CASE REPORTS

FANCONI ANEMIA PRESENTING LATE AS MALIGNANT BUCCAL CARCINOMA; A CASE REPORT

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ABSTRACT

Fanconi anemia (FA) is a rare, genetically and phenotypically heterogeneous autosomal recessive disorder. It is characterized by various congenital malformations, progressive bone marrow failure usually at a very young age and of tumor development. The mean age of diagnosis was 7 years and the average life expectancy was 25 years. We report a rare case of fanconi anemia presenting as a solid tumor of buccal cavity and found to be metastasizing on staging at an unusually late age i.e. 52 years.

Keywords: Buccal carcinoma, Fanconi anemia, Phenotypically heterogeneous.

INTRODUCTION

Fanconi Anemia (FA) is a rare, genetically and phenotypically heterogeneous recessive disorder characterized by various congenital malformations, progressive bone marrow failure usually at a very young age and of tumor development with a mean age of diagnosis being 7 years and the average life expectancy being 25 years[1].

CASE REPORT

An unmarried woman in her late 50s presented to the ENT department of Combined Military Hospital Rawalpindi, Pakistan with a painless swelling in her left cheek which was whitish in color and progressively increased in size over the period of 06 months.

A sample was taken through excisional biopsy, that constituted two gray-brown tissues masses collectively measuring 2.5cm x 1cm x 3.8cm. Microscopy revealed skin tissue with nests of atypical squamous cells having pleomorphic nuclei with prominent nucleoli. Keratin pearl formation was visible. A diagnosis of Squamous Cell Carcinoma (SCC), WHO-grade moderately differentiated was made. For staging purposes, a multi-slice, multi-detector contrast CT scan of the face and neck was carried out which revealed a soft tissue mass on left side of the cheek measuring 2.6cm x 2.2cm with irregular margins, which on peripheral contrast enhancement showed extensive erosion of left hemi-mandible including its hemi-ramus with involvement of superficial and deep portions of left Masseter muscle. The appearances were consistent with Squamous Cell Carcinoma of the Buccal Mucosa. The solitary enlarged metastatic lymph node was detected at level I-b measuring 1.1cm along its shortest axis. The tumor was staged as T4b, N1, Mo.

She was planned for trans-oral wide local excision with radical neck dissection. The pre-anesthetic assessment revealed decreased cell count in all three lineages with a Hb level of 6.4 g/dl, TLC 3.4 cells per liter and Platelets 41 x 10^3 per microliter and was referred to Hematology OPD for further management and disposal.

Further assessment revealed that she had irregular menstrual periods with profuse bleeding, history of easy bruising and multiple dark spots at the trunk. On examination she was pale, had cervical lymphadenopathy at the level of left I-b, microcephaly, micrognathia, left side oral ulcer involving Stenson's duct, and left-sided non-tender facial swelling, hypoplastic thumbs, hypoplastic radii with weak pulses, multiple café au late & hypo pigmented spots on body.

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The peripheral blood film showed anisocytosis, macrocytosis, reticulocytes 1.2%, Rouleaux formation, and dysplastic neutrophils. Bone marrow trephine biopsy revealed hypocellular bone marrow with reduced trilinear hematopoiesis with prominent lymphocytes, plasma cells and increased iron stores. Chromosomal breakage analysis with clastogenic agent like mitomycin C (MMC) revealed multiple breakages and aberrations confirming the diagnosis of Fanconi anemia.

Outcomes and Follow Up: Tans-oral wide local excision with radical neck dissection was done successfully for the SCC as histopathology showed tumor clearance.

DISCUSSION

This was the first case that was presented in Pakistan; that too at such a late age. The incidence of FA is yet to be determined within Pakistan.

FA is defined by the fragility of chromosomes to DNA cross-linking agents such as diepoxybutane and mitomycin C. Presence of mutations of in any one of the different FA genes (FANCD1, FANCJ, FANCM, FANCN, FANCO, FANCS) are involved in the development of FA. FA is then classified according to the gene mutated as A, B, C, D1, D2, E, F, G[2]. According to the International Fanconi Anemia Registry (IFAR) group A (65%), C (15%) and G (10%) are the most common[2,3].

Patients with FA that live long enough are at high risk of developing tumors of the head and neck region with the most common type being Oral Squamous cell Carcinoma and the most common sites being the tongue, gingiva, and buccal mucosa, respectively with females developing tumors significantly later than males[4,5]. The higher risk in men may be attributed to the development of behavioral risk factors like alcohol and tobacco, after 45 years of age[5].

The management of malignancies in FA patients is like that of the general population. They do however require preoperative hematological consultation. Due to the fragility of chromosomes, a less than the normal cancericidal dose of radiotherapy and/or chemotherapy is advised, if any. Since the difficulty in treating cancers in FA is high, the early diagnosis of malignancies in FA patients is extremely essential[2].

CONFLICT OF INTEREST

This study has no conflict of interest to be declared by any author.

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