Turcot syndrome (glioma polyposis)

Case report

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A case of Turcot syndrome (glioma polyposis) is described in a 22-year-old woman. The patient initially presented with a frontoparietal glioma, and was subsequently found to have segmental colonic polyposis with adenocarcinomatous changes. Her colonic polyposis was nonfamilial.

KEY WORDS • Turcot syndrome • glioma polyposis syndrome • cerebral astrocytoma • colonic polyposis • colonic adenocarcinoma

Since the description in 1959 by Turcot and his associates of the association between cerebral gliomas and colonic polyposis, case reports have been published sporadically in gastroenterological and general surgical journals. To date, 22 cases that fulfill the criteria of Turcot syndrome have been reported. Even though the presence of a glioma is an essential part of the syndrome, very few reports have appeared in the neurological or neurosurgical literature. We describe the case of a young woman who first presented with a frontoparietal astrocytoma, and was later found to have colonic polyposis with adenocarcinomatous changes in one of the polyps.

Case Report

This 22-year-old woman was admitted with a history of gradually progressive headache of 1 month's duration. Nausea and vomiting developed 1 week prior to her admission. She gave no history of bowel disturbance. Detailed family history on both the paternal and maternal sides yielded no history of colonic polyposis or cerebral neoplasms.

Examination. The patient was alert, and had bilateral papilledema with hemorrhages and exudate. There was no neurological deficit in the limbs. Computerized tomography (CT) scans of the brain showed a large intrinsic right frontoparietal tumor which enhanced in an irregular fashion with administration of contrast material (Fig. 1). Right carotid angiography showed a very vascular right frontoparietal tumor which had a primitive blood supply.

First Operation. Subtotal resection of the tumor was carried out. The tumor was soft, very vascular, and grayish in color, with small multiple cysts in its medial part. The histopathological examination showed this to be a cellular fibrillary grade II astrocytoma (Fig. 2 left). The immediate postoperative course was uneventful.

On the 11th postoperative day, the patient developed the sudden onset of bleeding per rectum and a partially prolapsed rectal mass. Sigmoidoscopy revealed intussusception associated with a large papillomatous mass, 6 × 4 cm in size. There were additional small polyps in the immediate vicinity. A biopsy was taken and the intussusception was reduced with a Gastrografin (meglumine diatrizoate) enema. Subsequently, a barium enema showed a cauliflower-like mass occupying almost the whole lumen of the distal sigmoid colon.

Second Operation. At laparotomy, the relevant segment of the colon was opened. A segmental resection of the colon was performed to include all the palpable polyps. Histological examination of the specimen showed definite adenocarcinomatous changes in the largest polyp (Fig. 2 right). The postoperative course was uneventful. The patient was transferred to the radiotherapy unit for further treatment of the glioma. She has completed a course of radiotherapy for the cerebral tumor, and at the follow-up period of 6 months she remains alert and well, without any neurological symptoms.
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FIG. 1. Computerized tomography brain scan showing a large right frontoparietal tumor with necrosis, pleomorphism, and contrast enhancement.

Discussion

In 1949, Crail first described the association between primary malignancies arising in the rectum, brain, and thyroid. Ten years later, Turcot, et al., first described the association between malignant tumors of the central nervous system (CNS) and familial polyposis of the colon, giving rise to the term “Turcot’s syndrome.”

Another name for this disorder is glioma polyposis syndrome. Since the article by Turcot, et al., various reports have appeared expanding the definition of the syndrome to include certain benign non-neuroepithelial tumors of the CNS, like meningiomas and pituitary neoplasms. Some authors have also included non-familial polyposis coli and adenocarcinoma arising in the colon. We believe that non-neuroepithelial tumors like meningiomas and pituitary tumors should not be classified as Turcot syndrome, but that the inclusion of non-familial colonic polyposis and of adenocarcinomatous changes occurring in these polyps is justified.

The association between a classical prototype of pre-cancerous disorder, like familial colonic polyposis and a neuroepithelial brain tumor, is of interest to clinicians. Colonic polyposis is associated with various other disorders, like polyposis with osteomas (Gardner’s syndrome), polyposis coli with multiple sebaceous cysts (Oldfield syndrome), polyposis coli with cartilagenous exostoses (Zanca syndrome), colonic polyposis with melanine spots of the buccal mucosa (Peutz-Jeghers syndrome), and generalized polyposis of the intestine with alopecia, dyspigmentation, and onychodystrophy (Cronkhite-Canada syndrome). The association of colonic polyposis with these various connective tissue and epithelial tumors has led to differing views regarding the mode of genetic inheritance of these tumors.

An important factor of interest in Turcot syndrome is whether it is an inherited disease and, if so, what is the mode of transmittal. Colonic polyposis is a well known hereditary precancerous disorder. The asso-

Fig. 2. Left: Photomicrograph showing an anaplastic astrocytoma. The presence of mitoses and the substantial variability among the tumor cells together with the hypercellularity point to the malignant nature of this tumor. H & E, x 350. Right: Photomicrograph showing a section of the infiltrating adenocarcinoma of the sigmoid colon removed during surgery. Note the papillary appearance of the malignant growth and deep penetration into the muscular layer. H & E, x 10.
cation between colonic polyposis and various other neoplasms is also recognized. Different views have been expressed on the subject of inheritance in Turcot syndrome. A recessive gene or a single dominant pleiotropic mutant gene (transmitted in the autosomal dominant mode) has been implicated. No definite opinion has emerged because of the small number of Turcot syndrome cases and also because this entity has been reported in patients with nonfamilial colonic polyposis. No family history of brain tumor or of colonic polyposis was found in our patient.

The possibility that some degree of immune deficiency plays a role in the genesis of multiple malignant tumors in these cases is of interest, especially as a substantial number of these patients had three or more malignant tumors. Some support for this theory comes from the occurrence of this syndrome predominantly in a young population. Of 22 reported cases, 20 patients were below the age of 25 years and 14 were below 19 years of age. In 1974, Kersey, and reported multiple neoplasms in children with primary immunodeficiency disease. Our patient, aged 22 years, showed no clinical or pathological features of immunodeficiency.

These patients are usually managed with the accepted treatment for glioma and colonic polyposis. For cerebral glioma, partial or subtotal excision of the tumor with radiotherapy and/or chemotherapy is the commonest method of treatment. For colonic polyposis, a segmental partial or total resection of the colon is advised depending upon the extent of the polyposis. In our patient, who presented initially with symptoms related to glioma, a subtotal resection of the glioma was carried out. One week after the craniotomy she developed the symptoms of colonic polyposis, and for this a segmental resection, with adequate margins of normal colon, was performed. She subsequently received radiotherapy for the glioma.

Contrary to our patient, most of the cases reported in the literature presented first with the symptoms of colonic disease. Colonic polyposis is associated with a myriad of other neoplasms, but, of all these, the only lethal tumor is cerebral glioma. Early recognition of a glioma in such patients may be of clinical importance with respect to increased survival potential. In view of this known association between colonic polyposis and glioma in Turcot syndrome, we recommend that patients with colonic polyposis be screened periodically for cerebral glioma, especially when presenting at a young age. In addition, in patients with familial colonic polyposis it may be worthwhile to perform similar screening of the siblings. Such screening can now easily be accomplished by periodic CT scanning.

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