Dysplastic cerebellar gangliocytoma

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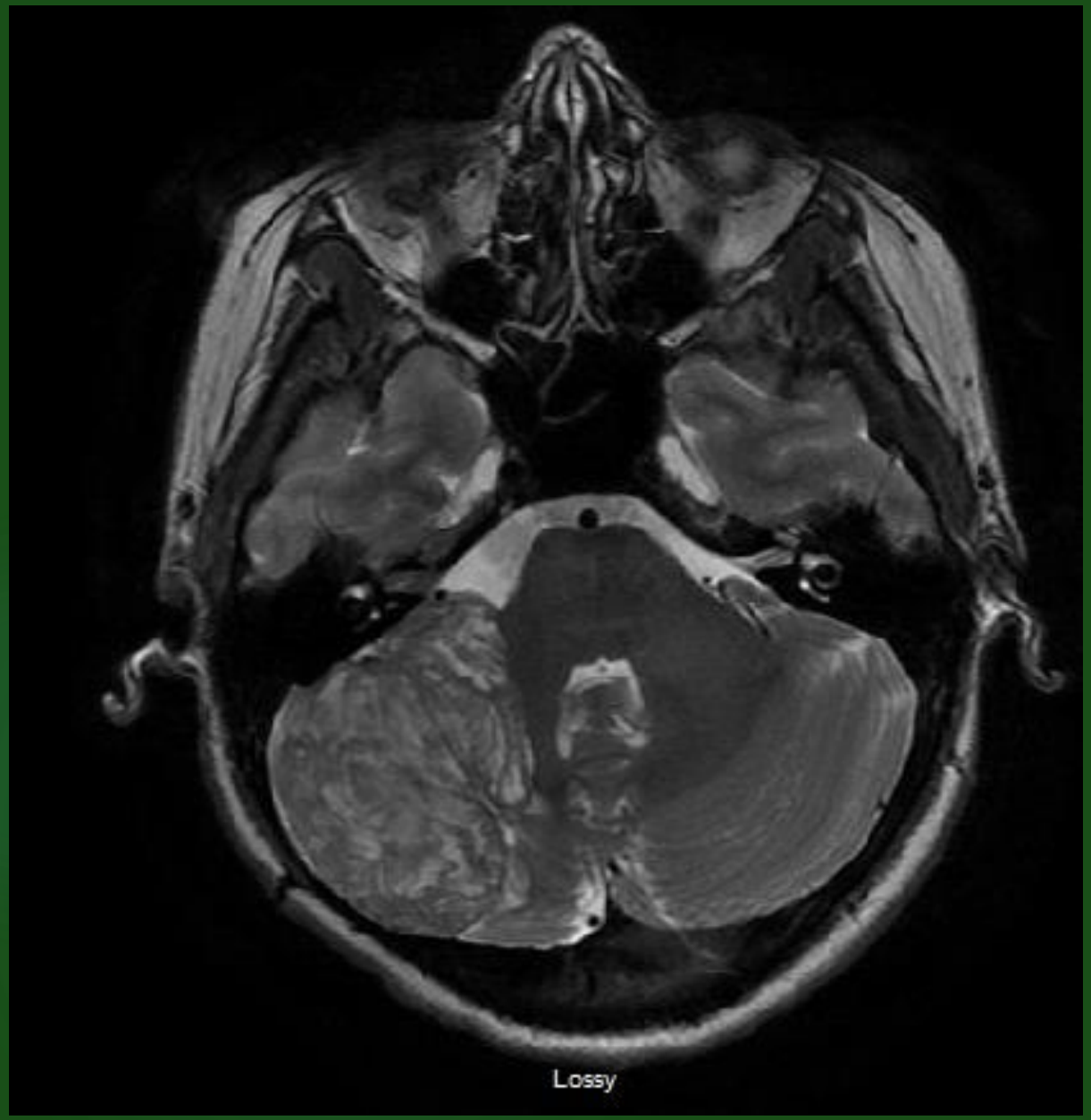
