Case Report

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A rare Congenital Condition Dyskeratosis Congenita Presenting with Carcinoma Hypopharynx in a 16 Year Old Child

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Abstract

Dyskeratosis congenita (DC) is an inherited bone marrow failure and cancer predisposition syndrome caused by defects in telomere biology. The most frequent solid tumors were head and neck squamous cell carcinomas followed by skin and anorectal cancer. The pediatric age group often present DC as a multisystem disorder. However, to date there has been no comprehensive quantitative analysis of cancer risk in Dyskeratosis Congenita. Since our patient is a case of malignancy, multimodality management is required which involves Radiation Oncology, Hematology, Medical oncology and Otorhinolaryngology. Proper counselling and multidisciplinary team approach is essential to treat a case of malignancy especially in Dyskeratosis congenita.

Keywords: Circulating tumor cells; Liquid biopsy; Microfluidics Ovarian cancer; Cancer research; Tumor; Gonad cancer

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Introduction

Dyskeratosis congenita (DC) is an inherited bone marrow failure and cancer predisposition syndrome caused by defects in telomere biology [1]. The exact prevalence of dyskeratosis congenita is unknown. It is estimated to occur in approximately 1 in 1 million people. The consequences of DC affect all body systems; these may include the diagnostic triad of abnormal nails, reticular skin pigmentation, and oral leukoplakia; bone marrow failure, pulmonary fibrosis, liver disease, neurological and ophthalmic abnormalities, as well as increased risk of cancer also occur. The most frequent solid tumors were head and neck squamous cell carcinomas followed by skin and anorectal cancer [2]. There is the presence of an excessive telomere shortening in this patient population, which in the absence of a deoxyribonucleic acid (DNA) damage response may lead to genomic instability and a predisposition for malignant transformation (**Figure 1**). The pediatric age group often present DC as a multisystem disorder, whereas adult patients generally present a very variable phenotype, not necessarily with the classical features of DC, and being a carrier of telomerase mutations usually acts as a risk factor [3].Since it is a multisystem disorder multidisciplinary team approach often required for management.

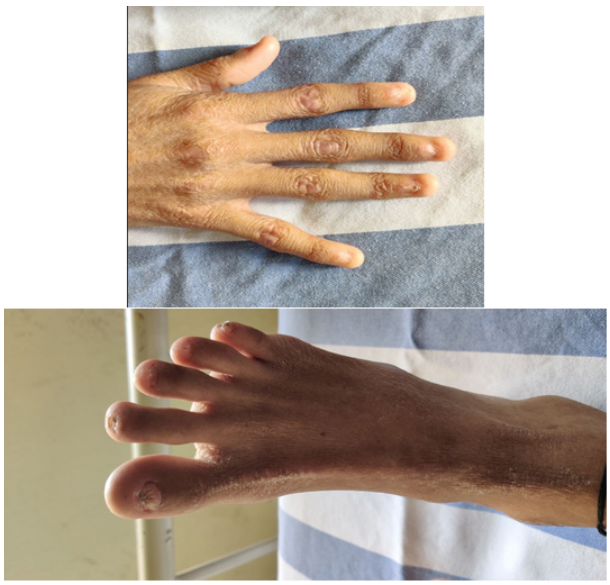

Figure 1 Reticular skin changes and Nail changes of Dyskeratosis Congenita.

Case Report

A child aged 16yrs came with complaints of pain in throat since 1 month. History of weight loss – non-quantifiable, history of decreased intake of food. Patient also gives history of change in voice since 1 month. Complaints of noisy breathing while eating. Patient is the first born of second degree consanguineous marriage. No known medical co-morbidities. Patient has no significant family or personal history. Clinical examination from Head to toe shows abnormal nails, reticular skin pigmentation all over the body and diagnosed with typical condition known as **Dyskeratosis congenita**. Right neck examination shown 6 X 7 cm node in mid and lower jugular region which is matted, tenderness present and examination of Left neck is normal. On Video direct laryngoscopy shows lesion arising from Pyriform fossa B/L Aryepiglottic folds more likely Malignancy arising from? Hypopharynx.

