

Case Report: Retinoblastoma

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Abstract:

Retinoblastoma is a cancer of the retina and is the most common type of eye cancer in children. The RB1 gene with a mutation usually develops retinoblastoma in both eyes and there are often several tumors within the eye such as melanoma in children. The main treatment for retinoblastoma are: Surgery; Radiation therapy; Photocoagulation; Cryotherapy; Thermotherapy; Chemotherapy; High-dose chemotherapy and stem cell transplant. Besides treatment, palliative care's patient is very important in quality of life. This paper presented 2 cases of retinoblastoma in one family with clinical signs, treatment and some palliative care contributing in diversifying quality of life for these patients.

Key words: Retinoblastoma, clinical signs, palliative care, quality of life.

1. Introduction

Retinoblastoma is an intraocular malignancy with primitive neuroendocrine origins that primarily affects young children. With ICD10 C69.2 is used for retinoblastoma. The estimated incidence of retinoblastoma varies by country from 3.4 to 42.6 cases per million live births. The incidence is 11.8 cases per million live births among children less than 5 years of age, and retinoblastoma represents 6.1% of all cancers in this age group in the United States [1]. The highest incidence of retinoblastoma typically affects young children with less than 4 years old [2]. It occurs equally in males and females. On presentation, approximately 60% of cases are unilateral, and the remaining 40% are bilateral [3]. Patients diagnosed with retinoblastoma are categorized by whether the mutation is germline or somatic.

This paper presents 2 cases of retinoblastoma in one family with clinical signs and some treatments are discussed contribution of diversifying take care of these conditions.

2. Case Report:

Two cases of retinoblastoma in one family are reported. The first is a son, a brother; the second is a girl child, a daughter. (Figure attach)

Case 1: A 7 year -old man child: He is a brother with a daughter of one family. He is a pupil at primary school. Onset 6 months ago he was observed with cat eye reflex, proptosis and exophthalmos. He was transferred ophthalmologist for checkup eye problems. General=Height 1,1meter -Weight 30kg- Pulse 90/' Arterial tension =110/60mmHg. Mental=Nothing abnormal. Right eye (RE)= Fundus exam: fungating mass; Left eye (LE)= Tumor. Visual acuity (VA) of RE= Counting finger (CF) 3meter; LE=No perception of light. Both eyes (OU) =Blindness

Diagnosis: A blind boy with:

RE=Retinoblastoma group A. LE= Retinoblastoma group E (According to International Classification of Retinoblastoma 1990s)

Treatment: LE=Enucleation+ artificial eye+ Radiation + Chemotherapy;

RE=Follow up.

Case 2: A 3 year- old girl child: He is a daughter with a brother of one family. Onset 3 months ago with cat eye reflex, proptosis and exophthalmos was the same condition with your brother but these developments was quickly. She was transferred ophthalmologist for checkup eye problems. General=Height 0,7meter -Weight 22kg- Pulse 90/' Arterial

tension=100/60mmHg. Mental=Nothing abnormal. Right eye (RE) = tumor; Left eye (LE) = Fundus exam: fungating mass. Visual acuity (VA) of RE=No perception of light (-); LE=Counting finger (CF) 1 meter; Both eye (OU) =Blindness

Diagnosis: A blind girl with:

+RE=Retinoblastoma group E. LE= Retinoblastoma group D (According to International Classification of Retinoblastoma 1990s)

+Melanoma = left face

Treatment: RE=preparation of enucleation+ Radiation; LE=Follow up.

The period of follow-up of 2 patients post intervention was 48 months in case 1 and 24 months in case 2. In case 1, he has been learning at blind school and in case 2 she has been living at Buddha pagoda's orphan house for disability children. Both cases have been continuing to learn, to participate with the same age child in community and their quality of life are acceptable.

Classification of Retinoblastoma: The International Classification of Retinoblastoma (ICRB) (see Table 1 at bottom) was developed to better predict those with intraocular retinoblastoma.

