Case Report

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Turcot Syndrome: A Synchronous Clinical Presentation of Medulloblastoma and Lower Rectal Tumor

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Abstract

We report the case of a 16-year-old patient who was operated for medulloblastoma with simple operative follow-up after radio and chemotherapy. Two years later, she was admitted, in a state of cachexia, for abdominal pain and rectorrhagia. The explorations found an infected lower rectal tumor. The patient underwent an urgent surgery. Immediate consequences were marked by the installation of a severe septic shock causing the patient's death.

Keywords: Turcot syndrome, Medulloblastoma, Genetic

Introduction

Turcot Syndrome (TS) is a rare hereditary disorder with genetic susceptibility to cancer which is characterized by the occurrence of colorectal and Central Nervous System (CNS) tumors.

Case Report

We report the case of a 17-year-old girl who underwent surgery for medulloblastoma (Figure 1) with simple operative follow-up. She had an adjuvant radio-chemotherapy.

Two years later, she consults, in a state of cachexia, for abdominal pain with rectorrhagia. The explorations concluded for infected the lower rectum tumor (Figure 2).

The patient underwent an urgent surgery. The immediate consequences were marked by the installation of a state of severe septic shock causing the death of the patient.

Figure 1: Axial CT and brain MRI showing medulloblastoma of the posterior fossa.

Figure 2: Pelvic computed tomography showing a large abscessed rectal tumor.

Discussion

Turcot syndrome is also known as brain-tumor polyposis and is characterized by the occurrence of primary tumors of the CNS and multiple colorectal adenomas and/or colonic adenocarcinoma [1,2]. From a genetic standpoint, TS can be divided into two subtypes: the first is manifested by the MMR gene and PMS2 with a high risk of developing glioblastoma, and the second type is manifested by the adenomatous polyposis coli gene germline mutation with a high risk of developing medulloblastoma [1][3].

The mode of inheritance of this disorder is controversial [4][5]. Autosomal dominant [5][6], autosomal recessive [7], and sporadic [7] modes of transmission have been proposed. Some have theorized that TS is an allelic variant of familial adenomatous polyposis (FAP) with the extracolonic manifestation of CNS neoplasia [6].

Controversy still persists regarding the mode of inheritance and whether Turcot syndrome constitutes a distinct genetic disorder. One common feature of these syndromes is association with inheritance of germline mutations in the DNA mismatch repair genes.

It is now proposed that inheritance of 2 mismatch repair mutations in an individual along with the unique tumor spectrum should be defined separately from Lynch syndrome I and II, or the subtypes Turcot and Muir-Torre and termed Lynch III, to identify individuals with constitutively compromised mismatch repair associated with biallelic mutations [8]. Seventy percent of patients with Turcot syndrome present with intestinal cancer and develop the CNS manifestations within 5 years; the mean age range of disease onset is in the second and third decades [1].

However, if brain tumors appear first, colonic polyps tend to become symptomatic within 1 year [9]. Our patient had an usual 02-year gap between the initial diagnosis of medulloblastoma and developing rectal cancer.

Recently, Sarin et al. Reported on a TS type-2 patient who had an unusual 22-year gap between the initial diagnosis of medulloblastoma and colon cancer [10]. Although the reported cases of TS-associated brain tumor have often been associated with enhanced survival time, however the reason remains in question.

Clinically, The usual presenting symptoms are those of a primary CNS tumor, often a brain tumor and rarely a spinal tumor. The neurologic signs and symptoms depend on the location of the tumor. There is usually a family history of familial adenomatous polyposis coli or colorectal carcinoma. Our patient did not have a such family history.

Skin manifestations may include cafe-au-lait and other pigmented spots, sebaceous cysts, and basal cell carcinomas [11]. Multiple cutaneous lesions are present in more than 30% of patients with TS.

Ophthalmological findings include congenital hypertrophic pigmented epithelium that correlates with the expression of polyps in familial adenomatous polyposis coli patients [12].

The diagnostic workup of a patient with a suspected CNS tumor is similar to that of primary brain and spinal cord tumors. Those with a family history of adenomatous polyposis coli should undergo screening and surveillance colonoscopy. Patients with TS should undergo genetic testing, and tissue removed at surgery should be genotyped [13].

The prognosis of Turcot syndrome is generally poor. More than two-thirds of the patients die within 5 years of the manifestation of the disease's first symptoms, but survival may be exceptionally long in some cases. The cause of death is usually the malignant brain tumor, but some patients die due to colorectal malignancy [13].

Conclusion

Turcot syndrome is a rare but quite serious disease characterized by clinical polymorphism and often late diagnosis. It must be searched in front of evocative personal or family history.

Molecular genetic allows early selection of subjects at risk and access to early stages treatment.

Conflicts of Interest

The authors declare that there are no conflicts of interest regarding the publication of this article.

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