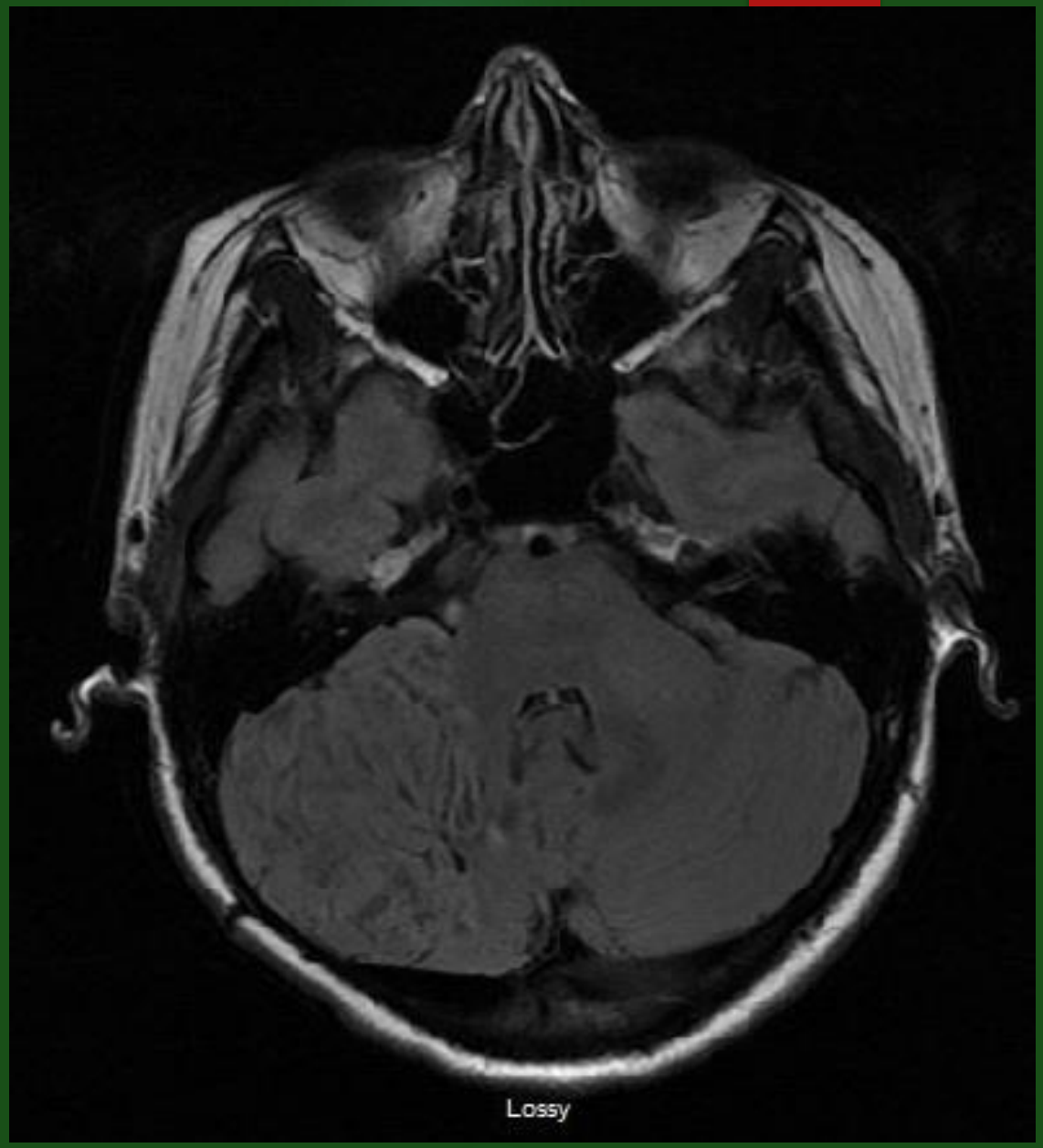
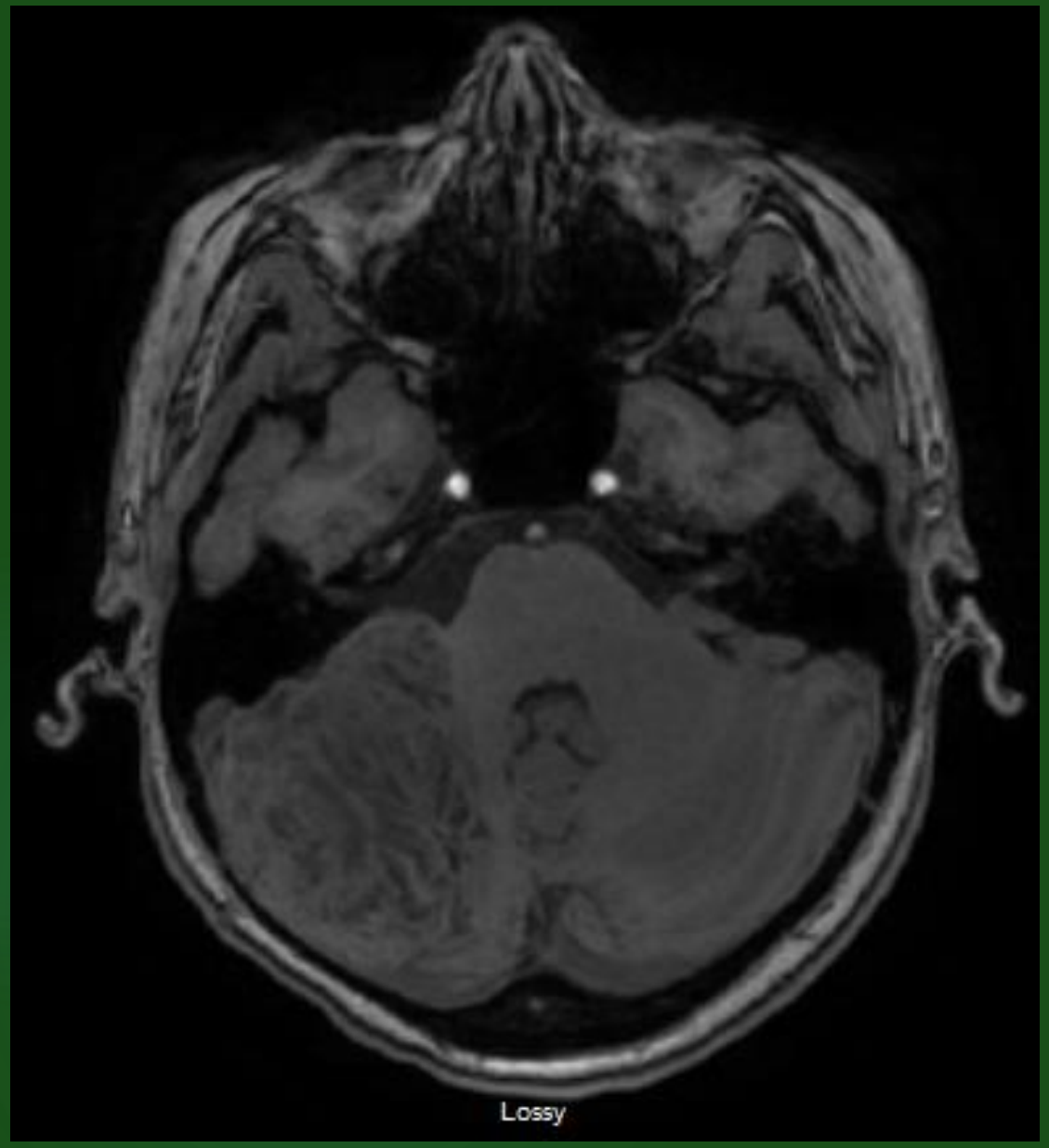
Clinical Presentation

