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Pure Pineal Germinoma in a 17-Year-Old Adolescent: A Case Report and Literature Review

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Abstract

We report the case of a 17-year-old adolescent presenting with a large pineal region mass initially interpreted as a ruptured mature teratoma with intraventricular dissemination, complicated by triventricular hydrocephalus. Stereotactic biopsy enabled a definitive diagnosis of a pure germinoma, confirmed by its immunohistochemical profile (SALL4+, PLAP+, CD117+, CK−). The patient received three cycles of BEP chemotherapy (bleomycin, etoposide, cisplatin) followed by panventricular irradiation (24 Gy) with an additional tumor boost (16 Gy), resulting in good treatment tolerance and sustained radiologic stabilization. This case highlights the diagnostic challenges in distinguishing germinoma from teratoma on imaging, underscores the importance of stereotactic biopsy, and illustrates the effectiveness of combined chemoradiotherapy in intracranial germinomas.

Keywords

Intracranial Germinoma, Intracranial Germ Cell Tumor, Pineal Region, BEP Regimen, Panventricular Radiotherapy

1. Introduction

Intracranial germ cell tumors (IGCTs) represent a rare and heterogeneous group of central nervous system neoplasms, sharing clinical and histopathological features with their more common gonadal counterparts [1]. Germinomas, also referred to as pure seminomas, account for approximately 0.2% - 1.7% of all primary intracranial tumors [2]and constitute nearly 60% of intracranial germ cell tumors, predominantly arising in the pineal and suprasellar regions [3]. In nearly 90% of

cases, they occur before the age of 20 years [4] [5], with a marked male predominance and a sex ratio of 2:1 [6]. Germinomas are characterized by high radiosensitivity, marked chemosensitivity, and a propensity for dissemination through the cerebrospinal fluid (CSF) pathways [7].

Radiological diagnosis may occasionally be misleading, as heterogeneous pineal masses can mimic teratomas. Therefore, histopathological and immunohistochemical confirmation remains essential to establish the diagnosis of pure intracranial germinoma and guide appropriate therapeutic decision-making.

We report the case of a 17-year-old adolescent presenting with a pineal mass initially suspected to be a ruptured mature teratoma, ultimately diagnosed as a non-secreting pure intracranial germinoma following stereotactic biopsy, and treated with BEP chemotherapy followed by panventricular radiotherapy, resulting in mass stabilization and favorable clinical evolution.

2. Case Presentation

2.1. Patient

This is a 17-year-old adolescent male with no significant past medical history. Surgically, he had undergone placement of a ventriculoperitoneal shunt (VPS) on November 26, 2021, for obstructive hydrocephalus, with a favorable postoperative course. There was no family history of similar conditions or any notable hereditary disorders.

2.2. History of Present Illness

The onset of symptoms dates back to March 2023, when the patient began experiencing progressively worsening headaches associated with episodes of vomiting. Several weeks later, the clinical course was complicated by the occurrence of seizures and a subjective decline in visual acuity, prompting his emergency consultation at our institution in June 2023. It is noteworthy that an earlier episode of obstructive hydrocephalus in 2021 had required placement of a ventriculoperitoneal shunt. At that time, a brain CT scan had already revealed a large mass in the pineal region suggestive of a teratoma ruptured into the ventricular system. The 2021 CT scan also showed a bilateral intraventricular fat–fluid level, indicating longstanding intraventricular dissemination. The CT performed in 2023 confirmed the persistence of this dissemination, now associated with a significant increase in tumour volume, which accounted for the recurrence of intracranial hypertension symptoms. The patient showed good neurological recovery after VPS placement and remained clinically stable until symptom recurrence in 2023. It should be noted that between 2021 and 2023, the patient was lost to follow-up and did not undergo regular clinical or radiological monitoring, returning only upon the recurrence of symptoms. The reappearance of signs of intracranial hypertension, combined with the onset of seizures, led to updated neuroimaging, which demonstrated an increase in the size of the pineal mass as well as worsening hydrocephalus, thereby justifying renewed medical evaluation and management.

2.3. Initial Imaging

Brain CT (19 November 2021):

A large heterogeneous pineal region mass was identified, displaying a triple-component architecture consisting of:

- a solid tissue component,
- a predominant fatty component,
- and a fluid-density area.

The lesion measured 69 × 39 × 47 mm, with intralesional calcifications. Evidence of intraventricular rupture was noted, producing bilateral fat–fluid levels within the ventricular system. The mass caused active triventricular hydrocephalus and cerebellar tonsillar descent.

Conclusion: Findings were highly suggestive of a ruptured mature pineal teratoma.

Brain CT (21 March 2023):

The pineal mass had increased in size to 87 × 78 × 58 mm, consistent with rapid progression, with imaging features remaining compatible with a ruptured mature teratoma.

The main CT examinations performed throughout follow-up, from the initial imaging to the post-treatment assessments, are summarized in Table 1.

Table 1. Radiological evolution of the pineal mass on CT scans.

