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Broken and Made Whole: A Rare Case of Pediatric Myelodysplastic Syndrome with Isolated 5q Deletion Treated with Lenalidomide

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INTRODUCTION

Most Myelodysplastic Syndrome (MDS) with transfusion- dependent anemia is associated with the deletion of 5q cytogenetic abnormality. Variable degrees of cytopenia are presented in this syndrome and there is an increased risk for developing acute leukemia, specifically Acute Myelogenous Leukemia. According to studies, Lenalidomide can decrease transfusion requirements in MDS. This case report aims to identify and prove the effects of this promising drug in children with MDS.

CASE SUMMARY

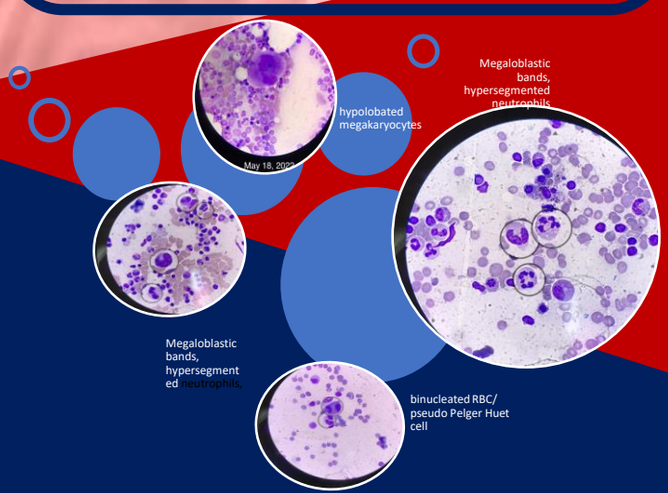
An 8 year old male initially presented with gum bleeding and pancytopenia. He was initially diagnosed with Aplastic Anemia, moderate. He was transfusion dependent with pRBC and platelet concentrate. BMA monitoring was done which showed trilineage dysplasia. Myelodysplastic Syndrome was considered and further work-up revealed isolated 5q31 deletion in EGRI probe of MDS FISH (Fluorescence In Situ Hybridization) Panel Assay. He was started with Lenalidomide 10mg/ dl daily and was monitored and observed closely every week wherein he showed rapid improvement based on hematologic and cytogenetic response to the drug. Further improvement was seen as early as post 2 cycles of Lenalidomide. He became transfusion independent with stable levels of hemoglobin and platelet count. The patient is still on treatment and remains to be asymptomatic.

DISCUSSION

There are limited reported case on the use of Lenalidomide in pediatric MDS. The treatment goals for MDS patients are to improve peripheral blood values (increase hemoglobin levels and to reduce bleeding and infections) and to change the natural progression of the disease. Lenalidomide is a thalidomide analog and an immunomodulatory drug. It is FDA approved and widely used in adult patients with Multiple Myeloma. In MDS, it suppresses clonal hematopoietic cells via synthetic lethality mediated by the cereblon-dependent degradation of haplodeficient proteins encoded within the deleted 5q region. It also has indirect immunomodulatory effects by increasing T- Cell and natural killer cell activation and decreased pro-inflammatory cytokine production. The recommended dosing of Lenalidomide was 10mg once daily for 21 consecutive days, with rest for 7 days (1 cycle = 28 days). Dose interruption nor reduction was not done due to good response of the drug in the index patient. The study of Pisani stated that the current treatment recommendation is to treat del(5q) with Lenalidomide until relapse or transfusion dependence or progressive disease (7). Consequences of the long- term effects of continuous treatment is unknown.

CONCLUSION

The patient presented in this case achieved both complete hematologic and complete cytogenetic remission after 2 cycles of Lenalidomide and was characterized by blood transfusion independence and disappearance of 5q deletion respectively. The duration of treatment is yet to be discovered but different journals suggests to give Lenalidomide indefinitely with modification if with complications and drug holiday to avoid drug resistance, improve tolerability enabling patients to stay on therapy for longer and thus optimizing clinical benefit.



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