principais alterações: -7/del7q ($p < 0,003$). **Discussão:** Nossos resultados mostraram que a CRI foi o subtipo mais comum, sendo as alterações associadas à evolução: -7, cariótipos complexos corroborando com a literatura. No entanto, diferente do esperado pela classificação do IPSS-R pacientes com del (11)(q23) e +8, também estiveram associados com a evolução da doença. Alterações epigenéticas na SMD-p são sutis sendo raras mutações *DNMT3A* e *TET2*. No entanto, observamos um aumento de expressão das *DNMTs* associada com a evolução da doença e a hipermetilação em $p15^{INK4B}$. Enquanto a expressão dos genes da maquinaria de desmetilação foi semelhante aos controles. Estudos mostram que o envelhecimento está associação há uma maior desregulação na maquinaria de metitilação e desmetilação do DNA dos pacientes com SMD, contudo podemos observar que essas alterações tentem a acentuar com a evolução da doença para alguns genes importantes como o $p15^{INK4B}$. **Conclusão:** Nossos resultados mostraram que durante o desenvolvimento e evolução de SMD-p para LMA há um desbalanço progressivo na maquinaria de metilação com aumento de expressão das *DNMTs*. A hipermetilação em $p15^{INK4B}$ foi um evento mais tardio associado à progressão da SMD, principalmente nos pacientes que apresentaram -7/del(7q). Em conjunto nossos resultados sugerem um modelo de evolução de SMD para LMA. **Auxílio financeiro:** INCA – Ministério da Saúde, FAPERJ.

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UNEXPECTED CONCOMITANCE OF ETV6-RUNX1 AND BCR-ABL1P210 IN A CHILD WITH B-CELL PRECURSOR ACUTE LYMPHOBLASTIC LEUKEMIA

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ETV6-RUNX1 and *BCR-ABL1* are recurrent primary events in pediatric B-cell precursor acute lymphoblastic leukemia (B-ALL), being the former the most common one. Elegant studies have shown that these are founder events, but additional genetic events are needed for leukemia development. Nonetheless, these common primary alterations are typically mutually exclusive. The presence of both *ETV6-RUNX1* and *BCR-ABL1* is a very rare event which has been previously reported in only two B-ALL cases, being one pediatric and one adult patient. Herein, we report a rare pediatric B-ALL case with simultaneous occurrence of *ETV6-RUNX1* and *BCR-ABL1* . A 5-year-old boy was admitted to Hospital Federal da Lagoa, Rio de Janeiro, Brazil. At clinical investigation, he presented hepatosplenomegaly, a white blood cell count of 0.7×10^9/L, 94% of blasts, Hb of 6.5 g/L, and platelet count of 16×10^9/L. The patient was treated according to the AIEOP-BFM protocol. He presented a poor prednisone response (at day 8; >1000 circulating lymphoblasts), the minimal residual disease at day 33 was negative (0.08%), and he was considered in complete remission. Four months after the diagnosis, the child is alive with negative levels of residual disease. The immunophenotyping was characterized by nTdT (low), CD10, CD13, CD19, CD21, CD22, CD24, CD33, CD34, CD38 and CD45, cCD66c, cCD79, CD81, CD123, and CD58 positive cells in 94.1% of lymphoblast cells and a diploid profile (DNA index of 1.09). *ETV6-RUNX1* and *BCR-ABL1* p210 were identified by reverse transcriptase PCR (RT-PCR). The presence of both rearrangements was confirmed by sanger sequencing. Moreover, additional copy number alterations (CNA) were evaluated by multiplex ligation-dependent probe amplification (MLPA) using SALSA MLPA P335-C1. Deletions affecting *IKZF1* (exons 2 and 3), *CDKN2A* (exons 2 and 5), *CDKN2B* (exon 2), *PAX5* (exon 1), and *ETV6* (exons 2, 3, 5 and 8) were detected. In consonance with previous studies, these CNAs are the most frequently secondary events found in patients with *ETV6-RUNX1* . Of note, *IKZF1* deletions are enriched in patients with *BCR-ABL1* . Due to the lack of material for FISH analysis, the clonality of both biomarkers could not be verified. However, we observed a low load of *BCR-ABL1* p210 transcript in comparison with the endogenous gene, suggesting that this gene fusion could be present in subclones. In conclusion, to the best of our knowledge, this is the second pediatric case identified with *ETV6-RUNX1* and *BCR-ABL1* gene rearrangement in concomitance, but is the first case reported with the p210 transcript, which is extremely rare in B-ALL. Previous studies have shown that the additional genetic events found can impact the usually favorable prognosis of *ETV6-RUNX1* – positive B-ALL. The clinical significance for these rare patients with both rearrangements remains unclear due to the small number of reported cases to date. Therefore, it is very important to describe more cases which will ultimately enable future studies with larger series and clarify this enigmatic subgroup.

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XANTOGRANULOMA SISTÊMICO INFANTO-JUVENIL – UM RELATO DE SUCESSO

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Paciente, sexo feminino, nascida à termo, APGAR 8/9, parto normal, sem intercorrências no período gestacional. Desde o nascimento com lesões nodulares palpáveis, azuladas e palpáveis, de 0,5 a 1 cm, inicialmente localizadas na face. Aos 30 dias foi internada em hospital pediátrico com irritabilidade e distensão abdominal progressiva. À admissão apresentava-se pálida e apresentava nódulos cutâneos visíveis e palpáveis na face e extremidades, além de hepatomegalia e esplenomegalia maciça. Os exames de sangue mostraram anemia leve e