

# Case #6

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# Clinical history

- 8-year-old female patient
- Autism, developmental delay, 15q11.2 deletion syndrome
- No headaches, nausea, vomiting or change in vision.

Post-contrast T1

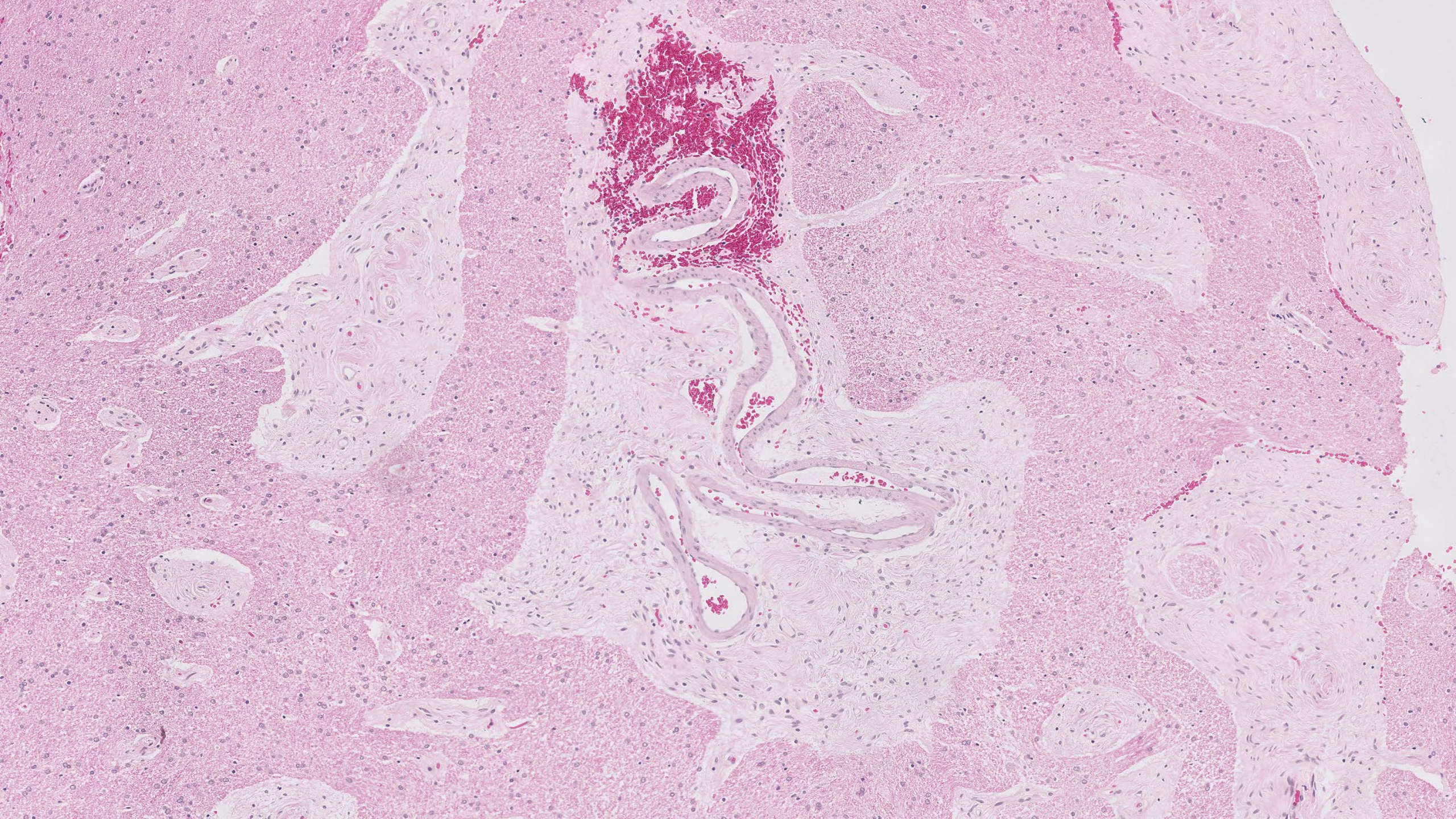