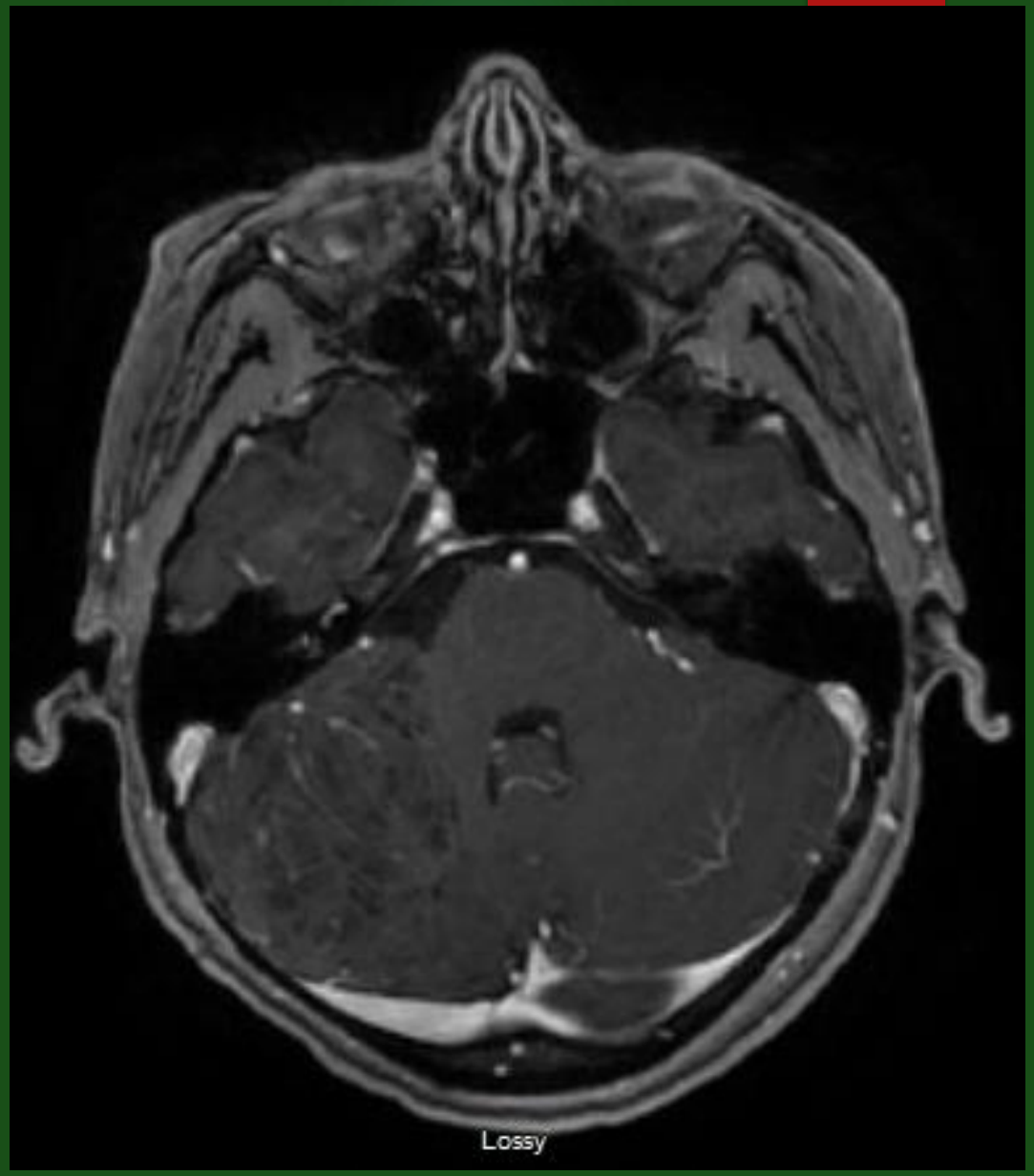
- 27-year-old female presented with history of worsening headaches located at the left occipital region associated with left-sided neck pain and occasional nausea and vomiting
- The patient also had witnessed seizure-like activity where she was found crouched on the floor hyperventilating, followed by tonic stiffening of both arms. However, during this entire episode, she was able to communicate and walk.
- She went to a walk-in clinic and was diagnosed with a panic attack with advice to follow up with her PCP.
- Her PCP referred her to a neurologist who obtained an MRI of the brain and EEG. MRI brain showed a right cerebellar mass and obstructive hydrocephalus. EEG showed few spikes and sharp waves in the left temporal region, and she was referred to neurosurgery for mass removal.



