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LEUKEMIA STEM CELLS AND MEASURABLE RESIDUAL DISEASE IN PATIENTS WITH ACUTE MYELOID LEUKEMIA UNDERGOING ALLOGENEIC BONE MARROW TRANSPLANTATION

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Introduction: Measurable residual disease (MRD) is predictive of relapse in acute myeloid leukemia (AML), including in the pre-stem cell transplant (SCT) setting. The presence of leukemia stem cells (LSCs) has been shown as another predictor of outcome in addition to MRD status. **Aim:** this study aims to assess the presence of LSC in association with MRD results and its impact on the survival of AML patients undergoing SCT. **Material and methods:** Patients, material and methods: One hundred and eleven patients with AML were retrospectively evaluated before SCT and 89 after SCT. The median age was 37 years (5-63), 47% were female, and 82 (91%) had de novo AML. The 2022 ELN risk classification was: 32% adverse, 28% intermediate, 4% favourable, and 36% with missing data. Clinical remission status was: CR1(52%), CR2(31%), $\geq$ CR3 (17%). The conditioning regimen were myeloablative (MAC) (59%) and reduced intensity (RIC)(41%). MRD was assessed before and after SCT using an analytically and clinically validated 10-color panel that combined backbone markers (CD33, CD34, CD38, CD 45, CD117 and HLA-DR) with markers such as CD2, CD7, CD11b, CD13, CD14, CD15, CD19, CD36, CD56, CD123, CD300e. LSC immunophenotypes (CD34+/CD38-) were identified using markers such as CD45RA, CD54, CD96, CD97, CD99, CD244, CLL1/TIM3. Samples were acquired on a BDFAC-Slyric flow cytometer and Infinicyt (Cytognos)™ was used for flow data analysis. At least 1 million events were evaluated per tube to achieve adequate test sensitivity. R-software v 4.1 was used for statistical analysis. The Kaplan-Meier method was used to assess overall survival (OS) and relapse-free survival (RFS). The log-rank test was used to compare the survival distributions of two or more groups. **Results:** In the pre-SCT setting, the 2-year OS and RFS of the patients were, respectively, 89% and 91% for the MRD-/LSC- group (n=44); 66% and 66% for MRD-/LSC+ (n=3); 63% and 53% for MRD+/LSC- (n=49); 42% and 45% for MRD+/LSC+ (n=15) (p < 0.001 and p < 0.005 respectively). In the post-SCT setting, OS and RFS at 2 years were, respectively, 90% and 92% for MRD-/LSC- (n=37); 100% and 89% for MRD-/LSC+ (n=9); 75.2% and 66% for MRD+/LSC- (n=36); 57% and 57% for MRD+/LSC+ (n=7) (p=0.001 and p=0.007 respectively). **Discussion and**

conclusion: Patients with MRD-/LSC- had significantly longer OS and PFS than the other groups both pre- (p=0.001) and post-SCT(p=0.009). MRD+/LSC+ characterized the group with the worst outcome. However, the MRD+/LSC- and MRD-/LSC+ groups presented intermediate overall survival and progression-free survival compared to the other groups, and presented similar outcomes both pre- and post-SCT. A shorter OS was observed in MRD+/LSC+ patients compared to MRD+/LSC- patients pre-SCT (p=0.02), but this analysis was hampered by the small number of patients in this group (n=3). In the MRD+ patient groups, OS and RFS were higher in the LSC-group than in the LSC+ group (p=0.009 and p=0.024 respectively) in the post-SCT setting. These findings show the relevance of the presence of LSC in the bone marrow of patients with AML and are in agreement with some data in the literature. Although the number of AML patients and LSC+ in this cohort is small to allow a more robust evaluation, these preliminary data contributed to highlight the importance of detecting LSC in the follow-up of these patients.

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LIGASE IV SYNDROME IN A 7-YEAR-OLD BOY: CLINICAL, MOLECULAR, AND TRANSPLANTATION ASPECTS OF A RARE IMMUNODEFICIENCY DISORDER

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Introduction: Ligase IV Syndrome (LIG4) is an extremely rare autosomal recessive disorder arising from mutations in the LIG4 gene, which encodes DNA ligase IV, a crucial component of the non-homologous end-joining (NHEJ) pathway responsible for repairing double-strand breaks (DSBs) in DNA. **Case report:** The patient is a male child, born on July 11, 2017, diagnosed with constitutional severe aplastic anemia secondary to a LIG4 gene mutation, confirmed through genetic testing. This rare autosomal recessive condition is classified under ICD D61.0 and is associated with defective DNA repair mechanisms and immunodeficiency disorders. Over recent months, he has required monthly transfusions of red blood cells and platelets and exhibited persistent severe neutropenia (< 500 neutrophils/ μ L), increasing his susceptibility to opportunistic and severe infections. The clinical and laboratory findings, consistent with bone marrow failure, led to the recommendation of allogeneic hematopoietic stem cell transplantation (HSCT), the only potentially curative treatment in LIG4 deficiency. Between January and February 2025, the patient was admitted several times to Hospital Infantil Gonzaga for neutropenia-associated bacterial infections and transfusional support. Episodes of febrile neutropenia were managed with broad-spectrum antibiotics, including intravenous vancomycin and meropenem, with good clinical response. No allergies