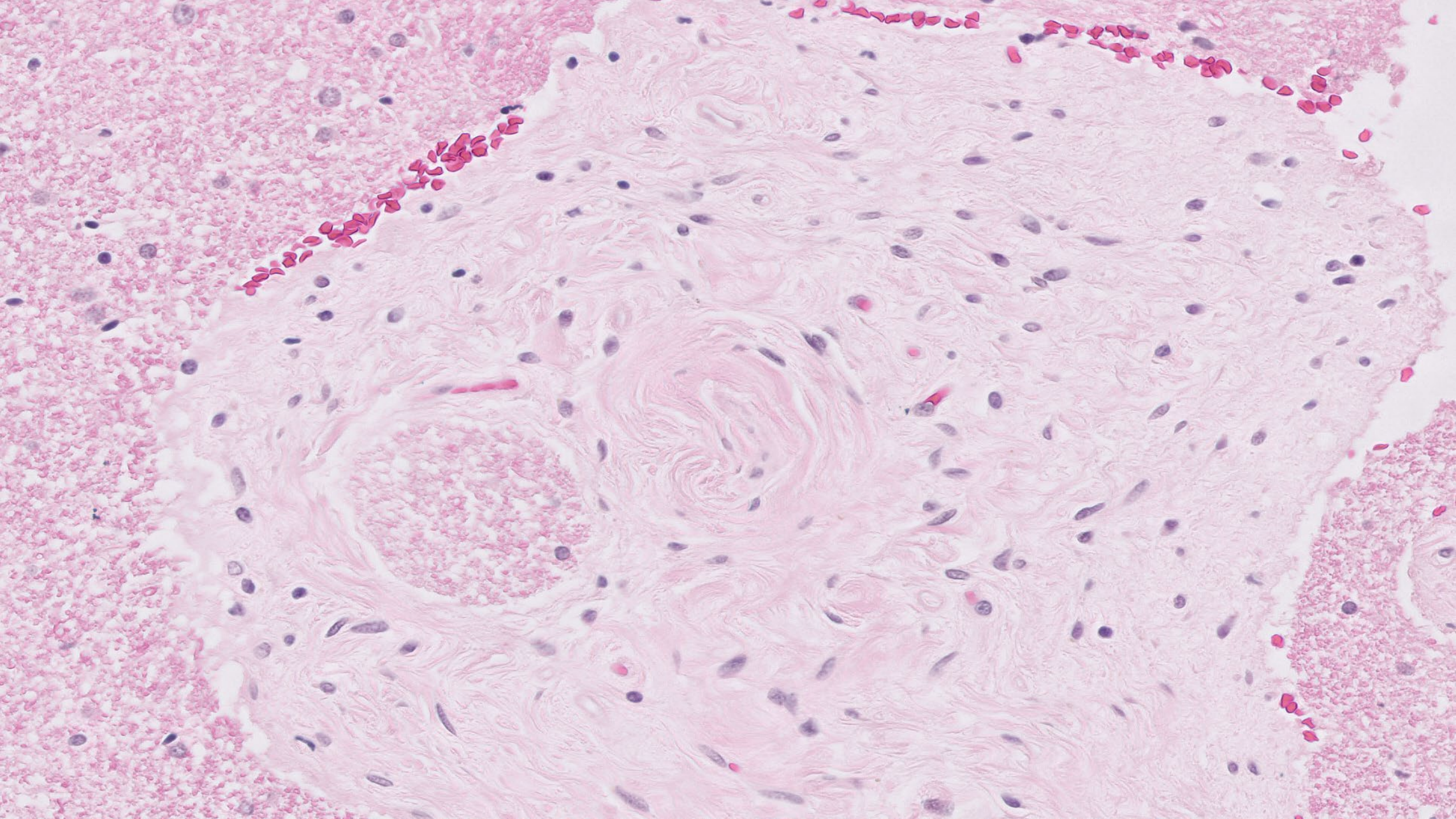
T2

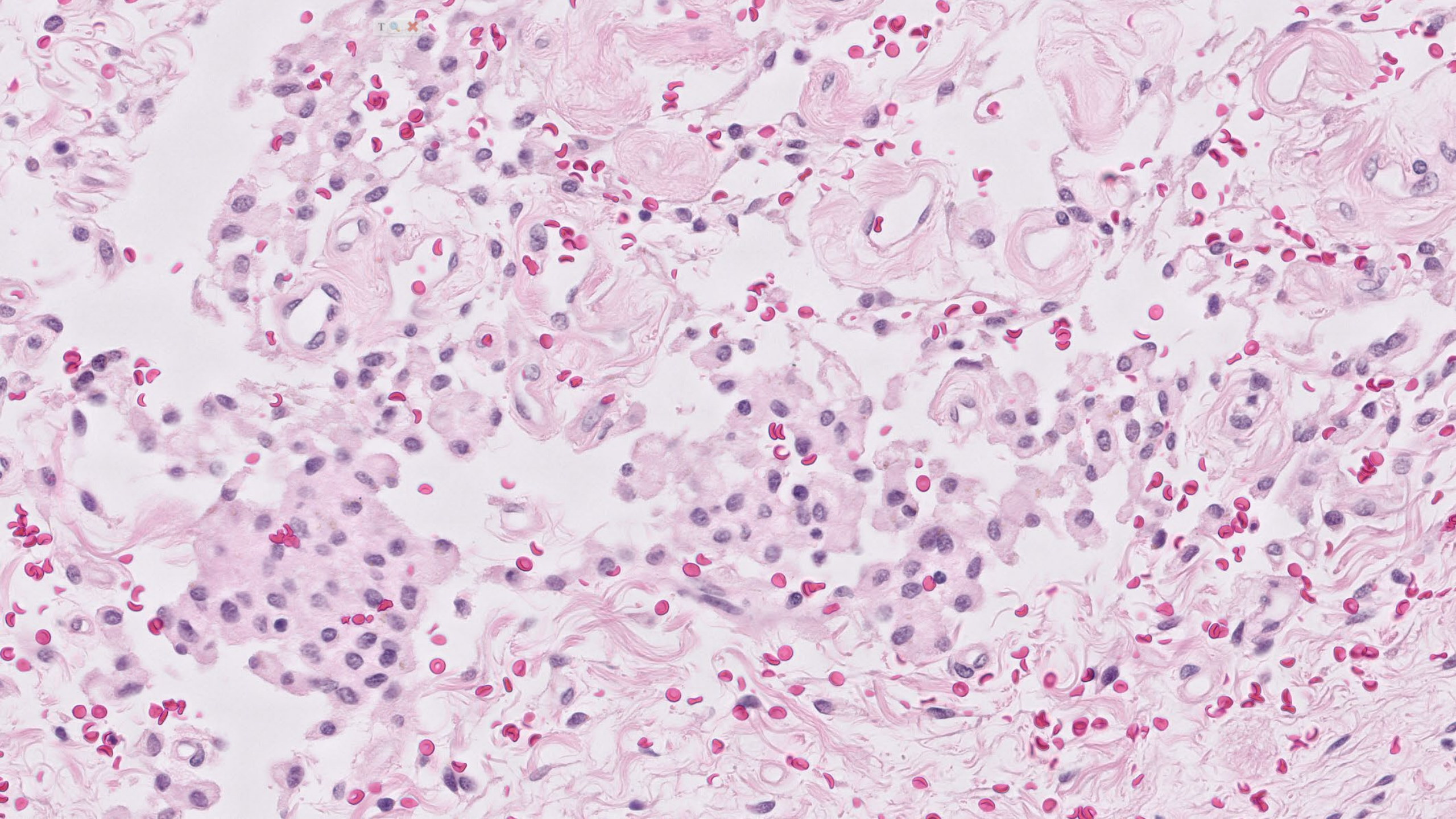


FLAIR

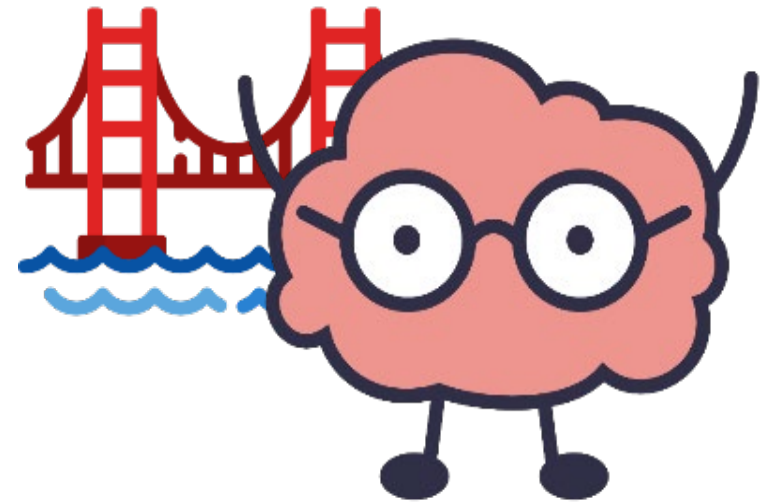