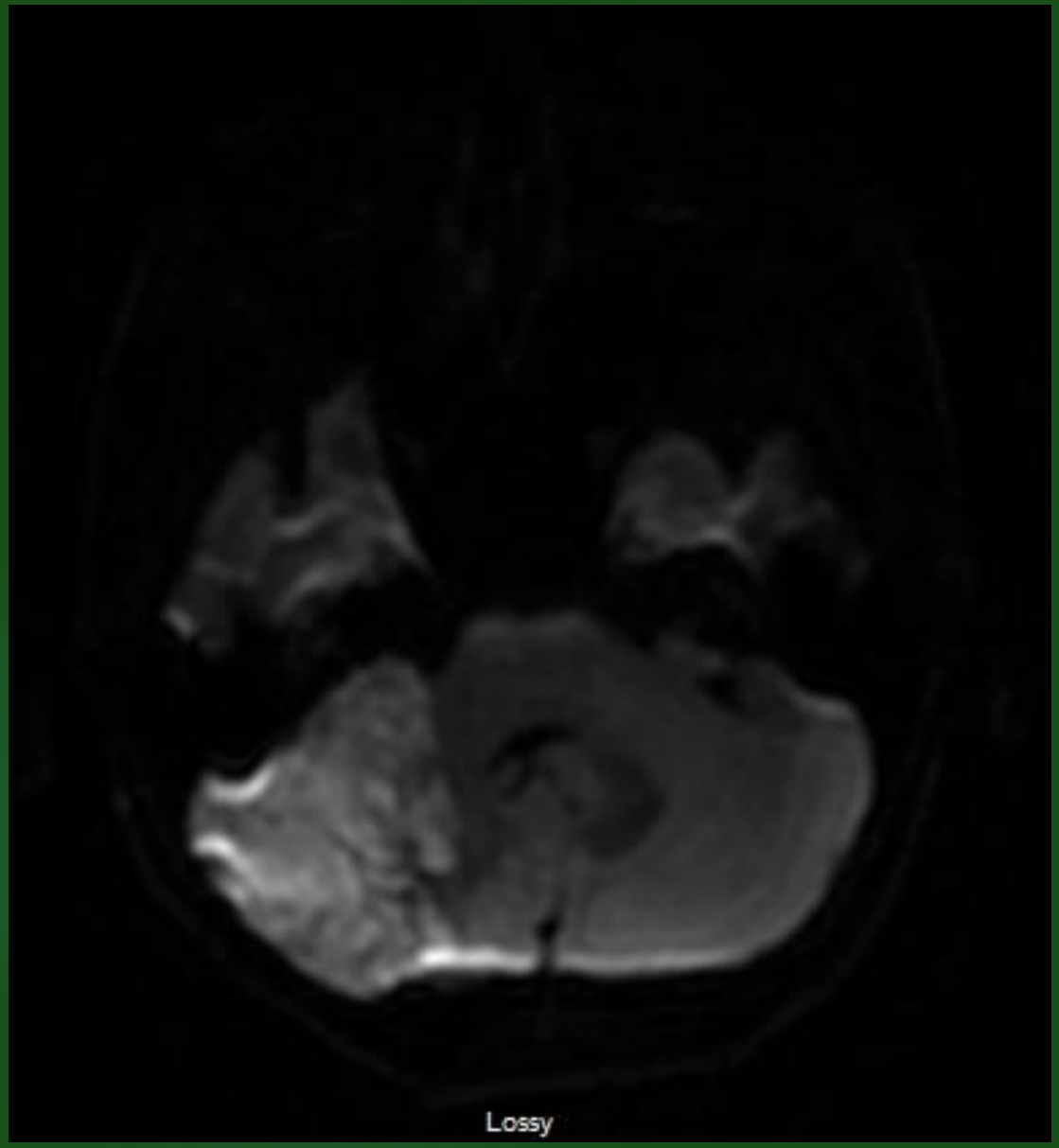


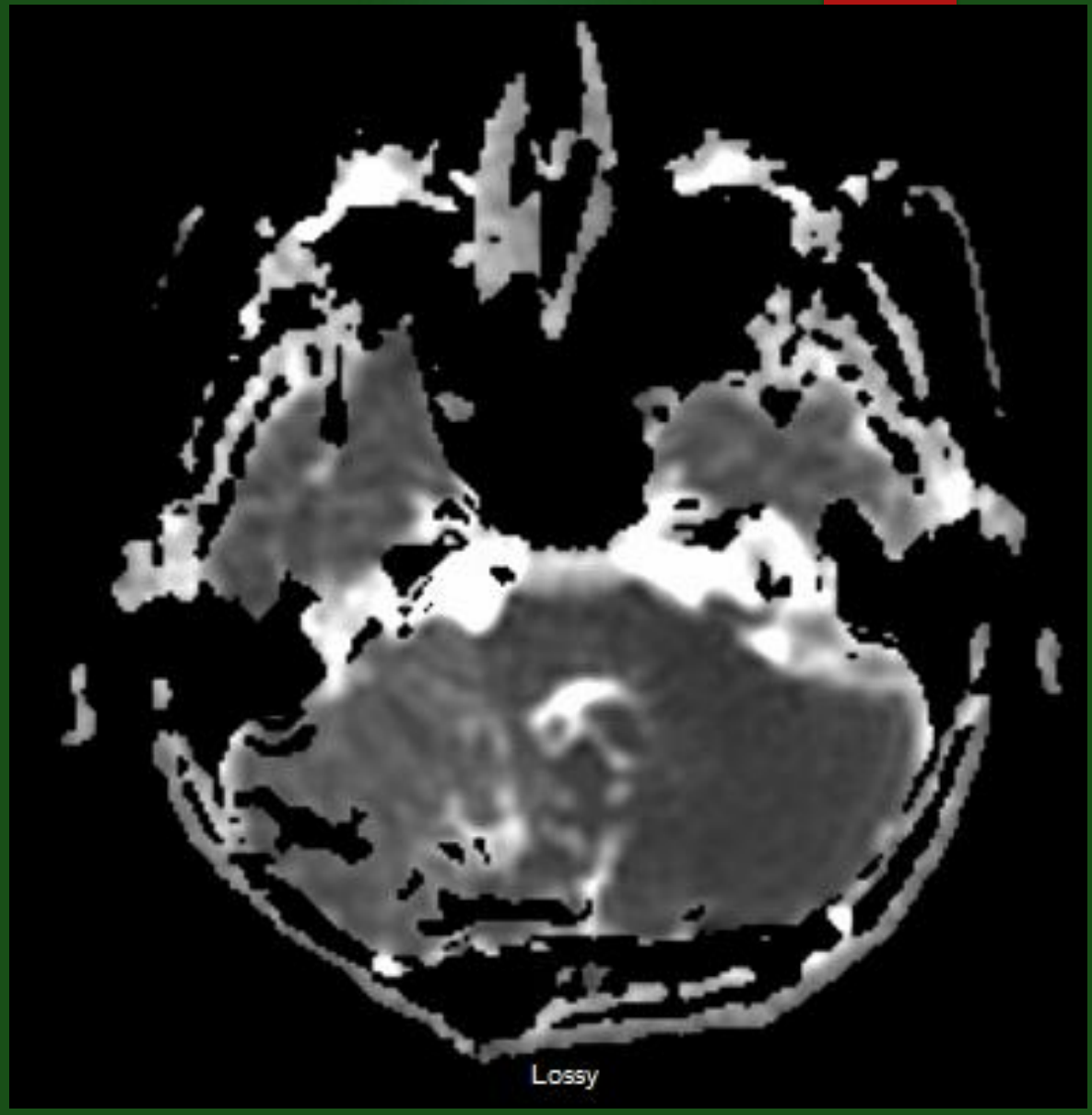
Large T2 hyperintense and T1 isointense right cerebellar mass involving the majority of the right cerebellar hemisphere.





