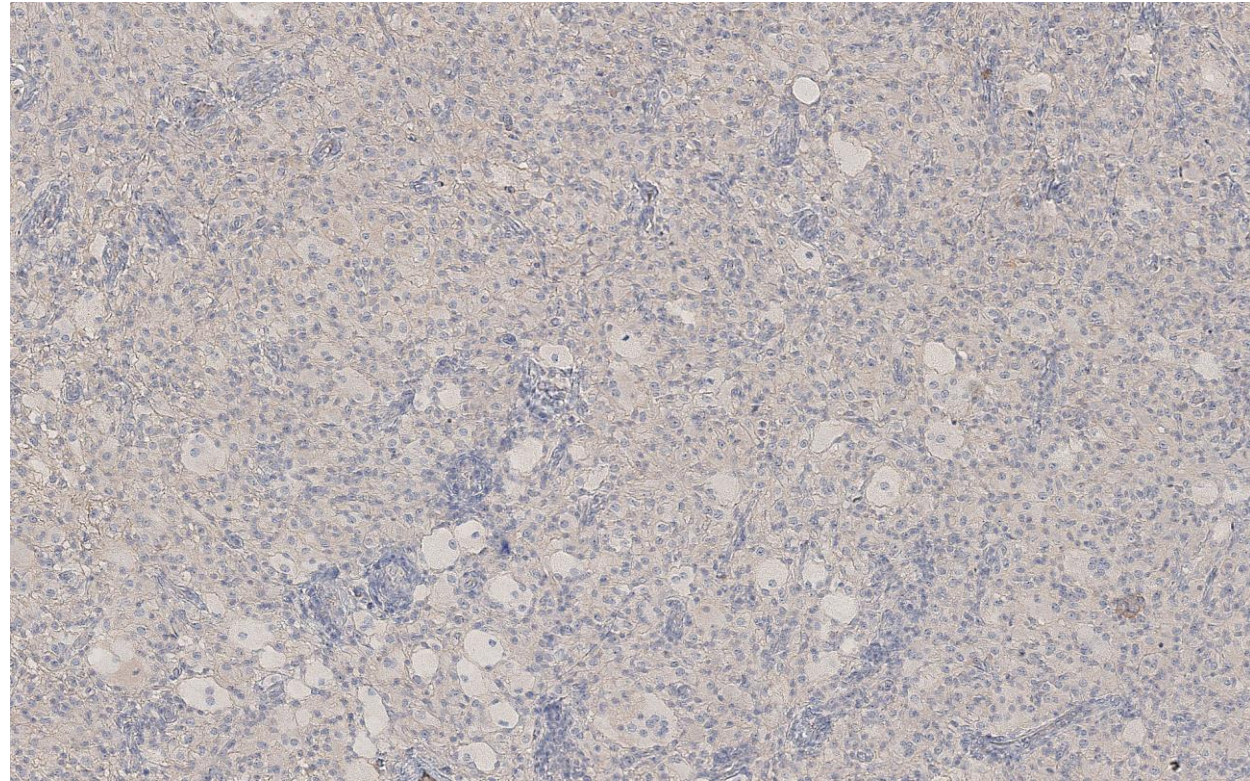
CD68 (PG-M1) 100X



CD14 100X



S100 200X



CD1a 200X

Diagnosi?

Diagnosi differenziale istiocitosi SNC

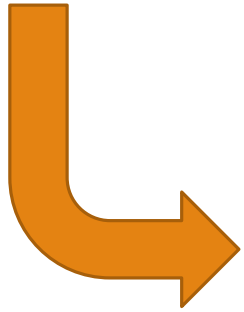
TABLE. Differential Diagnosis of Erdheim–Chester Disease with Langerhans Cell Histiocytosis and Rosai–Dorfman Disease

