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FANCONI SYNDROME IN FEBRILE NEUTROPENIC CARCINOMA PATIENT:A CASE REPORT

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Abstract

Fanconi syndrome is associated with the impairment of proximal tubular reabsorption, which leads to Renal Tubular Acidosis(RTA) characterised by increased excretion of electrolytes. Febrile neutropenia(FN) is a condition commonly seen in chemotherapy patients, with the signs of fever and low Absolute Neutrophil Count(ANC).A 72 year old male patient was admitted with the complaints of fever, breathlessness for the past two days, with one episode of haemoptysis. It is a known case of carcinoma lung® undergoing chemotherapy in other hospital. He had a temperature of 100°F.Supportive measures were taken. The haemogram showed Total Count(TC) as 2400cells/mm3, 8.1g% of Haemoglobin and 126mmol/L of sodium. Managed with 3%NS(100ml) and blood transfusion .On third day,ANC was 700cells/cumm, TC decreased to 1200cells/mm3 and CRP was 266.07mg/L, which indicates FN. Managed with Filgrastim(300mcg/0.5ml) along with Piperacillin-Tazobactam 4.5gQ6H and Gentamycin-80mgBD.Then, the patient had massive haemoptysis, managed with Tranexamic acid(100mg/5mlQ8H) .Later, serum potassium was found to be consistently low(3.6-2.1mmol/L),but improved to 3.7mmol/L with intensive potassium replacement therapy(20.25g in ten days).On the day of discharge, the levels of magnesium and bicarbonate was found to be 0.6mg/dl and 18mEq/L respectively. Thus, the diagnosis of Aminoglycoside induced Fanconi syndrome and Type 2 RTA was made. Gentamycin was withdrawn, supplementations were given and the patient got discharged for the next cycle of chemotherapy. The prevalence of FN condition is 17.5% in one lakh population. Patients undergoing chemotherapy are at high risk of electrolyte imbalance and haemogram variations. Therefore, careful monitoring is required for oncology patients with complications.

Keywords: Febrile neutropenia, Fanconi Syndrome

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A CASE REPORT ON PAEDIATRIC ANAPLASTIC ASTROCYTOMA WITH PIK3CA GENE MUTATION

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Abstract

Anaplastic astrocytoma is a rare malignant, diffusely infiltrating, astrocytic, brain tumour categorised under the high grade gliomas (WHO grade III-IV).A 12 year old male patient who is a known case of anaplastic astrocytoma, WHO GRADE III,1p/19q non-codeleted (diagnosed 07/03/2019) was admitted with complaints of fever, generalised weakness and excessive sleepiness. He was treated previously with chemotherapeutic drugs temozolomide, irinotecan, bevacizumab and PCV regimen but no progress was obtained and hence discontinued. The patient was given IV fluids, antibiotics, antiedema and anti-seizure medicines and other supportive measures. Tumour genomic DNA and RNA isolated from FFPE was used to perform targeted gene capture and clinically relevant mutation was identified in the PIK3CA gene of the subject on 20/01/2020. A missense variation that results in the amino acid substitution at codon 542 was detected in the PIK3CA gene of this subject. An FDA approved treatment regimen including a targeted PIK3CA inhibitor is available for specific subtypes of breast cancer. While PIK3CA variants are highly prevalent in many other cancer types, targeted therapies for PIK3CA variants are still in early clinical trial phases in other cancer types. However, the role of the variant identified in the subject is not very well documented in the medical literature for the tumour type in the subject.

Keywords: Anaplastic astrocytoma, gliomas, PIK3CA gene mutation.

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