Intracranial germinoma presenting as anorexia and unexplained weight loss

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**ABSTRACT**

**Objective:** To describe a patient with germinoma of pineal body who presented with anorexia and unexplained weight loss of 70 pounds. **Case Summary:** A 23-year-old man with no significant past medical history presented with nausea, vomiting, anorexia and weight loss of 70 pounds over a period of 6–8 months. Magnetic resonance imaging (MRI) scans demonstrated tumor lesions that were confirmed as germinoma histologically. A good treatment response to surgery, radiation, chemotherapy and alleviation of symptomatology was achieved. The postoperative neurological and MRI follow-up corresponded with the clinical course of the disease and the results of therapeutic procedures. **Discussion:** There were few reports of unexplained weight loss and anorexia, which was misdiagnosed as anorexia nervosa but later, found to be germinoma. Intracranial germinomas have a reported 90% survival to five years after diagnosis. Germinomas, like several other types of germ cell tumor, are sensitive to both chemotherapy and radiotherapy. **Conclusions:** In the initial stages, the differentiation of organic and functional symptoms and the diagnosis of central nervous system tumour may be difficult. Although their good prognosis, the review of available literature suggest that they are a diagnostic and therapeutic challenge. Our notices with this case signify the importance of considering organic disorder before making a diagnosis of depression or anorexia nervosa, particularly if the presentation is a typical. Through this case report, clinicians will be able to appreciate the need to be aware of germinoma as a possible cause of unexplained weight loss. Also, to bear in mind that the complaint of vomiting without a known etiology should facilitate considering a central cause.

**INTRODUCTION**

Tumors of the hypothalamic-pineal region may present with a wide variety of symptoms, including disturbed eating. A germinoma is a type of germ cell tumor that is undifferentiated upon examination. Germinomas are thought to originate from an error of development, when certain primordial germ cells fail to migrate properly. Metastasis has been noted in approximately 22% of cases at time of diagnosis. Males are approximately twice as commonly affected in developing germinomas. Germinomas are most commonly diagnosed between the age of 10 and 21. As with other germ cell tumors (GCTs) occurring outside the gonads, the most common location of intracranial germinoma is on or near the midline, often in the pineal or suprasellar areas; in 5–10% of patients with germinoma in either area, the tumor is in both areas.

**CASE REPORT**

Patient is a 23-year-old male who was referred by his PCP for further workup as he was being investigated for the cause of his weight loss of 70 pounds over a period of 6–8 months. His only complaint was nausea and vomiting. He reported throwing up most of the times he ate. He also complained of loss of appetite.

His previous workup has been negative including complete blood count, basic metabolic panel, sedimentation rate, Creactive protein, liver function tests, computerized tomography (CT) abdomen/pelvis, and ultrasound abdomen.
Esophago gastro duodenoscopy (EGD) was done which showed a single ulcer in antrum and he was started on Omeprazole about 2 months before he was seen in our clinic. Biopsy was negative for malignancy as per patient. Past medical history was significant for a back surgery in 2009 and a hand surgery a year ago because of a dog bite. Dog died the same day spontaneously but was negative for rabies. Family History was significant for ovarian cancer in Mom who is deceased. The patient chewed tobacco all day but did not smoke. Denied alcohol or drug use. Lived alone and was accompanied by his parents for all appointments. Previous history of high-risk sexual behavior and a negative HIV test. His medications included Citalopram and Omeprazole; was later started on Mirtazapine. Vitals have always been stable during all office visits. Depression was considered as a probable diagnosis and he was started on Citalopram a month prior but patient denied any depressive symptoms. Other tests that were done and were negative were HIV, syphilis, Lyme disease, Rocky Mountain spotted fever, Ehrlicchia. He had four visits with more tests including repeat CT abdomen/pelvis, chest Xray, prealbumin, albumin, total protein, anti-transglutaminase IgG and IgM, cortisol, stool for ova and parasites, Giardia antigen, white blood cells, guaiac for blood which were all negative. Patient was also referred to psychiatry for possible component of depression where he was evaluated and was asked to continue Citalopram. Repeat EGD w followup on gastric ulcer and duodenal biopsy for possible malabsorption syndrome was normal. Physical exam was significant for an old dog bite on right wrist, which had completely healed. Decision was made to get a magnetic resonance imaging (MRI) of the brain; it showed two separate masses involving the pituitary infundibulum and pineal area. His brain mass was biopsied; it showed germinoma (Figure 1). The neoplastic cells were strongly positive for c-kit, PLAP, D2-40. There was no other germ cell neoplasm component identified in the samples. He had an MRI of the spine including cervical, thoracic and lumbar regions, which was unremarkable. He underwent intradural resection of intra and suprasellar central skull base tumor. His appetite had improved subsequent to the surgery. He started feeling better and weight had stabilized. Repeat MRI of the brain after surgery showed postsurgical changes and decrease in size of suprasellar mass. The pineal region enhancing mass was found to be stable and measured 11 mm in size.

