the right, maintaining contact with the right lateral and anterior chest wall up to the skin; nodular lesions in the bilateral thyroid parenchyma; lesions compatible with lymphoproliferative disease. Abdominal tomography showed a solid mass in the body of the pancreas (5.8 × 5.2 cm), a solid mass in the topography of the left adrenal gland (10.4 × 6.2 cm), solid nodular lesions in the cortex of the bilateral kidneys (up to 3.2cm on the left); all lesions compatible with metastases. Performed on the lymph node conglomerate that diagnosed Burkitt's lymphoma, the patient concluded the stage without bone marrow involvement but with involvement in the cerebrospinal fluid, defining it as stage IV. Chemotherapy treatment was started with good clinical response and images. **Conclusion:** Based on this case and a review of the literature, secondary involvement of the adrenal, pancreas and thyroid does not affect the patient's prognosis like involvement of the cerebrospinal fluid or bone marrow. However, this case characterizes an unusual presentation of Burkitt's lymphoma with involvement of three endocrinological organs simultaneously.

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RESPONSE BY FLOW CYTOMETRY IN PATIENTS WITH BURKITT'S LYMPHOMA AND SECONDARY INVOLVEMENT IN THE CSF

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Introduction: Central nervous system (CNS) involvement is a poor prognostic factor of non-Hodgkin's lymphoma (NHL) in children. Therefore, early diagnosis and treatment of CNS disease is important to improve long-term disease-free survival rates in children. With the continuous progress of science and technology, the methods and technologies used for the diagnosis of CNS disease are constantly improving, from imaging and cerebrospinal fluid (CSF) morphology studies to cerebrospinal fluid CSF flow pattern and genetics studies, and so on. With the continuous progress of diagnosis technology, the clinical staging of patients, which has a vital role in follow-up treatments, has also become more accurate. Most patients with early CNS invasion have no obvious clinical manifestations. Therefore, to improve the diagnostic rate of CNS invasion, flow cytometry (FC) and conventional cytomorphology (CC) of CSF were performed in newly diagnosed NHL patients for the first time of intrathecal chemotherapy. We report the examination findings and flow cytometry (FC) and conventional cytomorphology (CC) of two patients diagnosed with a Burkitt lymphoma and treatment response. **Case Report:** : Case 1: Adolescent, male, 16 years old, with stage IV Burkitt lymphoma. In first evaluation cerebrospinal fluid, absence of atypical cells in cytological examination, immunophenotyping, presence of 38 cells/mL with immunophenotype: CD10+, CD19+, CD20+, CD38+, CD45+, Kappa+, CD34-, Lambda-. In the second CSF, absence of atypical cells and in the immunophenotyping, absence of monoclonal B lymphoid cells. Currently, the patient is 7 months out of treatment in remission. Case 2: Adolescent, male, 17 years old, with stage IV Burkitt lymphoma; in the first evaluation of the CSF with absence of atypical cells on cytology, on immunophenotyping, presence of 40% of anomalous B cells with immunophenotype: CD10+, CD19+, strong CD20, CD34 weak, CD38 strong. In a second CSF evaluation, the patient presented absence of atypical cells on cytological examination and absence of monoclonal B lymphoid cells. Currently, the patient is undergoing treatment with no evidence of CSF involvement. **Conclusion:** Central invasion is more common to be found in pediatric NHL patients with bulky disease and/or BM involvement and/or involvement of more than 4 organs. It was also common in the EBV with Burkitt Lymphoma. CSF detection by FC has important clinical significance in the diagnosis of CNS invasion in children with NHL, as it can significantly improve the detection rate of CNS involvement and evaluation during the treatment.

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COPY NUMBER ALTERATIONS ASSOCIATED WITH HYPERDIPLOIDY IN CHILDHOOD WITH B-CELL ACUTE LYMPHOBLASTIC LEUKEMIA

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Introduction: : The presence of genetic alterations is widely used to stratify B-cell precursor Acute Lymphoblastic Leukemia (BCP-ALL) treatment. The most common cytogenetic abnormality in BCP-ALL is hyperdiploidy (good prognosis) which can be classified as low or near-hyperdiploidy (47-49 chromosomes) and high hyperdiploidy (HeH, more than 50 chromosomes). However, the presence of secondary abnormalities, such as copy number alterations (CNA), associated with a hyperdiploid karyotype may alter the prognosis. **Objective:** : The aim of this study was to analyze the frequency of hyperdiploidy associated with CNA in a cohort of BCP-ALL patients. **Material and methods:** : Forty-nine bone marrow samples of BCP-ALL patients were tested with conventional karyotype, and the DNA undergone to the MLPA SALSA Probemix P036 to detect aneuploidies and to MLPA SALSA Probemix P335 to identify specific gene deletions and amplifications. **Results:** Most patients were aged less than 6 years-old (31/49). The molecular cytogenetic showed that 35/49 (71,4%) cases had low hyperdiploidy and the minority had high hyperdiploidy (28,6%). The presence of somatic trissomies of chromosome 21 was frequent. However, amplifications were observed in *CDKN2A/B* (5/49), *EBF1* (11/49), *IKZF1* (3/49), *PAX5* (11/49), *JAK2* (1/49), *ETV6* (7/49), *BTG1* (4/49), *RB1* (1/49), *CRLF2* (10/49) and PAR1 region (31/49). Deletions were observed in *CDKN2A/B* (7/49), *EBF1* (1/49), *IKZF1* (4/49), *PAX5* (7/49), *JAK2* (1/49), *ETV6* (2/49) and *RB1* (2/49). **Discussion:** : According to the

