boa resposta à TI em pacientes pediátricos. **Conclusão:** Muito há ainda para se entender sobre a associação de AA e HPN, porém a pesquisa de clone HPN em todos os pacientes pediátricos com AA deve ser realizada ao diagnóstico, podendo predizer maior chance de resposta à imunossupressão.

https://doi.org/10.1016/j.htct.2024.09.1083

IMPACT ON LEUKEMIC TRANSFORMATION OF HYPERDIPLOID KARYOTYPE IN PEDIATRIC MYELODYSPLASTIC SYNDROME

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Introduction: Myelodysplastic syndrome (MDS) comprises a heterogeneous group of clonal hematopoietic stem cell diseases. MDS is characterized by bone marrow dysplasias, peripheral blood cytopenias, and has an increased risk of progression to acute myeloid leukemia (AML). MDS mainly affects elderly patients over 65 years of age and it is rare in childhood. Clonal cytogenetic alterations are found in 30-70% of cases. The most frequent chromosomal abnormalities in pediatric MDS are alterations on chromosome 7 [-7/del(7q)]. Given the importance of karyotype in the prognosis of patients with MDS, it has been incorporated into prognostic risk systems. The most recent is the Revised International Prognostic Scoring System (IPSS-R). Karyotype is classified into five prognostic risk groups according to the IPSS-R: very good, good, intermediate, poor, and very poor. Hyperdiploidy is considered a rare karyotype in pediatric MDS. Therefore, its prognostic impact has not yet been elucidated. **Objective:** To report hyperdiploid karyotypes in children with MDS and their impact on the progression from MDS to AML. **Material and methods:** This study included five pediatric patients with MDS in a cohort of 200 patients. Chromosomal and clinical studies were performed between 2000-2023. Cytogenetic analyses were performed on bone marrow cells by G-banding and fluorescence *in situ* hybridization (FISH) according to the International System for Human Cytogenomic Nomenclature (2020). **Results:** The frequency of hyperdiploid karyotype was 2.5% of all cytogenetically analyzed cases (5/200). This chromosomal pattern was divided into two groups: patients with only chromosomal gains (3/5) and patients with structural alteration (2/5). The structural alterations associated with hyperdiploidy were: dup(1q) (one patient), der(6)del(6)(q21) and der(12)del(12)(p11) (one patient). Most of these patients had advanced subtypes with severe pancytopenia and hypocellular BM. All of these patients were indicated for allogeneic hematopoietic stem cell transplantation (HSCT), but four progressed to AML and three died before HSCT. Only two patients underwent HSCT; the patient with the initial subtype responded well to HSCT and is still alive. The patient with hyperdiploidy and structural alteration had cytogenetic and clinical relapse after HSCT and died. **Discussion:** Hyperdiploidy can be divided into two main subtypes: high hyperdiploidy (51-65 chromosomes) associated with favorable prognosis, and low hyperdiploidy (47-50 chromosomes) related to unfavorable prognosis in acute leukemias of childhood. In MDS, the hyperdiploid karyotype is rare. In this study, we describe 5 cases of hyperdiploid karyotype. According to the IPSS-R, hyperdiploid karyotypes are classified into the group of poor and very poor prognosis (three or more chromosomal alterations). In our study, hyperdiploidy was observed in the initial subtype and advanced subtypes. However, HSCT was successful only in the patient presenting the initial subtype. **Conclusion:** Hyperdiploid karyotype is a rare cytogenetic event in pediatric MDS. This karyotypic pattern was mainly associated with advanced subgroups and progression to AML. Our results suggest that hyperdiploidy in pediatric MDS is a predictive marker of leukemic progression and can be used to indicate these patients for HSCT, the only potentially curative treatment for patients with MDS. **Supported by:** : Fundação de Amparo à Pesquisa do Estado do Rio de Janeiro – FAPERJ (E-26/201.2018/2022).

https://doi.org/10.1016/j.htct.2024.09.1084

OSTEOPETROSE MALIGNA DA INFÂNCIA: RELATO DE CASO

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Introdução: Osteopetrose é um grupo de displasias ósseas raras e hereditárias, caracterizadas pelo aumento da massa óssea. Há diferentes subtipos e pode se manifestar de forma leve a fatal. A osteopetrose maligna da infância possui alta morbimortalidade, com formas graves surgindo logo após o nascimento. **Objetivos:** Descrever características clínicas, radiológicas e laboratoriais de uma paciente com Osteopetrose Maligna e realizar uma revisão bibliográfica não sistemática com base em artigos publicados em banco de dados de periódicos nacionais e internacionais nos últimos dez anos. **Relato do caso:** Lactente de 1 ano e 10 meses, previamente hígida, foi diagnosticada com anemia grave e plaquetopenia leve em exames pré-operatórios para correção de craniossinostose, sendo encaminhada para investigação pela Hematologia Pediátrica. Além da craniossinostose, apresentava esplenomegalia e sangue periférico leucoeritroblástico. O aspirado e a biópsia de medula óssea foram tecnicamente difíceis de serem realizados e imediatamente levantaram a suspeita de osteopetrose, a imunofenotipagem afastou leucemias. O inventário radiológico revelou aumento difuso da densidade óssea, com imagens características de osteopetrose, como vértebras em sanduíche. A tomografia de crânio mostrou hipertensão intracraniana compensada, com sinais de compressão de canal auditivo, órbita e nervo óptico; ainda sem manifestações clínicas, exceto pelo papiledema. Além de anemia, leucocitose e plaquetopenia, hipocalcemia e hiperfosfatemia também foram encontradas. A paciente foi encaminhada para avaliação pelo centro transplantador em cinco meses após o diagnóstico, para ser submetida ao transplante