Diagnostic Assessment

USG Neck shows Evidence of heterogeneously hyperechoic conglomerate matted mass lesion measuring 2.5 X 4.5 X 2.9cm and approximately 0.4cm deep to the skin surface noted in level II & III on right side. Another heterogeneous hyperechoic conglomerate matted mass lesion measuring 1.4cm X 1.3cm and approximately 0.6cm from skin surface noted in level III on left side. FNAC from right side of neck shows Metastatic Squamous Cell carcinoma. CECT Scan was done and shown heterogenously enhancing lesion measuring 3.1 x 2.8 x 7.7 cms involving B/L aryepiglottic folds, pyriform fossa and extending to cervical oesophagus and posterior pharyngeal wall-suggestive of growth. Multiple Ill Defined hypodense lymph nodes in B/L Ib, II and right III and IV ,largest measuring 3.4 x 2.4cms in the right level II. Patient was then sent for Biopsy from primary site which was reported as Well differentiated squamous cell carcinoma from the primary site PET CT was done which showed Increased FDG uptake is noted in the enhancing soft tissue mass involving the hypopharynx and cervical oesophagus (site of known primary), measuring 2.9 x 2.2 x 5.1 cm. It is extending superiorly to involve bilateral pyriform fossae and bilateral aryepiglottic folds. Increased FDG uptake is noted in the enlarged conglomerated right level II, III cervical nodal Masses, measuring 2.5 x 3.2 x 4.8 cm, Increased FDG uptake is noted in discrete right level IV, left retropharyngeal and level II cervical nodes, largest measuring 1 x 1.1 cm, SUV max 4.24 in right level IV node. No evidence of metabolically active disease elsewhere in the body (**Figure 2**).

Therapeutic intervention

After giving proper counselling about the disease & treatment, patient underwent Percutaneous Endoscopic Gastrostomy under short General Anaesthesia as patient had swallowing difficulty and then started on Definitive Radiation therapy to the Gross tumour involving hypo pharyngeal region and involved gross nodes. Patient was planned on 6 MV Linear Accelerator with Intensity Modulated Radiation Therapy with a dose of 70Gy (Gray) in 35#(Fractions)over 6.5 weeks. Patient tolerated the Radiation Therapy well. Chemotherapy was withheld in view of Age and Dyskeratosis Congenital. Reactions and Side effects were managed effectively during course of treatment.

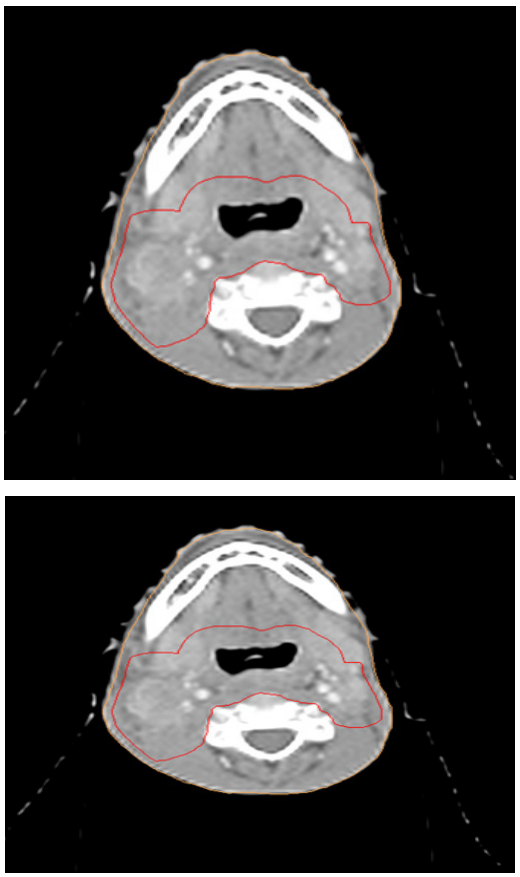

Figure 2 CT Scan showing Gross Hypopharynx disease and Gross involved Nodes.

Results

Patient underwent complete course of Radiation therapy. Reactions and side effects were managed symptomatically. Patient was asked to review after 1 month for follow up. On 1 month follow up patient complained of odynophagia and palpable cervical lymph nodes. Patient is referred to ENT department for Video Direct Laryngoscopy and it showed no abnormal or residual disease and USG Neck shown conglomerate lymph node mass with loss of fatty hilum of size 4x2.7x1 cms noted in Right Level II, 2.2X 1.2X0.5cms in Left Level II, 0.4X0.4mm node in Right Level IV. Patient has been put on Antibiotic coverage and asked to review for 6 month follow up. On 6 month follow up, patient came with complaints of Difficulty in swallowing and multiple palpable cervical lymph nodes and on examination there was a growth arising from the stroma site of Feeding tube. Patient was advised for PET-CT for whole body evaluation and shown Increased FDG uptake in the soft tissue mass involving the hypopharynx and cervical oesophagus, measuring 2.5 x 1.3 x 4 cm, SUV max 7.08, with infiltration into the right lobe of thyroid. Low grade FDG uptake is noted in partly calcified right level II, III cervical nodes, largest measuring 1.2 x 2.2 cm. Faint FDG uptake is noted in left level II cervical node measuring 5 x 9 mm, Non-FDG avid 3 mm sized left retropharyngeal node is seen. Low grade FDG uptake is noted in sub cm sized right supraclavicular nodes. Increased FDG uptake is noted in the large citatorylesion involving the

