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MYELODYSPLASIA WITH 20Q DELETION – A CASE REPORT OF UNUSUAL AGE AT PRESENTATION

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Abstract:

Myelodysplasia refers to a group of clonal stem cell disorders involving impaired hematopoiesis and persistent peripheral blood cytopenias, with an increased tendency of progressing to acute leukemias. The symptoms present according to the cell line affected i.e. fatigue and weakness from anemia, infections from neutropenia and bleeding from thrombocytopenia. We discuss the case of a 16-year-old female who presented with a 3-month history of fever, abdominal pain, shortness of breath and bleeding from gums.

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INTRODUCTION:

Myelodysplasia refers to a group of clonal stem cell disorders involving impaired hematopoiesis and persistent peripheral blood cytopenias, with an increased tendency of progressing to acute leukemias. The symptoms present according to the cell line affected i.e. fatigue and weakness from anemia, infections from neutropenia and bleeding from thrombocytopenia [1]. It has an incidence of 4.9 per 100,000 people per year [2]. Myelodysplasia shows an increased tendency in males as compared to females, is rare in adolescents and those under 40 years of age [3]. The median age of its onset is 71 years [4]. Diagnosis is based on qualitative and quantitative evaluation of the peripheral blood smear as well as bone marrow investigations [5]. The Revised International Prognostic Scoring System (IPSS-R) is a validated standard for assessing prognosis and divides patients into four risk groups stratified to predict prognosis, and median survival time [6]. Here, we discuss the case of a 16 year old female who presented with a 3 month history of fever, abdominal pain, shortness of breath and bleeding from gums.

CASE REPORT:

A 16-year-old female was admitted in the Medical Unit of Liaquat University Hospital, Jamshoro with the 3-month history of fever, abdominal pain, shortness of breath and bleeding from the gums. The patient had an unremarkable birth history but contracted meningitis as an infant, as a consequence of which she had developed Spastic Monoplegic Cerebral Palsy (with right lower limb involvement). The patient was intellectually impaired with an IQ level of 35%, non-trainable and had not achieved menarche. She had 2 deceased siblings who were also intellectually impaired and had an unknown blood disorder for which they had required repeated blood transfusions and had passed away at the ages of 7 and 12 years, respectively. Her past history was significant for having recurrent episodes of shortness of breath, productive cough and sore throat, nausea, and burning micturition. The patient was transfused 3 pints blood 1 month prior to presentation; no records were available.

Physical examination:

The girl appeared visibly stunted, pale and dyspneic with hepatomegaly but no splenomegaly or lymphadenopathy. The patient was febrile but otherwise vitally stable. She had thin depigmented hair, angular stomatitis, seeping gums and nostrils, with fungal infection in her nails. There was bruising noted over her arms, elevated JVP, bilaterally swollen ankles, and positive sternal tenderness. She had a visible, displaced apex beat in the 5^{th} intercostal space, 13 cm away from the midsternal point. An ejection systolic murmur was audible in all 4 cardiac areas (Grade 3/6), non-radiating. Fine inspiratory crackles were auscultated at the lung bases otherwise normal vesicular breathing was noted. She had no jaundice, clubbing, peripheral cyanosis, or lymphadenopathy.

Laboratory Investigations:

Her complete blood count revealed an Hb=1.9 g/dL, with an MCV=101.8 F, WBCs= 1.26, and Platelets=14,000. Her reticulocyte count was 0.5%, serum folate and LDH levels were normal while her cobolamine levels were low normal. The peripheral blood smear demonstrated anisocytosis, NRBCS and few atypical mononuclear cells. Her PT/aPTT/INR, Urea/Creatinine and Electrolytes, anti- TTG IgA and liver enzymes were unremarkable, while her serum albumin was 3.3 (Total protein=6.3). Her urine DR and stool DR were normal. Her ECG only showed Sinus Tachycardia. Based on her peripheral smear findings, her bone marrow aspirate was done which showed ineffective erythropoiesis and dysmyelopoeisis with increased ME ratio 3:1 and 8-10% blast cells. Consequently, her bone marrow biopsy was performed which revealed replacement of normal bone marrow structure, myeloid hyperplasia with maturation arrest, and dysmegakaryopoeisis.

Diagnosis:

A conclusive diagnosis of MDS RAEB (MDS, Refractory Anemia with Excess Blasts) was made. Chromosomal analysis was then carried out which revealed 20q deletion.

Management:

The patient was kept in isolation with O_2 inhalation, I/V furosemide, and transfused with 3 units of packed RBCs and 2 mega units of platelets as part of clinical management. As adjunct to her therapy, symptomatic treatment with broad spectrum antibiotics and oral antifungals was started and she was supplemented with Vitamin A, Methylcobolamine and Folic acid. Following stabilization of symptoms, the patient was

referred to the Hospital's Oncology Department for further evaluation and management.

DISCUSSION:

Myelodysplasia refers to a group of clonal stem cell disorders involving persistent peripheral blood cytopenias. At present, there are no recent studies that describe the occurrence of this spectrum in Pakistan. Our patient presented with symptomatic bleeding phenomena as well as signs of anemic failure. Features that make this case stand out are firstly, the unusually young age at presentation, as well as the fact that 20q deletions are rare in this age group [(3)]. We evaluated our patient based on the IPSS-R scoring system [6]and her cumulative score was 4.5 (1 for Cytogenetics -20q deletion, 2 for 8-10% bone marrow blasts, 1 for hemoglobin <8, and 1 for platelets <50,000). This gave her a risk category of "Intermediate" prognosis and a median survival rate of 3.02 years. Other treatment options of a supportive nature that were viable for our patient include Erythropoietin, Filgastrim, and Thrombopoeitin receptor agonist. Specific therapy involves epigenetic therapy, conventional chemotherapy, immunomodulatory therapy, immunosuppressive therapy, allogenic bone marrow transplantation and reduced intensity bone marrow transplantation [5]. Research is still ongoing on Myelodysplasia and its multiple variants, and the classification system continues to be scrutinized and adapted as per clinical evidence. It is imperative that cases be carefully monitored for a drop in cell lines and managed accordingly to reduce patient morbidity and mortality.

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