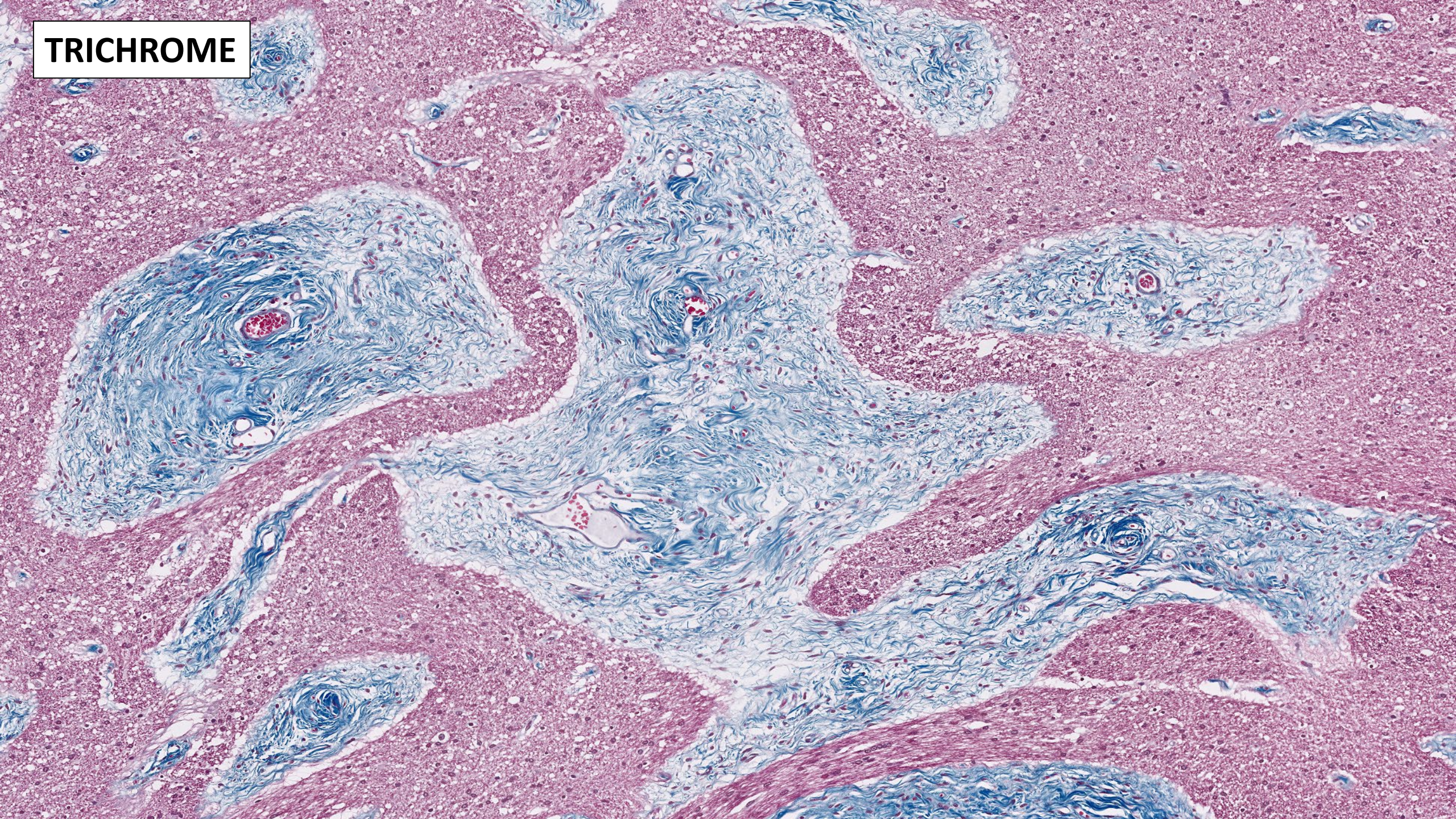




# Diagnosis?

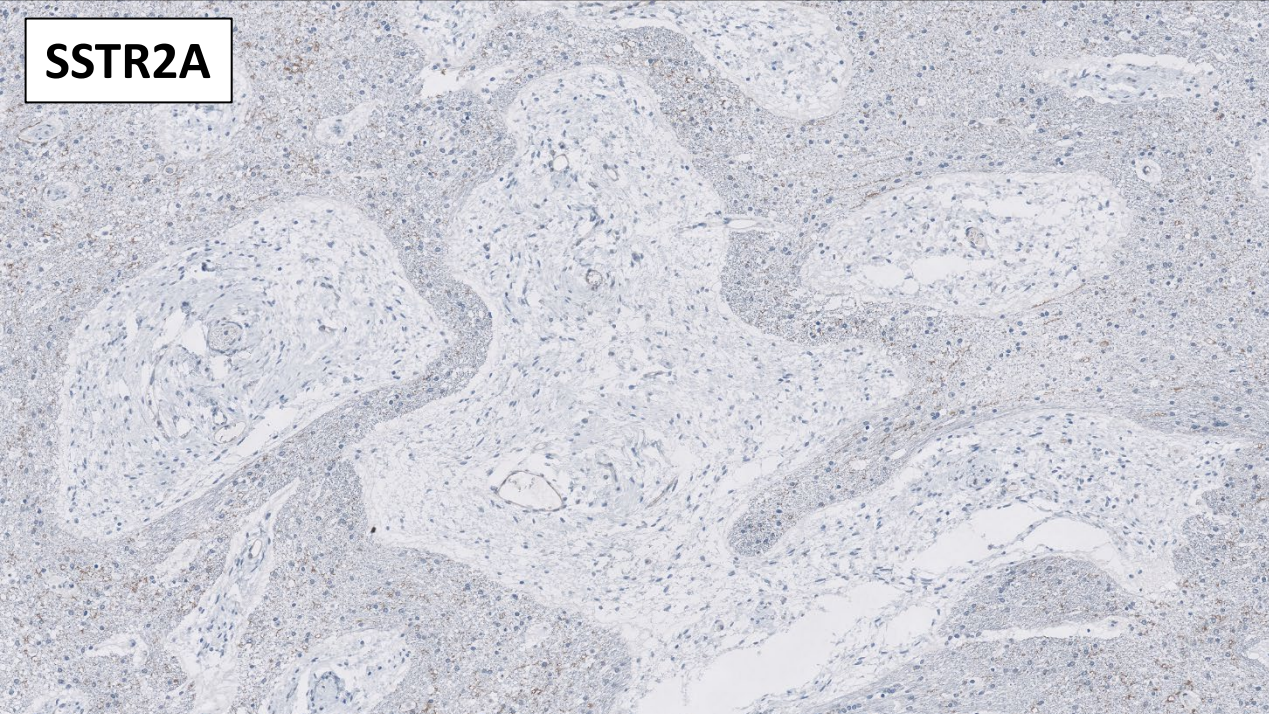


**TRICHROME**

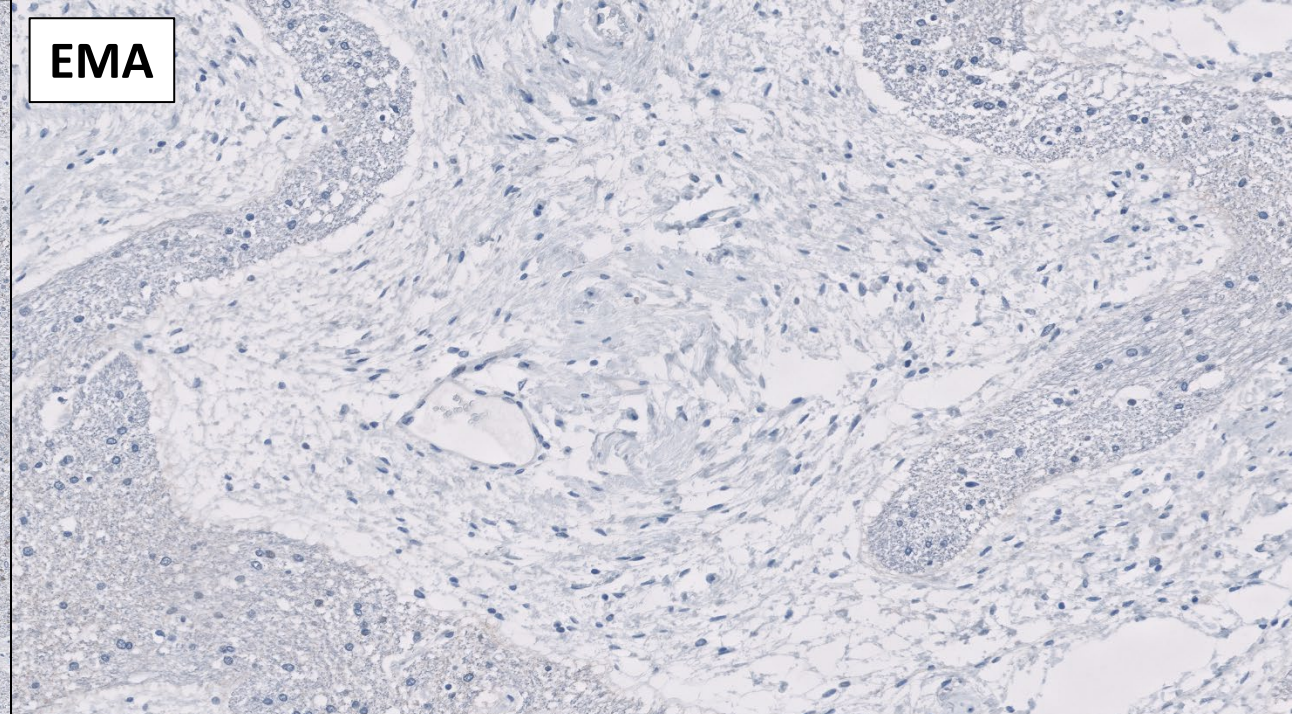