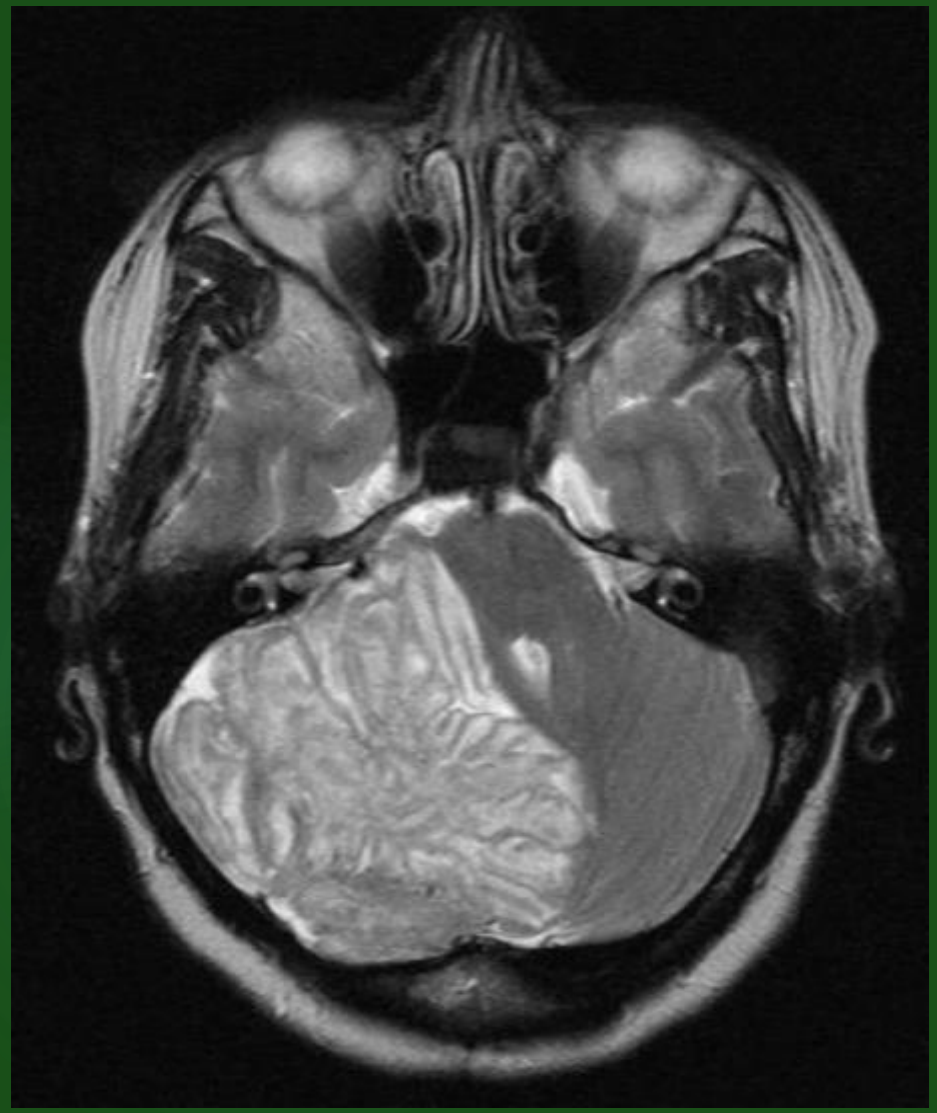
There are vessels traversing the lesion with very subtle areas of enhancement.





