SUCCESSFUL TREATMENT OF THREE DIFFERENT CONGENITAL HEMATOLOGICAL DISEASES WITH HEMATOPOIETIC STEM CELL TRANSPLANTATION

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Introduction: Congenital dyserithropoetic anemia and hereditary dehydrated stomatosis are causes of congenital hemolytic anemia and the main treatment approach is transfusion, which can lead to iron accumulation. It could be seen from the asymptomatic form to severe hemolytic crisis. Usually, it is characterized with mild jaundice and compensatory hemolytic anemia. Glanzmann thrombasthenia is a thrombocyte aggregation disorder, which is responsible for mucosal bleeding since early childhood. Thrombocyte suspension replacement, the uses of activated factor VII preparate and antifibrinolytic agents are the main treatment approaches. Hematopoietic stem cell transplantation (HSCT) is the main curative treatment choice both for benign and malign diseases currently. Case presentation:15-year-old girl whose parents are first degree cousins admitted to our hospital with easy bruising, epistaxis and concomitant anemia which required erytrocyte and thrombocyte transfusions. She was followed as congenital hemolytic anemia and Glanzmann thrombastenia according to flow cytometric analysis and hemoglobin electrophoresis. With high suspicion of congenital bone marrow failure syndrome, her Whole exome sequencing analysis (WES) was done, three different mutations were found: Homozygous mutation in ITGA2B gene (which was responsible for Glanzmann thrombasthenia), heterozygous mutation in PIEZO1 gene (which was responsible for dehydrated stomatosis) and homozygous mutation in SEC23B gene (which was responsible for congenital dyserytropoietic anemi Type II).After the conditioning regimen consisting of busulphan, cyclophosphomide and ATG, allogenic stem cell transplantation was done from fully matched sister. She had neutrophil engraftment on 15th day and thrombocyte engraftment on 28th day. She was discharged successfully at 35th day. After two months, she developed Grade 4 gastrointestinal system GVHD.Tacrolimus and metil prednisolone was started, mesencymal stem cell was given to improve the microenvironment in guts. At 100th day of transplantation, she discharged from hospital. After eight months of transplantation, she is now in remission with no complaints.
Conclusions: Hematopoetic stem cell transplantation is the main curative treatment both for benign and malign diseases currently. It is crucial to take a detailed disease and family history and perform WES analysis punctually, so that eligible patients can reach to treatments and transplantation successfully.