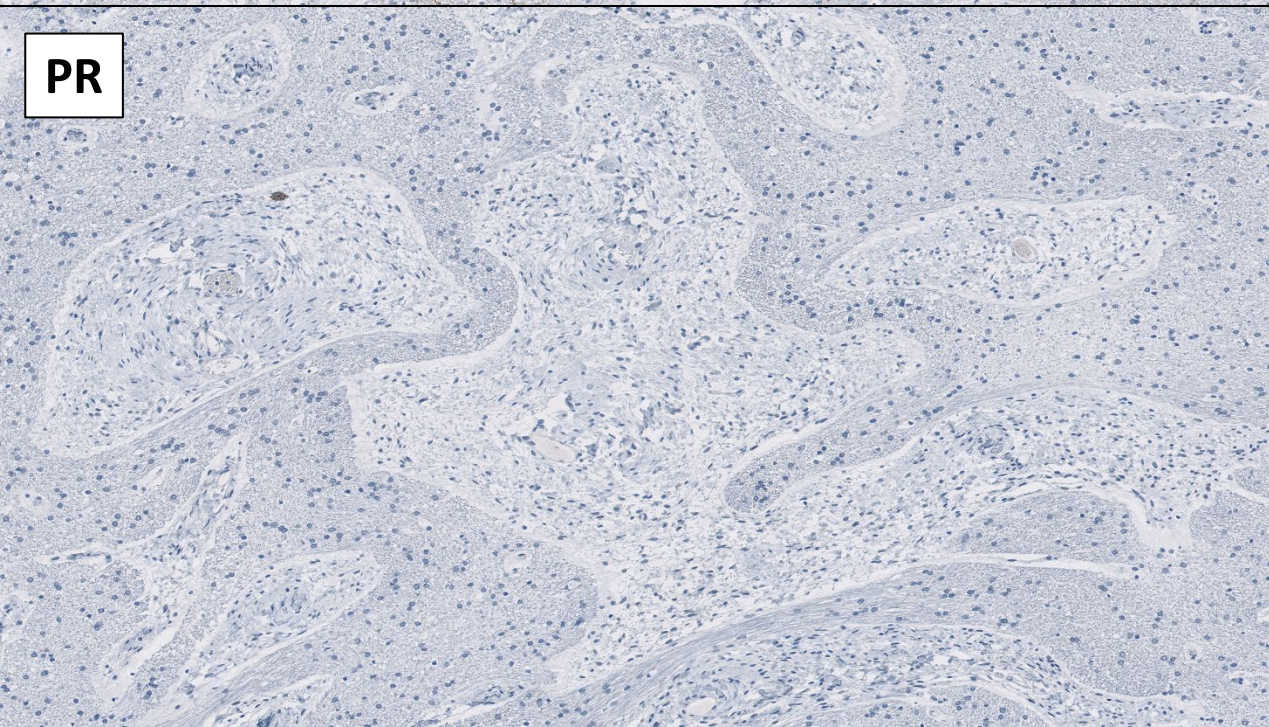
**SSTR2A**



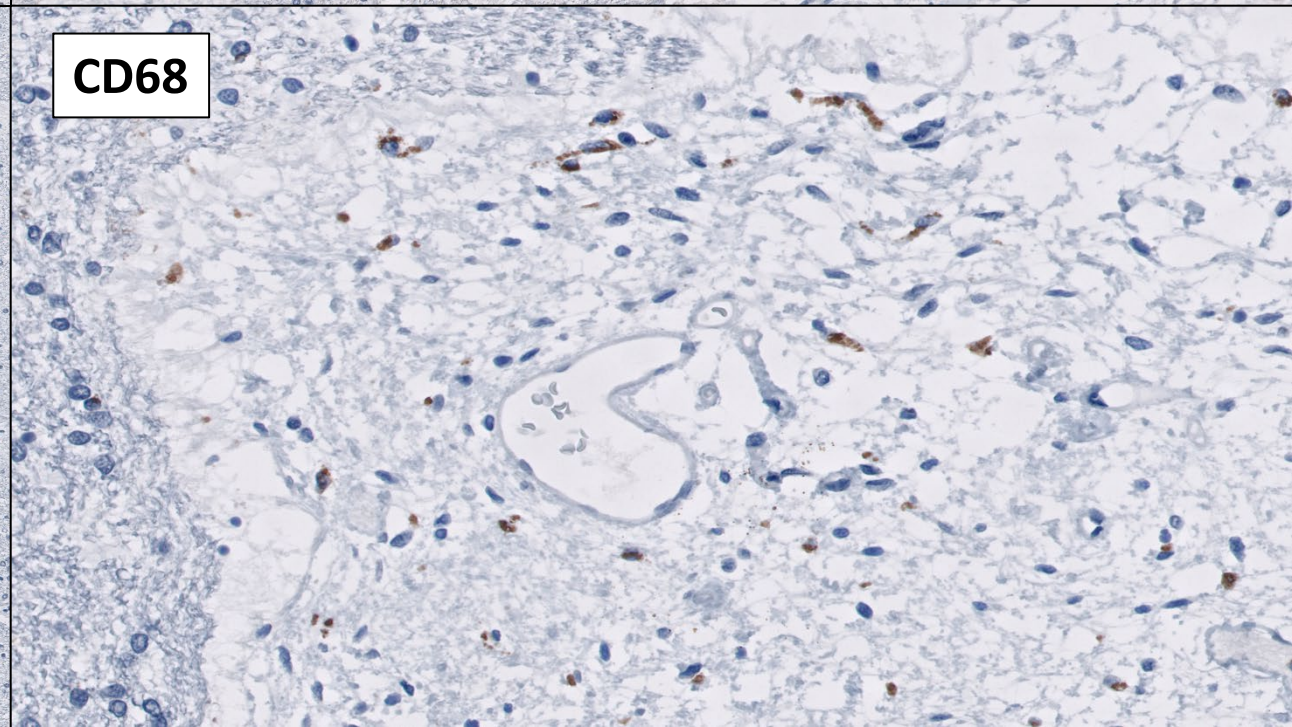
**EMA**



**PR**



**CD68**