DWI images showing hyperintensity due to T2 shine-through

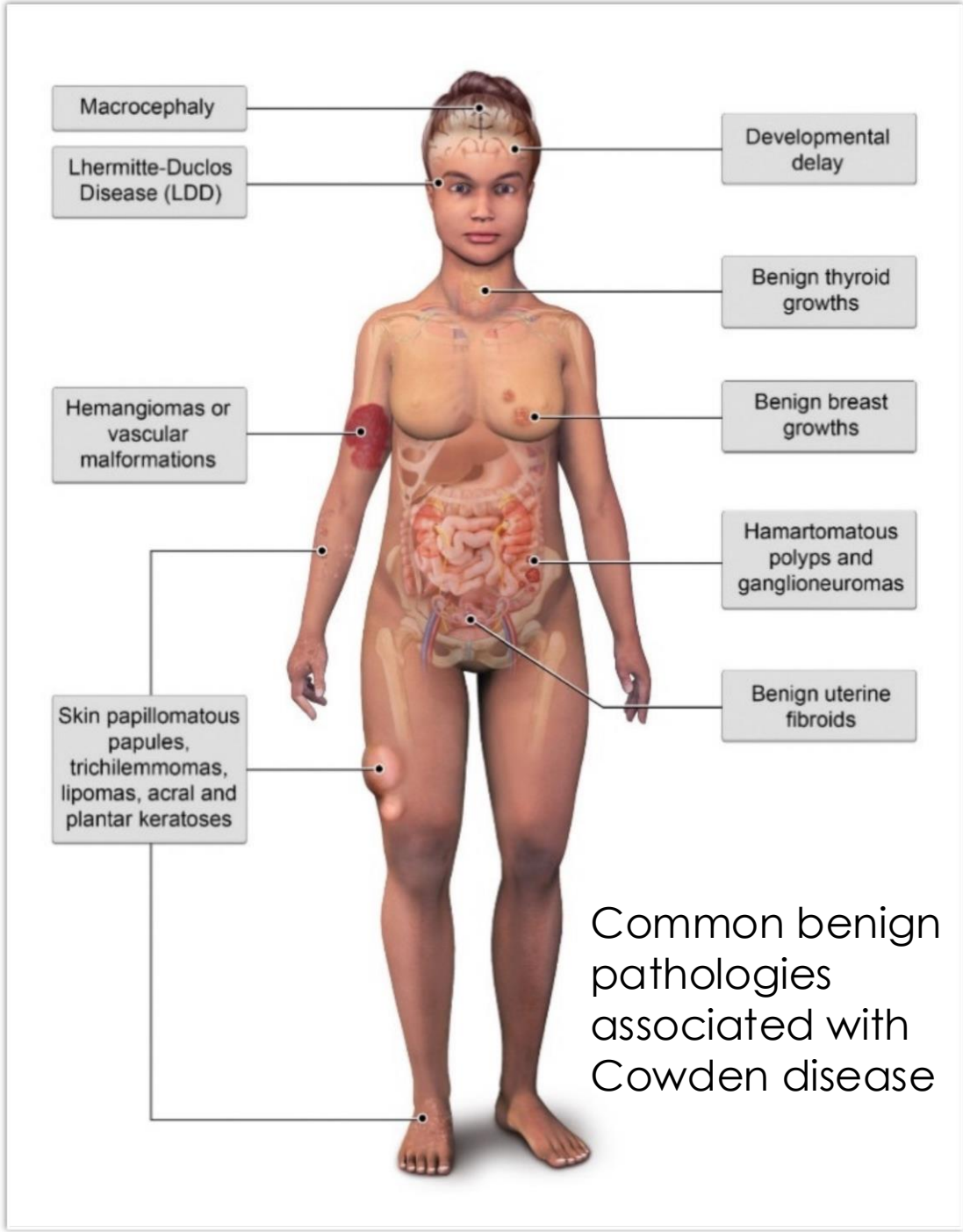
Imaging Features



- Usually unilateral with the right cerebellar hemisphere being more commonly affected
- T2: Hyperintense with a distinct striated pattern of preserved and widened cerebellar folia.
- DWI: The lesion demonstrates signal similar to normal cortex or may show hyperintensity due to T2 shine-through.
- Contrasted images: Enhancement is rare and if present, is usually superficial and possibly due to vascular proliferation.
- MR spectroscopy
  - Elevated lactate
  - Slightly reduced NAA
  - Reduced myo-inositol
  - Reduced choline
  - Reduced Cho/Cr ratio

Background

- Dysplastic cerebellar gangliocytoma, also known as Lhermitte-Duclos syndrome, is a rare cerebellar lesion generally considered to be a dysplastic hamartoma comprised of abnormally enlarged cells predominantly in the granule cell layer with a prevalence of about 1 in 1 million.
- It is a slow growing, space occupying lesion and the most common clinical presentation is headache, nausea, vomiting, papilledema, or visual disturbances.
- 75-80% of cases have a germline inactivation of the PTEN gene and over 1/3 of cases are associated with Cowden disease (multiple hamartoma-neoplasia syndrome).
- Cowden disease is a rare, autosomal dominant, multisystem clinical entity characterized by malignant and hamartomatous lesions developing particularly in the breast, endometrium, and thyroid.