	Erdheim–Chester Disease	Langerhans Cell Histiocytosis	Rosai–Dorfman Disease
Locations in CNS	Cerebellum, pons, dura, brain parenchyma	Cerebellum, pons, dura, brain parenchyma	Dura
Morphology			
Nucleus	Round	Epithelioid, frequent grooves	Often enlarged with mild atypia
Nucleolus	Small	Variable	Variable. May be prominent
Cytoplasm	Abundant, foamy, or lipid-laden	Moderate, pink	Abundant, dense pale or pink, frequent emperipolesis
Mitosis/necrosis	None	May be present	None
Other features	Touton cells	Occasional osteoclast-like giant cells	
Background	Fibrosis, plasma cells	Abundant eosinophils	Fibrosis and plasma cells
Immunophenotype			
CD1a	–	+	–
CD68	+	+	+
CD207 (langerin)	–	+	–
Factor XIIIa	+	–	–
S100	–	+	–
VE1	+*	+*	–
<i>BRAF</i> V600E	55%–60%	~64%	–
Other Mutations	<i>MAP2K1</i> <i>PIK3CA</i> 10.9% <i>NRAS</i> 3.7%	<i>MAP2K1</i> 12%–27.5% (mutually exclusive to <i>BRAF</i> V600E)	<i>KRAS</i> 19% <i>MAP2K1</i> 14% (mutually exclusive)

*Positive in the cases with *BRAF* V600E mutation.

Diagnosi: Localizzazione di **malattia di Erdheim-Chester**

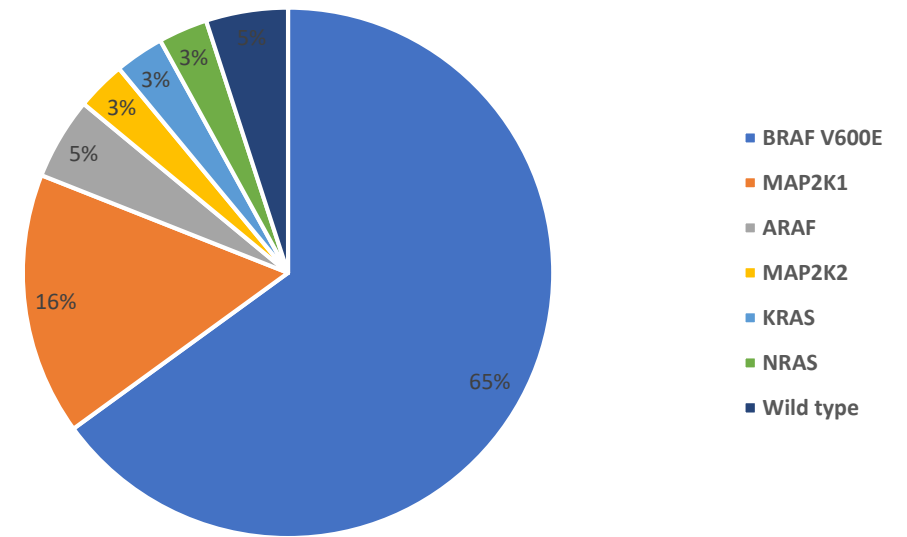
Sequenziamento *BRAF*: presenza di mutazione V600E
(*BRAF* c.1799T>A)



- Staging sistemico: negativo
- Iniziato trattamento con IFN- α
- Quadro clinico e radiologico stabile a 12 mesi dal reintervento

Malattia di Erdheim-Chester

- Diamond EL et al. Consensus guidelines for the diagnosis and clinical management of Erdheim-Chester disease. *Blood* 2014
- Zenggang P et al. CNS Erdheim–Chester Disease: A Challenge to Diagnose. *J Neuropathol Exp Neurol* 2017
- Oneal PA et al. FDA Approval Summary: Vemurafenib for the Treatment of Patients with Erdheim-Chester Disease with the BRAFV600 Mutation. *Oncologist* 2018
- Ozkaya N et al. The histopathology of Erdheim–Chester disease: a comprehensive review of a molecularly characterized cohort. *Mod Pathol* 2018



Ozkaya N et al., *Modern Pathology* 2018