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Early Bone Marrow Transplantation in GATA2 Deficiency-Induced MDS

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INTRODUCTION

GATA2 deficiency is considered one of the most common cancer predisposition syndromes determining myelodysplastic syndrome (MDS) in children. GATA2 deficiency clinical presentation with hematological changes can be suggestive of GATA2-related myelodysplastic syndrome

AIM

Early genetic and complete hematological investigations before serious complication or progression in advanced MDS or AML.

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METHOD

In this abstract we describe a case with GATA2 related MDS based on early clinical manifestation, diagnosis, treatment, and outcome.

RESULTS

Patient at the age of 3 was diagnosed with decrease in hearing, third degree. At age of 5 developed episodes of severe ulcerative stomatitis. In the last year presented with multiple subcutaneous haemorrhages on the lower limbs and buttocks. Thrombocytes were normal, and the patient was diagnosed with necrotic vasculitis. In May of 2021 blood count showed macrocytic anemia, leukopenia, neutropenia. Based on bone marrow (BM) aspiration morphology showed hypocellular bone marrow without blast cells, and cytogenetic investigation detected monosomy 7. Final hematological diagnosis was defined as MDS EB with monosomy 7. Due to the clinical phenotype of GATA2 deficiency, genetic tests were done which detected pathological variant of GATA2 gene. Tests detected one pathogenic c.1126_1143+54 del at site and one possibly pathogenic c.1192C>G, p.Arg398Gly mutations in GATA2, INVITAE04/22.
October 27, 2022, patient received transplant from MUD 9/10 donor, condition by Bu/Cy/ATG. GvHD prophylaxis with cyclosporin/MTX. Neutrophils engraftment at day +14. Full donor chimerism at day +19 -28.

CONCLUSIONS

Clinical and hematological phenotype in complex can be suggestive of GATA2 deficiency related.

REFERENCES

Prevalence, clinical characteristics, and prognosis of GATA2-related myelodysplastic syndromes in children and adolescents

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