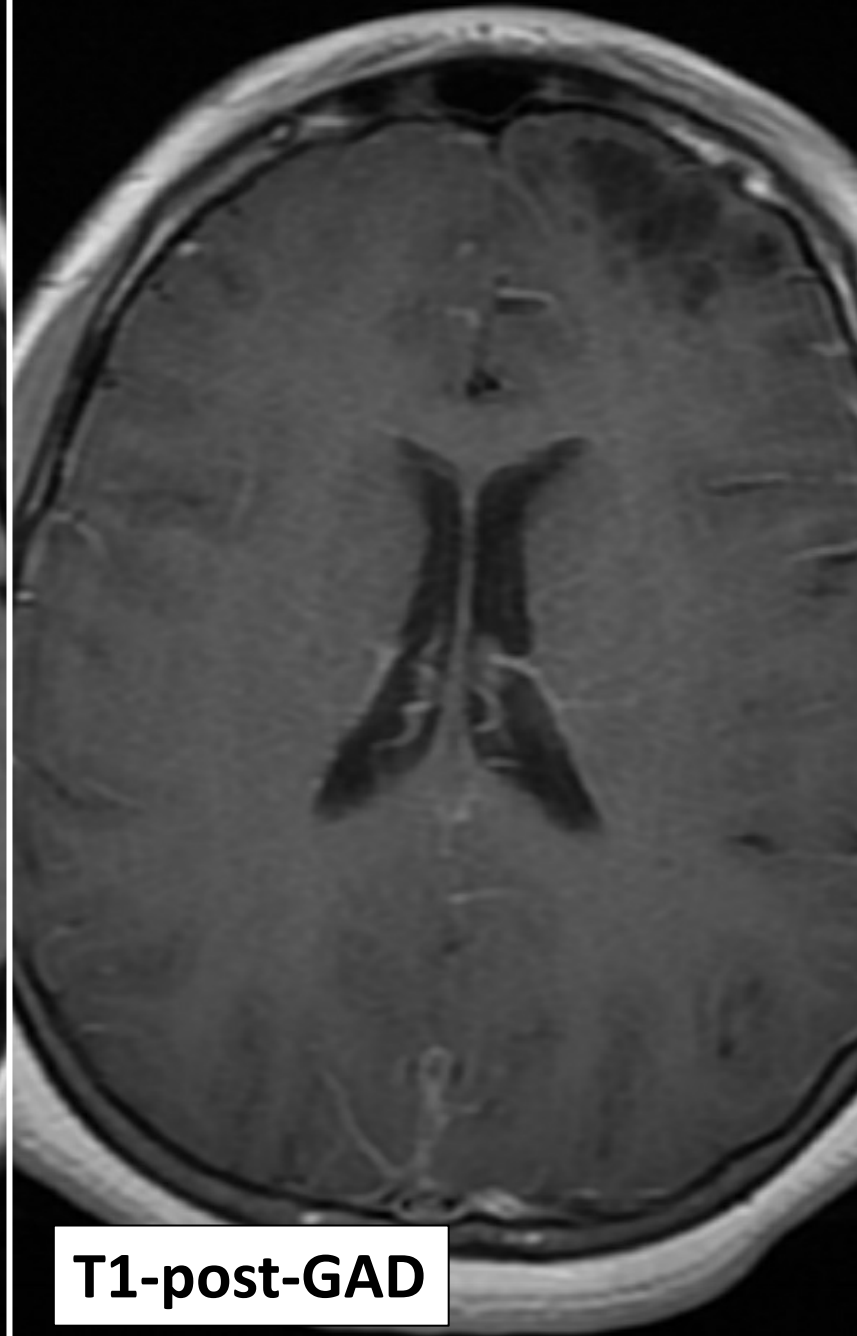
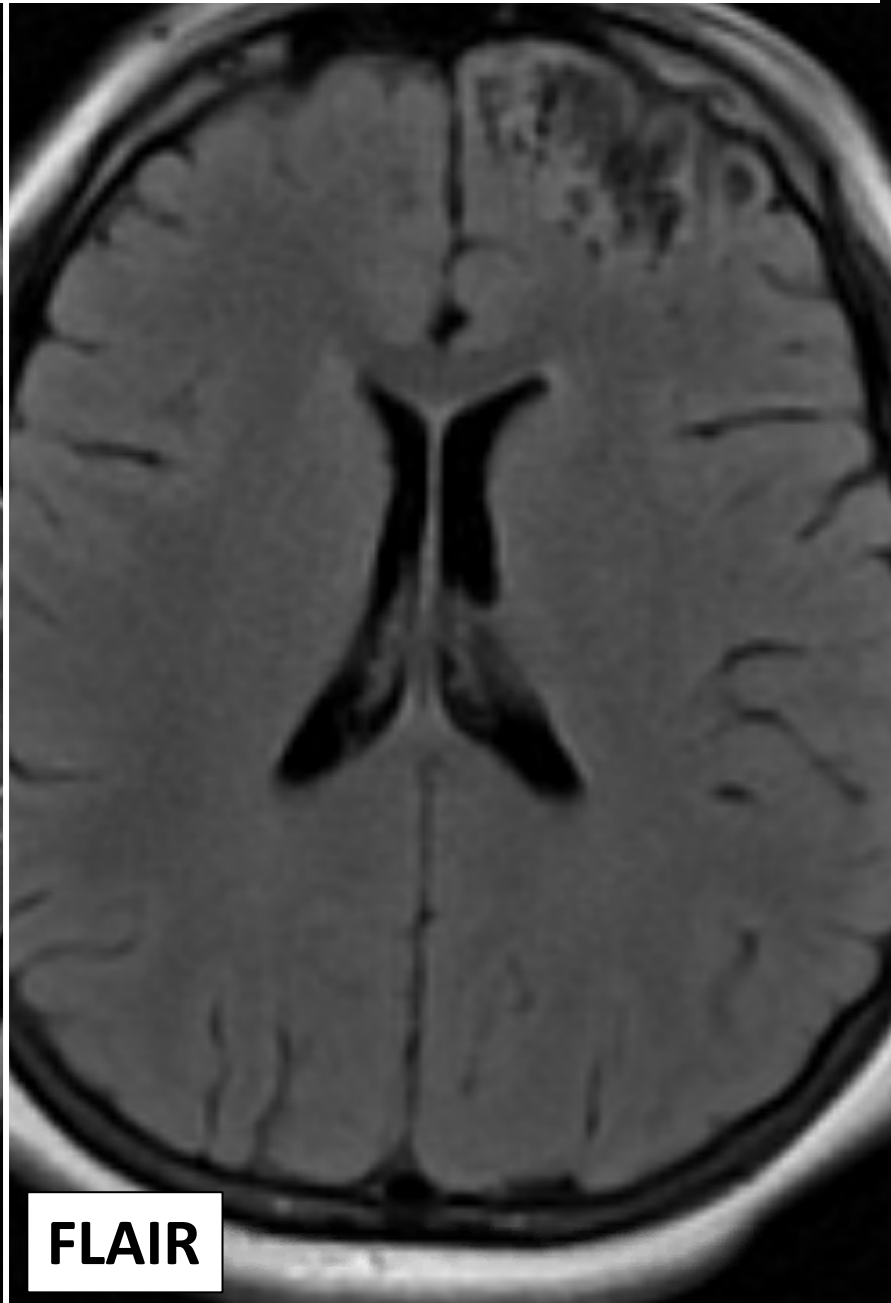
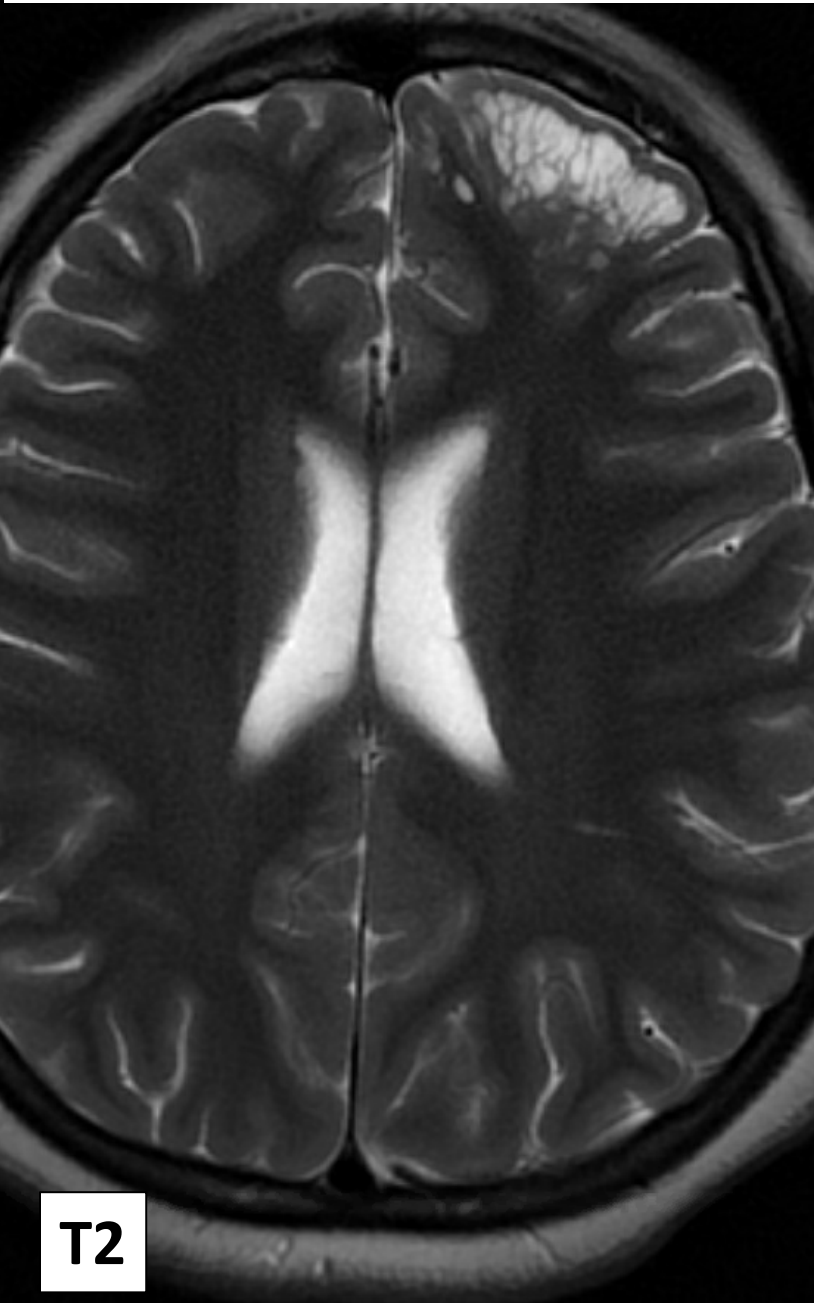
# UCSF500 NGS and Diagnosis

