Endodermal sinus tumor (EST)

Also known as yolk sac tumor (YST), is a member of the germ cell tumor group of cancers.

Epidemiology

It is the most common testicular tumor in children under 3, and is also known as infantile embryonal carcinoma. This age group has a very good prognosis. In contrast to the pure form typical of infants, adult endodermal sinus tumors are often found in combination with other kinds of germ cell tumor, particularly teratoma and embryonal carcinoma. While pure teratoma is usually benign, endodermal sinus tumor is malignant.

Diagnosis

The histology of EST is variable, but usually includes malignant endodermal cells. These cells secrete alpha-fetoprotein (AFP), which can be detected in tumor tissue, serum, cerebrospinal fluid, urine and, in the rare case of fetal EST, in amniotic fluid. When there is incongruence between biopsy and AFP test results for EST, the result indicating presence of EST dictates treatment.

This is because EST often occurs as small “malignant foci” within a larger tumor, usually teratoma, and biopsy is a sampling method; biopsy of the tumor may reveal only teratoma, whereas elevated AFP reveals that EST is also present. GATA-4, a transcription factor, also may be useful in the diagnosis of EST.

Diagnosis of EST in pregnant women and in infants is complicated by the extremely high levels of AFP in those two groups. Tumor surveillance by monitoring AFP requires accurate correction for gestational age in pregnant women, and age in infants. In pregnant women, this can be achieved simply by testing maternal serum AFP rather than tumor marker AFP. In infants, the tumor marker test is used, but must be interpreted using a reference table or graph of normal AFP in infants.

Treatment

A combination of operation and chemotherapy might be the effective management for EST in the posterior cranial fossa.

Outcome

While pure teratoma is usually benign, endodermal sinus tumor is malignant.

The serum alpha fetoprotein level is well correlated with the severity of the tumor.The prognosis of extragonadal intracranial EST is poor [1)].

Case reports

A case of YST of the medulla oblongata in a 50-year-old woman. She was followed up for 18 months without any tumor recurrence [2)].

Adenocarcinoma Arising in a Yolk Sac Tumor of the Pineal Gland [3)].

A 54-year-old Japanese man presented with disturbance of consciousness, Parinaud's syndrome, and gait disturbance. Magnetic resonance imaging revealed a pineal mass lesion, and subtotal resection of the tumor was achieved. The histologic diagnosis was MGCT, consisting mainly of YST. Although he underwent 5 courses of chemotherapy and craniospinal irradiation after surgery, tumor dissemination could not be controlled, and he died 10 months postoperatively.

The present case highlights the need for clinicians to include YST in the differential diagnosis of acute progressive lesions around the pineal region, even in adult patients [4)].

A case of a 6-year-old boy initially manifested symptoms of dizziness and vomiting. Computed tomography (CT) and magnetic resonance imaging (MRI) showed a large irregular oval tumor in the cerebellar hemisphere. They subtotally removed the tumor by microsurgery through the left suboccipital approach. Immunohistochemical staining showed that alpha fetoprotein (AFP) was positive and the Ki-67 proliferation index was high (60%), suggesting a germ cell tumor. After 3 months of follow-up, neither recurrence of tumor nor complications were found in the patient. The diagnosis of YST should be confirmed on the basis of clinical manifestations, neuroimaging and pathological findings. Gross total resection (GTR) is an ideal treatment for YST. However, due to the location of the tumor, GTR is usually difficult, and the rate of postoperative complications is high. This reported case shows that subtotal resection can be a good treatment strategy for YST [5)].

A patient with primary YST in the pineal region who achieved long term survival. Despite undergoing treatment, he experienced several recurrences over a 15-year period.

Brain magnetic resonance imaging (MRI) demonstrated the presence of space-occupying lesions in the pineal region and the medial tail of the left lateral ventricle. The tumors were excised, and the histological diagnosis suggested an intracranial YST.

The patient achieved long term survival after combined modality therapy including surgery, stereotactic radiosurgery (SRS)/intensity modulated radiation therapy (IMRT), chemotherapy, and targeted therapy.

The disease remained stable. However, the patient gave up treatment and passed away in October 2020, with a total survival of about 15 years.

To the best of our knowledge, this patient with intracranial YST had received a longer survival

compared with other published reports [6].

Gkampeta et al. reported the case of a 3-year-old boy with a primary posterior mediastinal yolk sac tumor who was managed initially with surgery, followed by chemotherapy and had a favorable prognosis. In the literature on yolk sac tumors presenting as a mediastinal mass, pediatric germ cell tumors have been reported very rarely in the posterior mediastinum [7].

1)
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