Teratoma in the pineal region in two brothers

Case reports

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The occurrence of pineal teratoma in two brothers is described, and the pathogenesis of this type of tumor is discussed. This is the first report of familial occurrence of pineal teratoma in two siblings.

Key Words: teratoma • pineal tumor • congenital disorder • brain tumor • pathogenesis

The familial occurrence of tumors of the nervous system, such as gliomas and presacral teratomas, has been well documented. Heredity has been found to play some part in the etiology of presacral teratomas and tumors in patients with phacomatosis. It has also been suggested that genetic factors may be related to the pathogenesis of intracranial gliomas. However, the familial occurrence of true teratomas in the pineal body has not been reported so far. To our knowledge, this is the first report of pineal teratoma occurring in siblings.

Case Reports

Case 1

This 13-year-old boy was admitted to Asahi General Hospital on March 24, 1973, with a 7-month history of polydipsia, polyuria, and loss of appetite. The patient was the eldest of three siblings, two boys and one girl, born of healthy parents without consanguinity. He was born at full term of a normal pregnancy, and his subsequent development was entirely normal until the present illness.

Examination. On admission he showed bitemporal hemianopsia, upward gaze palsy, horizontal nystagmus with lateral gaze, and ataxic gait. There was no other focal neurological abnormality. Precocious puberty was not found. Plain skull film revealed no calcification in or near the pineal body.

Course. After a ventriculoperitoneal shunt for hydrocephalus was placed on May 16, 1973, the patient's general condition improved gradually. However, he still showed upward gaze palsy and horizontal nystagmus with lateral gaze. One month later, he became mute, and his general condition deteriorated. The second pneumoventriculogram, performed on October 24, disclosed a small mass in the pineal region. He received a course of radiation therapy with 2500 rads of cobalt-60, but he died on December 31, 1973.

Postmortem Examination. An infiltrating tumor with a small cyst, 0.8 cm in diameter, was found in the pineal region. The tumor invaded the wall of the third ventricle and the lateral ventricles. The meninges were thickened and opaque, which was compatible with the finding of purulent meningitis (Fig. 1). Histologically, the tumor was composed of keratinized squamous epithelium, sebaceous glands, fat tissues, and columnar epithelium, with goblet and Paneth cells indicating intestinal structures (Fig. 2 upper left). In addition to these three germinal components, the tumor contained germinomatous elements, which infiltrated into the...
FIG. 1. Case 1. Photograph of the autopsy specimen. An infiltrating tumor with a small cyst (about 0.8 cm in diameter) is observed in the pineal region (arrow). The wall of the third ventricle is invaded by the tumor, and the aqueduct is dilated, with occlusion at the level of superior colliculus (arrowhead).

parenchyma of the surrounding brain (Fig. 2 lower left). The histological diagnosis was made of a pineal teratoma with some germinomatous components.

Case 2

This 17-year-old boy was admitted to the University of Tokyo Hospital on May 28, 1979, because of a progressive right-sided hemiparesis with a 2-year history of diplopia. This patient was a younger brother of the patient in Case 1, and the middle of the three siblings. He too was born at full term of a normal pregnancy. At 11 years of age, a plain skull film showed abnormal intracranial calcification.

Examination. The patient had upward gaze palsy and horizontal nystagmus with lateral gaze. There was a spastic hemiparesis and a hemihypesthesia with extensor plantar response on the right side. Computerized tomography (CT) revealed a large mass, with calcification in the pineal region, that encroached upon the posterior portion of the third ventricle. There was a low-density area in the left frontal horn, which was considered to be caused by fat globules from the ruptured tumor (Fig. 3 left). Carotid and vertebral angiograms showed a mass in the posterior third ventricle and a diffuse vascular shadow in the left posterior portion of the lateral ventricle, which was also found on CT with contrast enhancement (Fig. 3 right). Titer of alpha-fetoprotein and human chorionic gonadotropin in both serum and cerebrospinal fluid were within normal range.