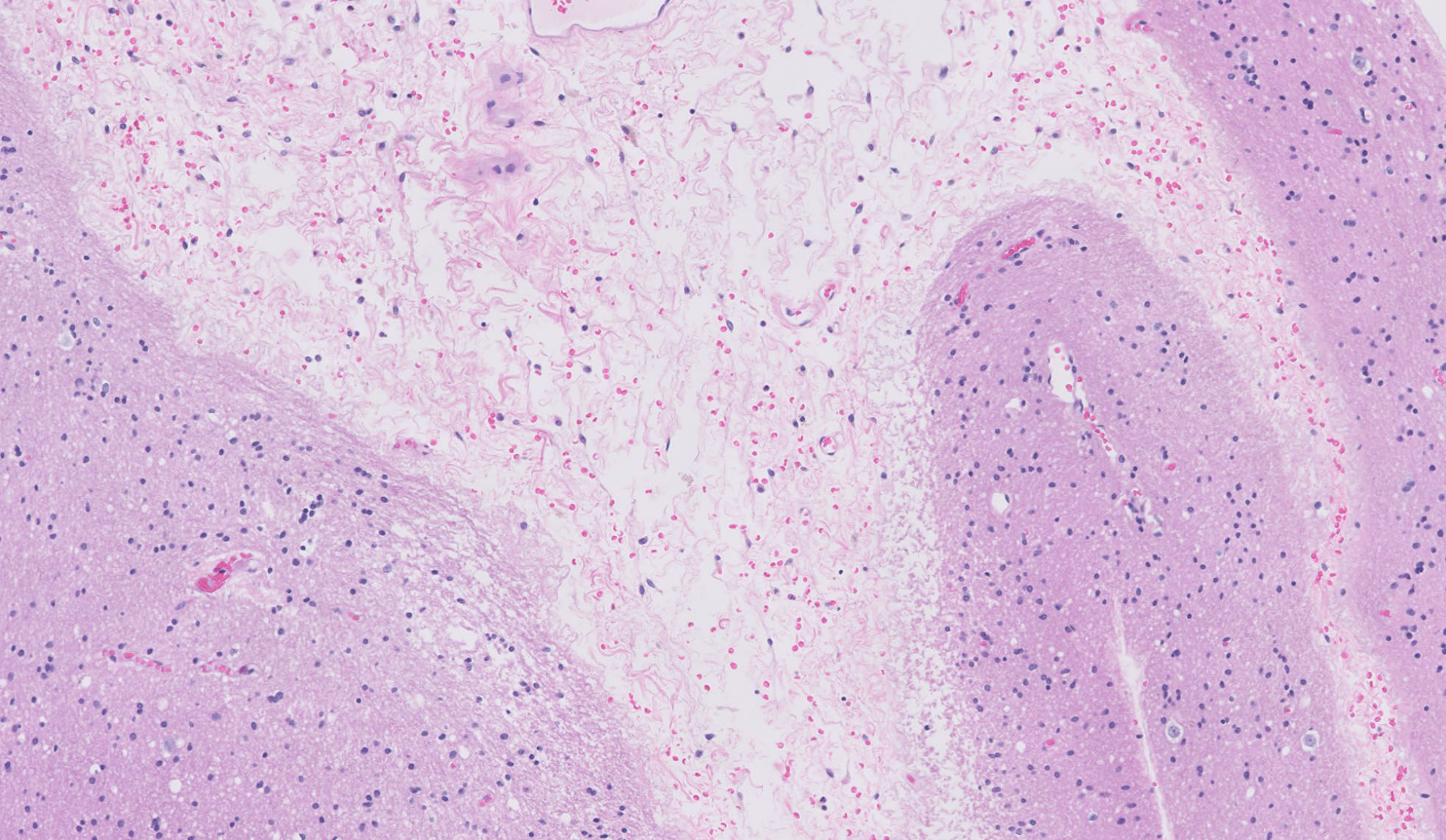
PATHOGENIC AND LIKELY PATHOGENIC ALTERATIONS				
VARIANT	TRANSCRIPT ID	CLASSIFICATION	READS	MUTANT ALLELE FREQUENCY
PDGFRA p.D842del	NM_006206.4	Likely Pathogenic	608	12%