literature, the most frequent deletions present in hyperdiploidy occurs in *IKZF1* , *CDKN2A/B* , *PAX5* and *ETV6* genes. Besides, these genes play a key role in B-cell development or cell cycle regulation. However, amplifications are not frequently reported. Deletions in genes present in the PAR1 region (*CRLF2* , *IL3RA* , *CSF2RA* and *P2RY8*) are often associated with the genetic fusion *P2RY8::CRLF2* . Although, the most frequent amplifications observed in our study were present in PAR1 region, *PAX5* and *EBF1* . Also, the investigation of CNAs is relevant since further studies revealed that patients with hyperdiploid karyotypes and deletions in *IKZF1* gene were associated with a higher MRD and resistance to prednisone and L-asparaginase. **Conclusion:** : In this study, we showed the importance of molecular methods to characterize the genetic alterations in pediatric patients with BCP-ALL to aid in prognosis and in treatment choice. **Supported by:** FAPERJ, Ministério da Saúde - INCA.

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TROMBOEMBOLISMO VENOSO NA INFÂNCIA: ANÁLISE E REGISTRO DE CASOS EM UM HOSPITAL PEDIÁTRICO TERCIÁRIO

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Objetivo: O diagnóstico de tromboembolismo venoso (TEV) na infância aumentou nas últimas décadas. O avanço no reconhecimento dos eventos tromboembólicos, bem como as particularidades na população pediátrica, baseia-se principalmente na elaboração de registros, bancos de dados e estudos de coorte retrospectivos. Desde 2016, um registro prospectivo de tromboembolismo em crianças foi iniciado no Instituto da Criança/Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo (ICR-HCFMUSP). O objetivo foi descrever o perfil epidemiológico da trombose venosa pediátrica em um serviço de saúde terciário em São Paulo. **Métodos:** Análise retrospectiva do registro institucional de doença tromboembólica pediátrica (idade < 18 anos) de janeiro de 2016 a novembro de 2022. Foram avaliados dados demográficos, fatores predisponentes, local da trombose, diagnóstico radiológico, trombofilias, intervenções terapêuticas e desfechos. **Resultados:** No período do estudo, foram registrados 256 eventos, 142 (55,5%) afetando o sexo masculino. Foram relatadas 164 tromboses venosas, além de 66 tromboses intracardíacas e 26 embolias pulmonares. O sistema venoso superior foi sítio de TEV em 74 casos (28,9%) e o sistema venoso profundo inferior em 44 (17,2%). A trombose venosa cerebral foi diagnosticada em dezenove pacientes (7,4%) e em 27 crianças (10,5%) os eventos tromboembólicos foram em sítios incomuns. Um pico de prevalência foi observado em crianças de 1 a 5 anos, inesperadamente. Dos 256 eventos reportados, 198 (77,3%) estavam relacionados a cateter venoso central e 153 (59,7%) pacientes apresentavam infecção aguda. História prévia de doença crônica foi relatada em 69,9% dos pacientes registrados. As doenças mais prevalentes foram câncer (28,1%), doença renal (12,5%) e doença cardíaca (10,9%). A ultrassonografia doppler foi o exame mais utilizado para o diagnóstico de trombose venosa profunda (46,0%), seguida pela tomografia computadorizada em 46 pacientes (18,0%). O perfil de trombofilia foi investigado em 26 casos (10,2%); destes, seis (2,3%) apresentavam alterações. Terapia antitrombótica foi administrada em 86,0% dos eventos referidos, sendo heparina de baixo peso molecular utilizada isoladamente em 89,8% dos pacientes, seguida de heparina não fracionada. Trombólise após tratamento conservador foi realizada em oito pacientes (3,6%). O acompanhamento por imagem demonstrou recanalização em 71,0% dos eventos (total em 47,6% e parcial em 23,4%). **Discussão:** O presente estudo demonstrou concordância com a análise dos principais Bancos de dados e registros publicados na literatura. Os casos de TEV foram mais prevalentes no sexo masculino, associaram-se a dois ou mais fatores de risco em sua maioria, sendo o cateter venoso central o mais descrito, seguido de infeção e doença crônica. Ultrassonografia com estudo doppler foi o método diagnóstico por imagem mais utilizado, seguido de tomografia computadorizada. A pesquisa de trombofilia não foi realizada de rotina. A modalidade terapêutica mais utilizada, heparina de baixo peso molecular, também foi a mesma descrita na literatura. **Conclusão:** Apesar das limitações, este estudo oferece dados relevantes sobre TEV pediátrico em um serviço de saúde terciário no Brasil. Os resultados são semelhantes aos descritos em outros registros pediátricos e contribuem para o estabelecimento de uma rede para estudos futuros.

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ESTUDO FAMILIAL REVELA O PRIMEIRO CASO DE HEMOGLOBINA OTTAWA EM COERANÇA DE ALFA TALASSEMIA NO BRASIL

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Objetivo: Relatar o primeiro caso no Brasil de interação da hemoglobina (Hb) variante Ottawa com alfa talassemia em um estudo familial. **Materiais e métodos:** Amostra de sangue total de uma criança do sexo feminino de dez meses, proveniente da APAE – Salvador/BA, foi recebida pelo Laboratório