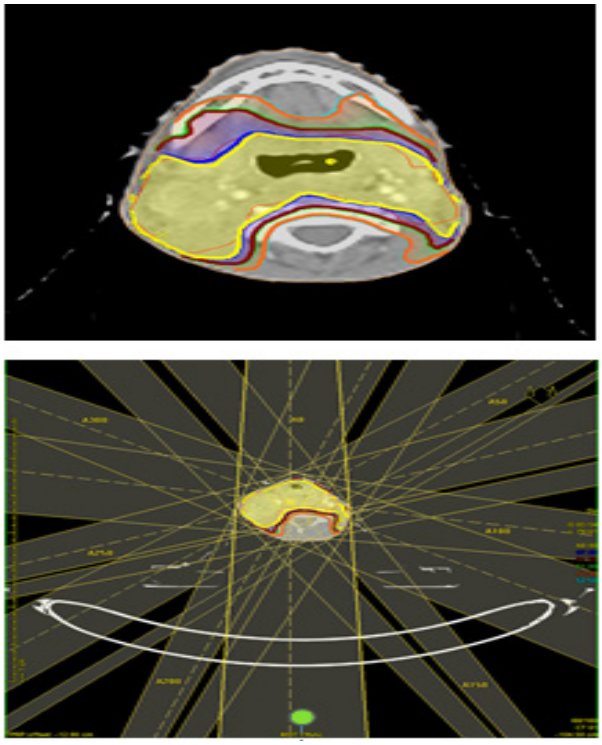

Figure 3 Dose Distribution and Beam placement for Radiation therapy

lower lobe of left lung. Increased FOG uptake is noted in the circumferential wall thickening involving the GE junction with perigastric extension. Discrete smallerHypermetabolic soft tissue deposit beneath the anterior abdominal wall, superior to the above mentioned mass, is a new finding - likely to be metastatic. Hypermetabolic peritoneal deposit abutting the right lateral upper rectal wall is a new finding - likely to be metastatic (**Figure 3**). Overall PET CT Shows Residual disease and whole body metastasis. Opinion was sought from Medical oncologist and surgical oncologist regarding and need for further management. But unfortunately due to advanced disease and disease progression patient couldn't survive.

Discussion

Dyskeratosis congenita (DC) is a rare multisystem bone marrow failure syndrome that displays marked clinical and genetic heterogeneity. X-linked recessive, autosomal dominant and autosomal recessive forms of the disease are recognized. The gene that is mutated in the X-linked form of the disease is DKC1. The DKC1 -encoded protein, dyskerin, is a component of small nucleolar ribonucleoprotein particles, which are important in ribosomal RNA processing, and of the telomerase complex. The autosomal dominant form of DC is due to mutations in the gene for the RNA component of telomerase [4]. The link between Dyskeratosis Congenita and cancer is particularly intriguing, because DC is associated with defects in telomere biology. Patients with Dyskeratosis Congenita have very short telomeres and mutations have been identified in telomere biology genes. However, to date there has been no comprehensive quantitative analysis of cancer risk in Dyskeratosis Congenita. There is increased risk for squamous cell carcinoma and hematolymphoid neoplasms. Approximately 90% of patients exhibit nail dystrophy, which affects the fingernails first and then the toenails in most cases. Mucosal leukoplakia is a pathognomonic feature and occurs in approximately 80% of patients. In childhood, Bone Marrow Failure is the most frequent complication of Dyskeratosis Congenita, whereas pulmonary fibrosis is a frequent cause of mortality in adults. The skeletal, gastrointestinal, and genitourinary systems also may be affected. DC is a multisystem disorder, so it is important to monitor many systems of the body [5]. A lot of studies and literature is there regarding treatment of Dyskeratosis congenital. Since our patient is a case of malignancy, multimodality management is required which involves Radiation Oncology, Hematology, Medical oncology and Otorhinolaryngology. Proper counselling and multidisciplinary team approach is essential to treat a case of malignancy especially in Dyskeratosis congenita.

Conclusion

Dyskeratosis congenital is a rare congenital disorder. Bone marrow failure is the most common cause of death in children. Malignancy is a troublesome problem in patients suffering from Dyskeratosis congenital. Since it is a congenital disorder, it is advised that all siblings of affected patient should undergo Genetic screening and parents should be counselled for the same and the need for early management.

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