Sequencing of this benign perivascular proliferation demonstrates a single likely pathogenic in-frame deletion in platelet-derived growth factor receptor, alpha (PDGFRA). This deletion occurs within the kinase domain of the protein and missense mutations at the same position (p. D842V) are known to be activating and recurrent in certain tumors such as gastrointestinal stromal tumors (Ref.1-3). In-frame deletions at this position are not common but have been observed previously (Ref.4), thus we have classified this variant as likely pathogenic.

**Diagnosis: Perivascular spindle cell proliferation with a likely pathogenic in-frame PDGFRA deletion.**

Couple months later...A 13-year-old female patient





## Immunohistochemistry

- SSTR2A: Patchy
- EMA: Patchy
- PR: Negative.
- CD34: Blood vessels.
- Ki-67: Rare cells.

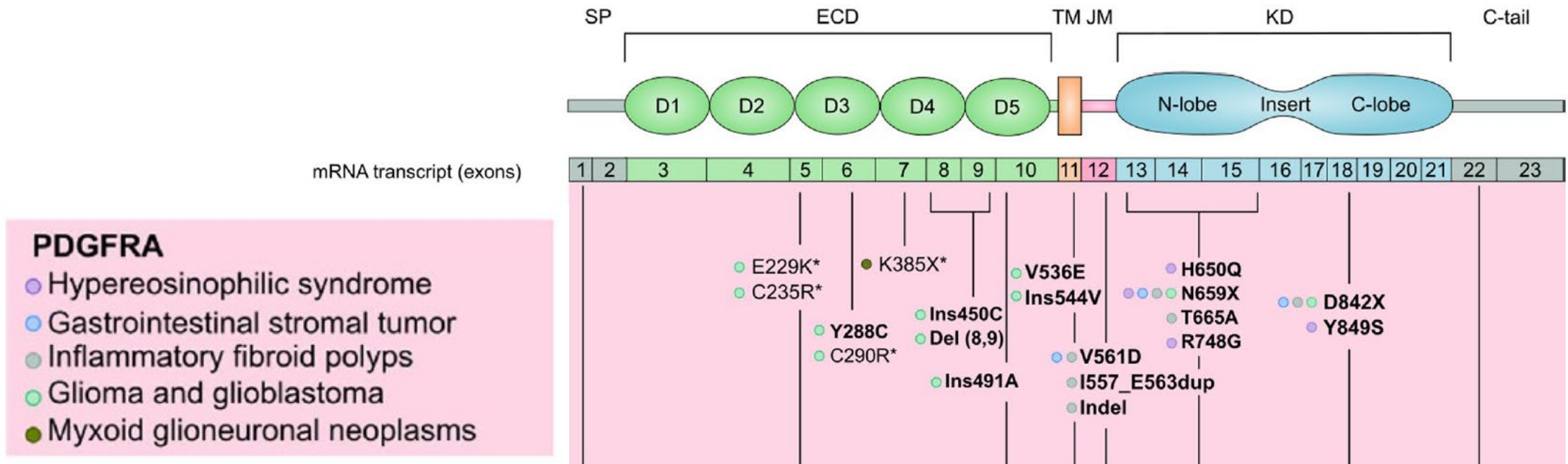
### **PATHOGENIC AND LIKELY PATHOGENIC ALTERATIONS**

VARIANT	TRANSCRIPT ID	CLASSIFICATION	READS	MUTANT ALLELE FREQUENCY
PDGFRA p.D842V	NM_006206.4	Pathogenic	783	2%

# Differential Diagnoses

- Meningioangiomatosis
- Giant tumefactive perivascular spaces
- Gastrointestinal stromal tumor (GIST)
- Myopericytoma/myofibroma

# *PDGFRA* is expressed in mesenchymal cells and its alterations were identified in various disorders

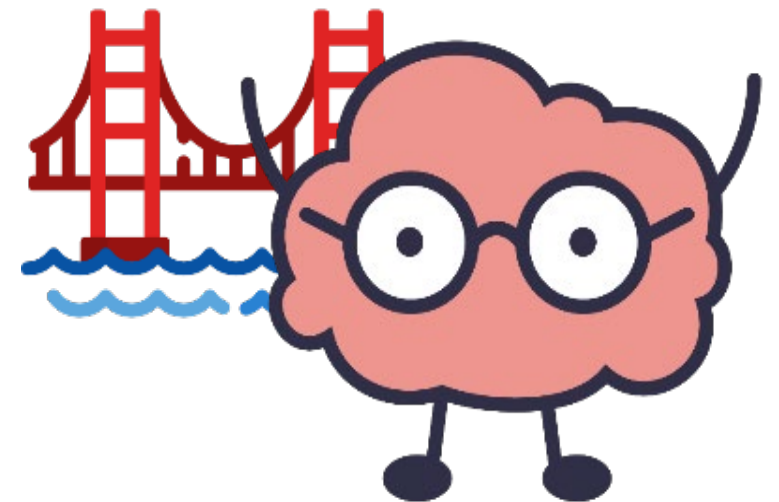


- Glioblastoma, IDH-wildtype, CNS WHO grade 4
- Astrocytoma, IDH-mutant
- Diffuse pediatric type high-grade glioma
- Low-grade glioneuronal tumors such as myxoid glioneuronal tumors
- **Could this be an expansion of this specific progenitor pool?**
- **Is this entity potentially targetable by an RTK-inhibitor?**

Guérit E, et al. Cell Mol Life Sci. 2021  
 Korshunov A et al. Acta Neuropathol. 2017 PMID: 28401334  
 Korshunov A et al. Acta Neuropathol. 2016 PMID: 26482474.  
 Brennan CW et al. Cell. 2013 PMID: 24120142  
 Iwayama T et al. Genes Dev. 2015 PMID: 26019175

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- UCSF Neuropathology Division



# References

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