Common benign pathologies associated with Cowden disease

Management

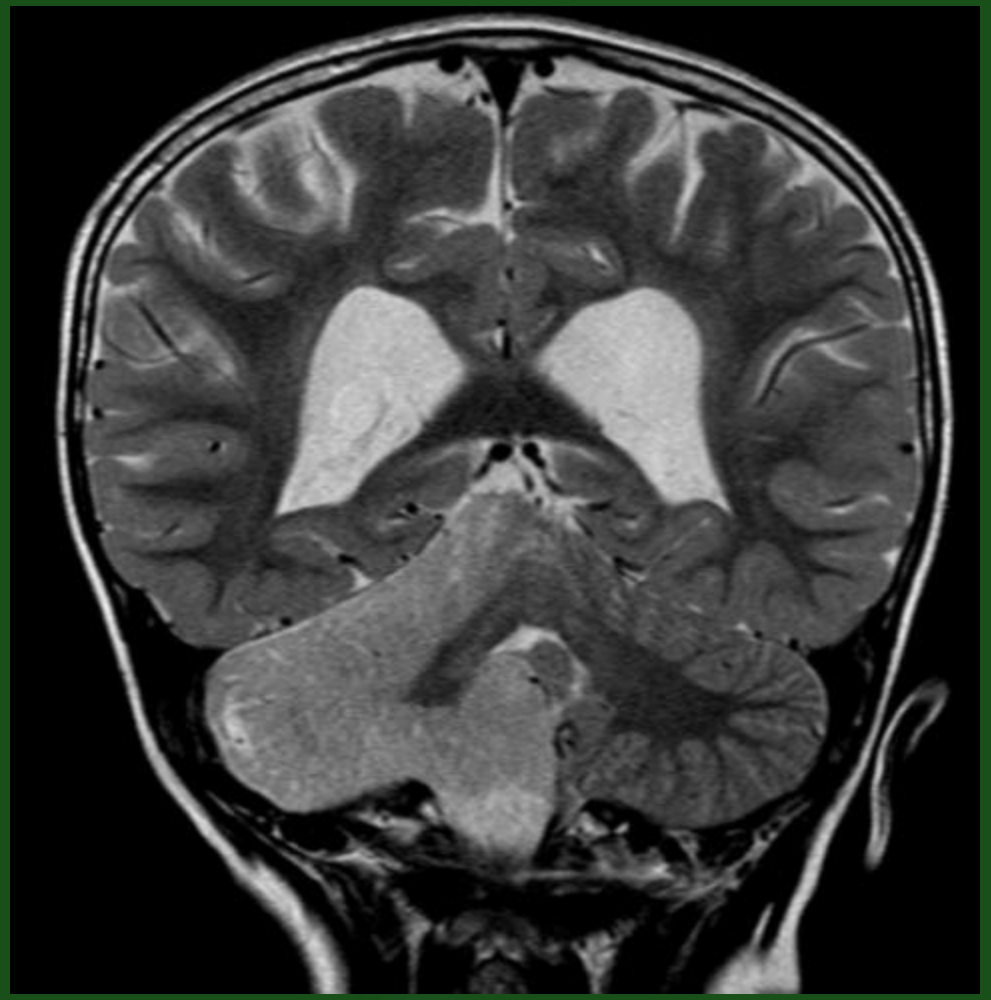
- MRI imaging facilitates better surgical planning as resection margins are difficult to determine due to preservation of gyral pattern.
- For diffuse lesions, alternative options to resection include suboccipital decompression and/or CSF shunting.
- Surgical intervention has resulted in relief of symptoms and increased long-term survival. However, recurrence has been reported at a rate of 8.6%.
- Cowden syndrome carries increased risk of breast cancer, endometrial cancer, and thyroid cancer which is managed with early cancer screening and/or prophylactic surgery.

Outcome

- The patient had subtotal resection of the mass and pathology showed dysplastic gangliocytoma of the cerebellum (Lhermitte- Duclos disease).
- Genetic testing confirmed c.740delT mutation in the PTEN gene consistent with Cowden's syndrome.
- Other features of Cowden disease were present including multinodular goiter. The patient had a total thyroidectomy, prophylactic hysterectomy and bilateral mastectomy.
- The patient has been headache free since the surgery although she has residual ataxic dysarthria, right sided dysmetria, and ataxic gait.

Take Home Points

- Dysplastic cerebellar gangliocytoma, also known as Lhermitte-Duclos syndrome, is a rare, slow growing cerebellar lesion considered to be a dysplastic hamartoma.
- Over 1/3 of cases are associated with Cowden disease which carries an increased risk of thyroid, endometrial, and breast cancer.
- The lesion is typically unilateral with the right cerebellar hemisphere being more commonly affected, and it is T2 hyperintense with a distinct striated pattern of preserved and widened cerebellar folia.
- Surgical intervention has resulted in relief of symptoms and increased long-term survival. However, recurrence has been reported at a rate of 8.6%.



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