He was seen at Radiation Therapy Institution and the decision was made for chemotherapy followed by radiation. Serum tumor markers were within normal range. CSF markers were essentially normal with the Beta-hCG in the CSF being slightly above normal (4 with normal range 0–3). He completed four cycles of etoposide and cisplatin (EP) followed by adjuvant radiotherapy. Restaging MRI after two cycles showed significant improvement. He subsequently completed radiation and his last MRI after completion of therapy showed no evidence of tumor (Figure 2). Patient’s symptoms had resolved and patient had started gaining weight.
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**DISCUSSION**

There were few reports of unexplained weight loss and anorexia, which was misdiagnosed as anorexia nervosa but later, found to be germinoma.\(^2\)\(^-\)\(^3\) Pineal neoplasms are rare tumors representing 0.4–1% of all intracranial tumors that usually present with symptoms of increased intracranial pressure and pressure on the midbrain. In rare cases, they are accompanied by abnormal skeletal growth and premature sexual development in young boys. The median age at presentation is 20 years (range 6–49 years) with a median duration of symptoms before diagnosis of 17 months (range 1–35 months). Polyuria was the commonest presenting symptom with suprasellar masses.\(^4\) Diabetes insipidus occurs in several patients, as also partial or complete anterior pituitary failure. Visual failure, anorexia, weight loss and disturbed thirst sensation were also common.

Although the history of our patient may have been suggestive of anorexianervosa, he did not meet all of the DSM-IV criteria for this disorder, and there were clinical features that raised questions about the diagnosis. Although anorexia may occur in males, it is more commonly seen in adolescent females. Our patient had a loss of appetite early in his illness accompanied by nausea and vomiting. He did not have the psychological symptoms of a body-image disturbance or changes in self-perception.

Positron emission tomography scanning was occasionally useful in the evaluation of suprasellar tumours/pituitary stalk lesions deemed too risky to biopsy. Intracranial germinomas have a reported 90% survival to five years after diagnosis. Germinomas, like several other types of germ cell tumor, are sensitive to both chemotherapy and radiotherapy.\(^5\) For this reason, patients’ chances of long-term survival, even cure, are excellent with treatment. A ‘central nervous system-friendly’ chemoradiotherapy regimen comprising vincristine, etoposide and carboplatin and differential daily dose irradiation, usually administered using a partial transmission block technique, produced a 5-year survival of 100% with low morbidity.\(^6\) Treatment may not correct previously abnormal endocrine function although it might improve vision.

**CONCLUSIONS**

In the initial stages, the differentiation of organic and functional symptoms and the diagnosis of central nervous system tumour may be difficult. The presentation of synchronous suprasellar and pineal germinomas is a rare occurrence with clinical expression non-specific for either region.\(^7\) The differential diagnosis is based on the typical clinical features, contrast-enhanced CT/MRI, specific endocrine and lab tests, as well as histological verification.\(^8\)

Although their good prognosis, the review of available literature suggests that they are a diagnostic and therapeutic challenge.\(^9\) Our notices with this case imply that organic disorder should always be considered before making a diagnosis of depression or anorexia nervosa, particularly if the presentation is a typical.

We wish to thank the patient described for consenting to the publication of this case report.

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**Figure 2.** Repeat MRI scan showing no evidence of tumour.
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REFERENCES