3. Discussion:

For decades, the retinoblastoma susceptibility gene RB1 is a tumor-suppressing gene. It encodes a protein with a regulatory function in the cellular growth cycle at the G1 checkpoint. It is located on subband 13q14.2. Both alleles of the retinoblastoma gene have to be inactivated for tumor development [2]. Trilateral retinoblastoma is the term often applied in the setting of bilateral retinoblastoma. It is a rare neoplasm that occurs in patients with germline mutations. While this is more common in bilateral cases, some patients with unilateral retinoblastoma that carry germline mutations are also at risk for developing this tumor [4]

A careful history of present illness, family history, and thorough ophthalmic examination are critical for prompt diagnosis. The doctor must specifically inquire about a family history of blindness, eye tumors, childhood malignancies, or enucleations. A family history of other cancers such as sarcomas may also be suggestive. Two cases in this paper are in one family. In rare cases, retinoblastoma can present with pain and inflammation and have a similar presentation to endophthalmitis, uveitis, hemorrhage or preseptal or orbital cellulitis. Case 2 is specific for these ophthalmic signs

Follow up: Patients with hereditary retinoblastoma are at

increased lifetime risk of developing secondary malignancies throughout the body. The most common secondary tumor is osteosarcoma [5]. Other tumors include fibrosarcoma, and melanoma. Patients who have been treated with radiation are at higher risk for secondary tumors in the field of treatment. Long term follow-up of all retinoblastoma patients is mandatory with special vigilance if patients have germline mutations

Screening for detecting: The American Academy of Pediatrics policy statement on Red Reflex Examinations in Neonates, Infants, and Children recommends that all neonates, infants, and children should have an examination of the red reflex before discharge from the neonatal nursery and at all subsequent routine health supervision visits. [6]

General treatment: Spontaneous regression of retinoblastoma occurs, but is rare. The priorities in the treatment of retinoblastoma are to preserve life, preserve globe, and vision. Treatment modalities include systemic chemotherapy with focal consolidation, intra-arterial chemotherapy, and for small tumors, focally destructive therapy (cryopexy, laser photocoagulation, hyperthermia and plaque irradiation).

Surgery: Enucleation may be needed to remove the eyeball. This may help keep the cancer from spreading to other areas of the body. After removing the eyeball, the surgeon places an artificial eye. Patients who have an eye enucleated will continue to be followed to ensure there is no evidence of tumor in the other eye. In patients with advanced bilateral retinoblastoma, traditionally the more severely affected eye has been enucleated. In 2 cases mentioned above, the tumor is large and there is poor vision. So, an indication of enucleation is necessary.

In developing countries including Nigeria, the survival of patients with retinoblastoma is very low [7]. In advanced countries, however, as a result of the advances in treatment and early presentation of patients with retinoblastoma to the medical facilities, survival rate of retinoblastoma cases is more than 90% [8]. Enucleation with chemotherapy was the intervention with the highest number of survival rate after 4 year follow-up. There was weakly positive association between the survival rate and the intervention employed [8]. The earlier diagnosis can be done, the more likely it can be treated effectively.

Palliative care: The role of palliative care at the end of life is to relieve the suffering of patients and their families by the comprehensive assessment and treatment of physical, psychosocial, and spiritual symptoms patients' experience. [9]

Quality of life: The period of follow-up of 2 patients post intervention in this paper was between 24-48 months. Two patients are continuing to learn, to play, and to participate all activities with the same age child. All of these as a part of palliative care, can be a reason make the patients decreasing worry about their disability. Despite of a developing country the quality of life index of Vietnam is ranged 60 in 2018 [10].

4. Conclusion:

These severe and total blindness cases are contributing of confirming clinical eye signs of retinoblastoma. Their mental are being well development by integrating with community as a palliative care in order to get better quality of life. So the firstly is artificial eye to have got both eyes in esthetic view. The secondly, patients have to learn, to participate all activities with the same age child. These can be a reason make the patients

decreasing worry about their disability, as a part of palliative care, contributed quality of life.

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Author Profile

Duong Dieu graduated MD 1978 and PhD 2002 He was Head of Ophthalmology Department between 1980-2010. From 2010-2018 he was Dean of Optical Ophthalmology Faculty of Nguyen Tat Thanh University Vietnam. He now is a lecturer of Faculty of medicine of Nguyen tat Thanh University VN. Add 300A Nguyen Tat Thanh street-Dist 4-Hochiminh City Vietnam.

Table 1: International Classification of Retinoblastoma (ICRB) 1990s

Group A	Small intraretinal tumors (< 3mm) away from foveola and disc.
Group B	Tumors > 3mm, macular or juxtapapillary location, or with subretinal fluid.
Group C	Tumor with focal subretinal or vitreous seeding within 3mm of tumor.
Group D	Tumor with diffuse subretinal or vitreous seeding > 3mm from tumor.
Group E	Extensive retinoblastoma occupying >50% of the globe with or without neovascular glaucoma, hemorrhage, extension of tumor to optic nerve or anterior chamber.

