functional roles of DEGs in Myelodysplastic Syndrome (MDS) pathobiology. We categorized the DEGs of CD34+ HSC and PBMC into functional categories using GO analysis. In CD34+ HSC, the most enriched pathways were signal transduction, negative regulation of transcription, and immune response. In PBMC, the immune response, signal transduction, and innate immune response pathways were significantly activated. We also performed KEGG pathway analysis for upregulated and downregulated DEGs in both CD34+ HSC and PBMC samples. In CD34+ HSC, the top pathways were involved in hematopoietic cell lineage, cell receptor signaling, and primary immunodeficiency. These pathways were also identified in PBMC, along with cancer and cell cycle pathways. **Discussion and Conclusion:** To summarize, our study uncovered disparities in DEGs between CD34+ HSC and PBMC cell types in MDS. However, we also observed a certain degree of similarity in the activated pathways of both cell samples based on Gene Ontology and KEGG pathways enrichment analyses. Of particular importance in our study was the role of the immune system and the activation of hematopoietic cell lineage pathways as the most activated in MDS, regardless of the cell type evaluated. These findings provide novel insights into DEGs biomarkers associated with MDS pathogenesis, holding clinical significance.

https://doi.org/10.1016/j.htct.2023.09.805

CASE REPORT: ADVANCED SYSTEMIC MASTOCYTOSIS ASSOCIATED WITH HEMATOLOGICAL NEOPLASIA

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Objective: Report a clinical case of Advanced Systemic Mastocytosis Associated with Hematological Neoplasia (MAS-ANH) under treatment with the AZAVIT-ABCDEF protocol, 7 days every 28 days (azacytidine associated with high doses of vitamins B1, C, and D, with erythropoietin and filgrastim). Brazil Platform CAAE: 53015421.0.0000.5463. After obtaining informed consent. **Methodology:** Assessment of patient's information from HSPE's computer-based patient records. **Clinical case:** A 71-year-old male with fatigue, diarrhea, pruritus, and extensive skin lesions in the groin, lymph node enlargement, and splenomegaly. Hb = 8.2 g/dL, platelets = 64,000/mm^3, and leukocytes = 11.38 thousand/mm^3 (1/5/54/0/0/37/3), Alkaline phosphatase = 239 U/L (30-120 U/L). Myelogram showed dysplastic changes, 4% blasts, and moderate mastocytosis. Bone marrow biopsy revealed 60% cellularity and mastocytosis. Immunohistochemistry: CD117 (+). Lymph node and skin biopsy showed infiltration by mast cells. Bone scintigraphy showed increased uptake in the skull, pelvis, spine, and femurs. Initially treated with alpha interferon, which resulted in increased cytopenias and worsening of constitutional symptoms. Decided to start zoledronic acid and AZAVIT-ABCDEF protocol, receiving 27 cycles every 28 days. Showed improvement in hemogram with sporadic blood component transfusions and reduction in splenomegaly. Pruritus and diarrhea improved significantly, and the patient gained weight. Triptase level still at 145 μg/mL (normal = 11 μg/mL) with slight improvement in skin lesions. Current blood count: Hb = 10.8 g/dL, platelets = 71,000/mm^3, and leukocytes = 9.280/mm^3 (1/5/59/1/0/14/20). **Discussion:** MAS-ANH is a rare entity associated with a mutation in the KIT D816V gene that encodes the receptor for stem cell factor (CD117). Additional mutations are commonly found, such as TET2 (29%), ASXL1 (17%), and CBL (11%). The main manifestations are hematological and gastrointestinal disturbances. Other symptoms include pruritus, urticaria, skin infiltration, bone pain, fatigue, fever, headache, and anaphylactic reactions. A prognostic index showed worse outcomes for cases with age > 60 years, thrombocytopenia, anemia, elevated alkaline phosphatase, and the presence of adverse mutations (ASXL1, RUNX1, and NRAS). Treatment options include KIT gene inhibitors (midostaurin, imatinib, and avapritinib), alpha interferon, and 2-CDA, with short-lived responses and the possibility of worsening cytopenias. Antihistamines and corticosteroids are used symptomatically. The good hematological response observed in this case probably occurred due to the presence of hematological clonal abnormalities resembling myelodysplastic neoplasia without mastocytosis. We believe that the protocol promotes differentiation by increasing the transcription of factors related to cell differentiation and improving aerobic metabolism. **Conclusion:** MAS-ANH is an aggressive pathology with a median survival of 20 months, even with the use of KIT gene loop inhibitors. At diagnosis, the patient presented with 4 out of 5 poor prognostic features and has already achieved a survival of 41 months. Further studies will be required to confirm the improvement in quality of life and survival with the AZAVIT-ABCDEF protocol.

https://doi.org/10.1016/j.htct.2023.09.806

PLATELET DYSFUNCTION IN A PATIENT WITH CHRONIC MYELOMONOCYTIC LEUKEMIA (CMML) AND RECURRENT SUBDURAL HEMATOMA.

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Objectives: To report a case of a patient diagnosed with CMML with recurrent subdural hematomas and platelet dysfunction. **Materials and methods:** The case report was based on data recorded in the electronic medical record and literature review. **Results:** A 62-year-old female diagnosed with CMML in February 2022 during investigation of persistent monocytosis associated with anemia and thrombocytopenia. Hemoglobin: 8.9 g/dL; White blood cell count: 11,860/mm^3 (69/0/0/9/20); Platelets: 45,000/mm^3. BCR-ABL and JAK2 tests were negative. Bone marrow examination revealed erythroid and granulocytic hypercellularity with dysplastic changes.