Operation. A right occipital craniotomy was carried out on June 7, 1979. The tumor was approached by splitting the splenium. The mass that occupied the posterior part of the third ventricle and the pineal region was found to consist of white cheese-like material, hair, and finger-tip sized bone, three-quarters of which were removed piece by piece. The left half of the mass was quite vascular in nature, infiltrating the left thalamus and posterior limb of the internal capsule. On histological diagnosis of this part during the operation this tumor was reported to be a germinoma. Since this tumor is known to be radiosensitive, it was decided to abandon further removal of the mass in order to preserve function. Ventriculoperitoneal shunting was performed on June 23, 1979, and later the patient was treated with a course of radiation therapy (6000 rads to the whole brain). He remains bedridden.

Pathological Examination. The tumor consisted of germinatal elements, including skin with sebaceous glands, smooth muscle, bone, and intestine (Fig. 2 upper right). A germinoma-like structure that had cells with relatively large nuclei intermingled with small lymphoid cells was present in the specimen taken from the left half of the tumor (Fig. 2 lower right). The diagnosis was pineal teratoma with some germinomatous components.

Comment

The sister of these two patients, who was the youngest of the three siblings, underwent CT scanning but no abnormality was revealed.

Discussion

It is surprising that teratoma of the pineal region has not previously been found in siblings, since it is known that this type of tumor is congenital in origin. In Japan, the incidence of teratomas in the pineal region is reported to be about 0.4% to 2.1% of all intracranial neoplasms, an incidence about two to 10 times that in Western countries. The familial occurrence of presacral teratoma has been documented in six families by Hunt, et al. It was associated with a sacrococcygeal bone defect,
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Fig. 2. Photomicrographs of the tumor. Upper: Three germinal components are found in the tumor of both Case 1 (left), and Case 2 (right). H & E, × 45. Lower: Germinomatous portions of the tumor in Case 1 (left), and Case 2 (right) are shown, in which large spheroid cells are intermingled with small lymphoid cells. H & E, × 225.
vesicoureteral reflux, skin dimples, retrorectal abscess, and anorectal stenosis. These authors suggested that this tumor is inherited as an autosomal-dominant trait, considering the pattern of occurrence. In our cases occurring in the pineal region, associated malformations indicative of hereditary factors were not observed.

Medulloblastoma occurring in siblings has been described in seven reports.\textsuperscript{5,6,7,8,9,10,11} Recently, we encountered medulloblastoma in two brothers (one was 11 years old and the other 10 years old at the onset of symptoms). In view of the striking similarities in the sex, age at onset of symptoms, clinical manifestations, and pathological findings in affected members in each reported family, Yamashita, \textit{et al.},\textsuperscript{10} suggested that heredity must play a role in the etiology of medulloblastoma, at least as one of several factors.

Regarding the pathogenesis of pineal and intraspinal teratoma, Rogers\textsuperscript{12} presented the hypothesis that if one dizygotic twin with epithelial defects is partially destroyed by fetus to fetus interaction \textit{in utero}, the damage may be recognized as a teratoma or as an area of inappropriate skin cover to the defects. If this hypothesis is true, intrauterine factors rather than heredity must be related to the pathogenesis of this tumor. Thus, such a teratoma occurring in two siblings as reported here would be just an incidental phenomenon. Provided that the incidence of brain tumor is 14.5 among 10\textsuperscript{8} patients per year,\textsuperscript{11} and 2.1\% of all brain tumors are pineal teratomas,\textsuperscript{12} the chance of pineal teratoma in two siblings would be \((14.5/10^8 \times 2.1/100 \times 50)^2 = 2.3/10^8\) during 50 years. This calculated occurrence rate is extremely low. This low occurrence rate, as predicted by Rogers' hypothesis, together with the paucity of reported cases in siblings, support the role of intrauterine factors in the pathogenesis of this tumor.

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\textbf{References}

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