A 17-year-old patient was diagnosed with severe aplastic anemia after a history of fatigue, mucocutaneous bleeding and pancytopenia. Bone marrow (BM) aspirate was hypocellular and had no dysplastic changes. Trephine biopsy confirmed 5% cellularity and absence of increased immature progenitors; reticulin fibrosis was not observed. Conventional cytogenetics showed a 46, XY karyotype; a small paroxysmal nocturnal hemoglobinuria (PNH) clone was present in red cells, neutrophils and monocytes. Chromosome breakage analysis for Fanconi anemia was unremarkable, and telomere length was within the normal range. The patient had no HLA-matched donor, and once hATG was discontinued in Brazil and eltrombopag was unavailable, he was included in a group of patients treated with upfront immunosuppressive therapy (IST) with subcutaneous alemtuzumab and cyclosporine; The patient achieved a partial response after 3 months of treatment. Cyclosporine was gradually tapered and was completely stopped after a year of treatment. This patient was lost to follow-up visits for six months and returned after four years of IST, complaining about worsening of fatigue and gingival bleeding. Complete blood count was notable for a loss of the achieved PR, and the presence of a small population of dysplastic monocytes. Revaluation of BM showed a hypercellular smear, and the presence of 60% of immature myeloid cells composed by myeloblasts and monocytes precursors. BM immunophenotyping was compatible with acute myelomonocytic leukemia. Conventional cytogenetics detected a monosomy of chromosome 7 and a trisomy of chromosome 21. Molecular studies for FLT3 and NPM1 mutations, and RUNX1-RUNX1T1 and CBFB-MYH11 fusion genes were negative. The PNH clone size remained stable. The patient was started on standard AML chemotherapy with cytarabine and daunorubicin. An hematologic complete remission with a negative minimal residual disease was achieved. Due to an acute appendicitis during AML induction, the patient awaits consolidation therapy with an haploidentical hematopoietic stem-cell transplant (HSCT) from his brother. **Discussion:** Frontline therapy for severe aplastic anemia patients younger than 40 years who have an HLA-matched sibling donor is HSCT. Triple IST with eltrombopag, hATG and cyclosporine is now considered the standard frontline therapy for patients older than 40 years and those who lack a suitable HLA-matched donor. Once hATG was discontinued in many countries, previous studies have reported outcomes of replacing hATG with rabbit ATG, cyclophosphamide, or alemtuzumab. Clonal evolution to myeloid neoplasms is a recognized complication of IST and patients with AA may develop secondary MDS/AML, with the main associated factors being longer disease duration, presence of mutations with an adverse prognosis, a short telomeric length and the development of cytogenetic abnormalities (CAs). The most recurrent CAs include total or partial loss of chromosome 7, trisomy 8 and 13q deletion, and, less frequently, trisomy 6, trisomy 15 and trisomy 21. **Conclusion:** T-cell-mediated autoimmune diseases, such as AA, have a higher risk of progression to AML/MDS dur to development of somatic mutations and CAs. Monosomy 7 independently represents a poor prognostic factor due to the high rate of progression to acute leukemia, such as occurred with the patient reported.

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FISIOPATOLOGIA DOS HAPLÓTIPOS BANTU E BENIM EM CRIANÇAS COM ANEMIA FALCIFORME

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Objetivos: A doença falciforme (DF) é caracterizada pela presença da hemoglobina S e pode apresentar diversas manifestações clínicas no decorrer da vida do paciente. Dentre estas, podemos destacar a síndrome torácica aguda (STA) e as crises de dor. Desta forma, esse trabalho objetivou fazer uma análise da ocorrência da STA e da frequência das crises de dor com relação ao haplótipo em crianças de 5 a 10 anos. **Material e métodos:** Foram avaliadas 58 crianças, com idade entre 5 e 10 anos com diagnóstico confirmado de anemia falciforme (genótipo SS). Destes indivíduos, 33 foram classificados como Bantu/Bantu, 15 como Bantu/Benim e os outros 10 em outros haplótipos, como Benim/Benim e Benim/Camarões. Foram avaliados dentro de cada grupo a quantidade de crises de dor no último ano antes da coleta e, também, se apresentavam STA. **Resultados:** Dos indivíduos classificados como Bantu/Bantu, 57% apresentavam STA, 72% apresentaram de 0 a 2 crises de dor no último ano, 21% tiveram de 3 a 5 crises de dor e 7% relataram mais de 6 crises de dor no ano corrente. Já com relação aos pacientes Bantu/Benim, 53% relataram STA, 73% apresentaram de 0 a 2 crises de dor e 27% apresentaram de 3 a 5 crises de dor no ano anterior à coleta. **Discussão:** A anemia falciforme é caracterizada pela variação da sua fisiopatologia, sendo que a síndrome torácica aguda e as crises de dor são alguns dos sintomas mais frequentes. A literatura mostra que os indivíduos com haplótipo Bantu/Bantu apresentam as manifestações clínicas mais graves. Nosso estudo demonstra que a ocorrência de STA nos dois haplótipos foi semelhante e, também, a frequência de 0 a 2 crises de dor no ano. Contudo, pode-se destacar que apenas nos indivíduos com haplótipo Bantu/Bantu houveram relatos de mais de 6 crises de dor. Cabe ressaltar que, tratando-se de crianças, as concentrações de hemoglobina fetal no sangue ainda são maiores ($9 \pm 5{,}98\%$) do que os valores de referência (0 a 1%). A hemoglobina fetal tem maior afinidade pelo oxigênio, o que pode atuar em um processo atenuante das manifestações clínicas nestes indivíduos. **Conclusão:** Conclui-se, então, que, com relação à ocorrência da síndrome torácica aguda e das crises de dor não houve diferença significativa nesta amostragem específica.

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