Date	Modality	Tumor size	Main characteristics	Hydrocephalus	Interpretation/Comment
19/11/2021	CT	69 × 39 × 47 mm	Heterogeneous, three components (solid tissue, fat, cystic area), intralesional calcifications, bilateral intraventricular fat–fluid level	Triventricular active	Appearance suggestive of ruptured pineal teratoma
21/03/2023	CT	87 × 78 × 58 mm	Large, heterogeneous mass, persistent, calcifications	Increased hydrocephalus	Rapid growth, still compatible with suspected teratoma
25/08/2023	CT	Marked regression	Small residual lesion in the third ventricle	VP shunt functioning well	After BEP chemotherapy and before radiotherapy
28/07/2025	CT	67 × 47 × 45 mm	Stable, bilateral subdural collections unchanged	VP shunt in place	Long-term post-treatment follow-up

2.4. Stereotactic Biopsy (3 April 2023)

Initial histopathological assessment:

- Dense polymorphous lymphoid infiltrates,
- No expression of glial markers (GFAP–, Olig2–),
- Heterogeneous expression of CD3 and CD20,
- CD30 negative,
- Differential diagnosis: encephalitis vs. lymphoma.

Additional immunohistochemistry following clinico-radiological correlation:

- Cytokeratin negative,
- CD117+, PLAP+, SALL4+ in scattered characteristic tumor cells,
- No epithelial or mesenchymal components identified.

Final immunohistochemistry conclusion:

Findings were consistent with a germinoma (pure seminomatous intracranial germ cell tumor).

2.5. Staging Work-Up

Thoraco-abdomino-pelvic CT (27 April 2023):

No evidence of metastatic disease.

Serum tumor markers (23 May 2023):

- β-hCG: 2.46 mIU/mL
- AFP: 1.21 ng/mL
- LDH: 134 U/L

Testicular ultrasonography (5 June 2023):

- Multiple microlithiasis,
- No focal testicular mass,
- Bilateral hydrocele.

Interpretation:

Absence of a primary gonadal lesion, supporting a primary intracranial origin of the tumor.

2.6. Treatment

The patient received three cycles of chemotherapy according to the BEP protocol, initiated on 15 June 2023 and completed on 26 August 2023, with good overall tolerance. Serum tumor markers (β-HCG, AFP, LDH) remained within normal limits throughout treatment and follow-up, consistent with the non-secreting nature of intracranial germinomas. Although most pure intracranial germinomas are non-secreting, non-germinomatous intracranial germ cell tumors (teratomas, choriocarcinomas, yolk sac tumors) frequently produce β-HCG and/or AFP. Measurement of these serum markers therefore represents a useful diagnostic tool to guide the work-up and differentiate between tumour subtypes [8][9].

Chemotherapy was followed by radiotherapy delivered using IMRT (Varian®), consisting of pan-ventricular irradiation to a total dose of 24 Gy, complemented by a localized tumor boost of 16 Gy. Radiotherapy was administered between 28 February 2024 and 1 April 2024, with excellent clinical tolerance.

2.7. Follow-Up and Outcome

Radiological follow-up demonstrated a marked reduction in the pineal mass on the CT scan performed on 25 August 2023, with stability of the small residual lesion within the third ventricle and adequate functioning of the ventriculoperi-

toneal shunt. Thoraco-abdomino-pelvic imaging during the same period showed no abnormalities. At the subsequent follow-up on 28 July 2025, the pineal region mass remained stable, measuring 67 × 47 × 45 mm (vs. 41 × 54 × 47 mm in 2024), with unchanged bilateral subdural collections and both ventriculoperitoneal shunts in place. No evidence of recurrence or clinical progression was observed.

Clinically, the patient remained in good general condition (ECOG 1) with a Glasgow Coma Scale score of 15. He reported no further headaches, vomiting, or visual disturbances, and no neurological deficits were detected. His current status is considered stable in the long term.

3. Discussion

Germ cell tumors arise predominantly in the gonads, while their occurrence within the central nervous system remains exceptional [10]. Among these, germinomas—also referred to as dysgerminomas or pure seminomas—account for approximately 60%. They primarily affect children and young adults, with the highest incidence during the second decade of life (45%) [11], and most commonly originate in the pineal gland or suprasellar region [3]. Tumor dissemination or the presence of multiple nodular implants along the lateral and third ventricles is observed in about 10% of cases [6].

Although pineal localization is typical, the radiological presentation of these tumors may be misleading, particularly when differentiating them from teratomas, which are far more heterogeneous and often contain fatty or cystic components. Neuroimaging (CT/MRI) of pineal tumors lacks sufficient specificity to reliably distinguish a germinoma from a teratoma, as some germinomas may present with calcifications or atypical heterogeneity mimicking a teratomatous lesion [12]. In our case, the initial imaging strongly suggested a ruptured pineal teratoma due to the presence of fat density areas and intralesional calcifications. Histological and immunohistochemical confirmation was decisive: the dense lymphocytic infiltrate, together with the specific expression of germ cell markers (PLAP+, CD117+, SALL4+), enabled a definitive diagnosis of pure germinoma. This case underscores the importance of clinico-radiological correlation and the use of stereotactic biopsy to avoid diagnostic errors in midline germ cell tumors.

