FINAL DIAGNOSIS:

Pineal parenchymal tumor of intermediate differentiation.

DISCUSSION:

Tumors of the pineal region are rare, composing less than 1% of intracranial neoplasms1. Pineal parenchymal tumor of intermediate differentiation (PPTID), a neoplasm of the pineal parenchyma with features intermediate between a pineocytoma and pineoblastoma. This neoplasm accounts for 45% of all pineal parenchymal tumors. There is a slight female

preponderance (M: F = 0.8:1) with a median age of 33 years. (range = 3.5-64 years) 2. The patients present with features related to raised intracranial pressure because of aqueduct-neuro-ophthalmic dysfunction (Parinaud syndrome), and brainstem or cerebellar dysfunction 3.

PPTID is characterized by a characteristic small in frame insertions of KBTBD4 4. PPTID are graded from WHO grade 2 to 3; however, the grading system remains ill-defined with different grading systems utilizing mitosis, Ki-67 proliferation index and neurofilament protein as the criterion 2. Low-grade PPTID carries an estimated 5-year overall survival of 74% versus 39% for high-grade PPTID 5.

Due to rarity of these tumors, treatment protocols are not very well defined. Maximal safe surgical resection, followed by assessment and decision to use chemotherapy and radiation is the protocol used in majority of the tumors. This decision is influenced by the degree of resection and presence of spinal or cerebrospinal fluid metastases 6.

PPTID has a wide range of differential diagnoses including central neurocytoma, ependymoma, and oligodendroglioma. In the present case glial neoplasm is excluded as GFAP and OLIG2 were negative. EMA negativity excluded an ependymoma. Furthermore the imaging localization of the mass in the pineal region further helped in the diagnosis. A papillary tumor of the pineal region was also considered, which was excluded based on absence of keratin immunoreactivity. A midline location of the tumor warranted work up for midline gliomas; however. our tumor had a negative H3K27M profile with retention of H3K27Me3. INI1 was also done to rule out unusual pineal tumor. Moreover the typical morphology of a desmoplastic myxoid tumor of the pineal region, SMARCB1 mutant was not observed in the present case. Due to the young age and location, a pineoblastoma was the first clinical possibility; however, the lack of primitive looking areas, nuclear molding, and presence of well defined cytoplasm and cytoplasmic borders, a diagnosis of PPTID was the rendered.