Pineal germinomas typically cause a relatively rapid onset of symptoms, mainly due to compression of the aqueduct of Sylvius and secondary obstructive hydrocephalus. Patients classically present with headaches, nausea, vomiting, visual disturbances, and sometimes transient vision loss or oculomotor abnormalities, as widely described in the literature [13]. In our observation, the patient exhibited this typical constellation of symptoms—progressive headaches, vomiting, seizures, and decreased visual acuity—consistent with the usual clinical presentation of pineal germinomas and highlighting the central role of intracranial hypertension as the revealing manifestation.

Intracranial germinomas are often characterized histologically by a dense lymphocytic infiltrate composed predominantly of T lymphocytes, but also including

B lymphocytes, plasma cells, and numerous macrophages. This feature may lead to an initial misdiagnosis, such as encephalitis or T-cell lymphoma, particularly when only a small biopsy sample is available [14]. Although the initial biopsy suggested a differential diagnosis between encephalitis and lymphoma, these hypotheses had already been considered clinically. The progressive evolution of symptoms, the large size of the pineal mass on imaging, and the absence of systemic infectious signs made encephalitis unlikely. Likewise, the patient's adolescent age and the radiological appearance of the lesion were less typical of lymphoma. These considerations justified the prompt performance of a stereotactic biopsy, which enabled a definitive diagnosis of pure germinoma through immunohistochemical analysis.

Diagnostic confirmation therefore relies heavily on immunohistochemistry, which enables the identification of specific markers such as PLAP, CD117, and SALL4 (13). In our case, these markers were decisive in confirming the pure seminomatous nature of the tumor despite the abundance of lymphocytes. PLAP (placental alkaline phosphatase) is a classical marker of germ cells, CD117 (c-KIT) is characteristically expressed by undifferentiated germ cells, and SALL4 is a transcription factor expressed in pure germ cell tumors. The co-expression of these three markers is therefore highly indicative of a pure intracranial germinoma, allowing it to be distinguished from other germ cell or lymphoproliferative tumors [15] [16].

Intracranial germinomas are well known for their high sensitivity to both chemotherapy and radiotherapy, which allows excellent clinical outcomes even in the presence of large tumors or ventricular extension [17]. Due to the lack of specific recommendations in major oncology guidelines—namely NCCN and ESMO, which do not provide dedicated protocols for primary intracranial germ cell tumors—management is typically guided by European and international expert consensus statements such as the EANO/SNO/EURACAN recommendations [17]. These generally advocate induction chemotherapy followed by tailored radiotherapy depending on tumor volume and the risk of ventricular or leptomeningeal dissemination.

In our case, the patient received three cycles of BEP chemotherapy (bleomycin, etoposide, cisplatin) followed by pan-ventricular radiotherapy (24 Gy) with a 16 Gy local boost, consistent with established evidence regarding the chemosensitivity and radiosensitivity of intracranial germinomas. For instance, in a series of 60 patients with localized intracranial germinomas, neoadjuvant chemotherapy followed by 40 Gy focal radiotherapy resulted in significant tumor regression and high event-free survival, while underscoring the importance of field design to reduce periventricular relapse [18]. Similarly, the prospective non-randomized SIOP CNS GCT 96 study, which included 190 patients with localized germinomas, compared the efficacy of chemotherapy followed by 40 Gy focal radiotherapy with that of craniospinal irradiation (24 Gy) plus a 16 Gy tumor boost without chemotherapy [19]. These studies collectively support the role of induction chemotherapy in

reducing tumor volume and enabling a more conservative radiotherapy approach while ensuring effective tumor control.

The prognosis of intracranial germinomas is generally favorable, with reported 5-year overall survival rates exceeding 90%, owing to their high responsiveness to therapy. Nonetheless, close surveillance is essential, incorporating clinical evaluation, imaging, and biological monitoring (serum markers and cerebrospinal fluid when appropriate) to detect potential recurrences or late treatment-related complications. Although follow-up strategies vary across protocols, published data support intensive monitoring during the first two years, followed by progressively more spaced evaluations in cases of sustained stability [17].

Furthermore, the long-term implications of therapeutic choices must be considered, particularly in adolescents. The BEP regimen carries risks of gonadal, pulmonary, and renal toxicity, whereas panventricular radiotherapy may lead to late neurocognitive, endocrine, or vascular effects. Given the potential risk to fertility, the patient underwent sperm preservation beforehand, after being informed of the possible impact of treatment. In our case, no late complications have been observed to date, but prolonged follow-up remains essential to allow early detection of potential late toxicities.

Nevertheless, it should be emphasized that conclusions drawn from a single case remain limited, and larger series or prospective studies would be required to confirm the generalizability of these observations.

4. Conclusion

This case illustrates a pure germinoma of the pineal region in an adolescent, initially mimicking a mature teratoma with intraventricular rupture on imaging. The initial histological findings were non-specific, underscoring the diagnostic challenge and the critical importance of immunohistochemistry. Combined treatment with BEP chemotherapy and pan-ventricular irradiation resulted in a favorable clinical and radiological outcome. The publication of this case is relevant due to its distinctive diagnostic and therapeutic features.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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