=0.007), poor-risk karyotype (HR 4.8 95% CI 2.29 – 10, p <0.001), hemoglobin < 8.5 (HR 3.44 95% CI 1.56 – 7.88, p =0.002) and platelets < 80.000 (HR 3.5 95% CI 1.49 – 8.23, p =0.002). In multivariate analysis, parameters associated with worse OS were transfusion dependency (HR 2.26 95% CI 1.27 – 4.04, p = 0.006) and poor karyotype (HR 2.53 95% CI 1.41 – 4.57, p =0.002). Thirty-five patients (50%) had febrile neutropenia, and 30% delayed one or more cycles. **Discussion:** The results reinforce the poor outcomes of higher-risk MDS despite the use of HMA. Some real-world studies previously published had already showed a suboptimal drug benefit, probably by the underutilization and low persistence of the treatment, in a subgroup of critically ill patients. **Conclusion:** This retrospective analysis confirms the poor prognosis of higher-risk MDS treated with HMA. Poor-risk karyotype and transfusion dependency are the main factors that impact the outcomes.

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CYTOGENETIC ANALYSIS OF 2,205 CASES OF HEMATOLOGICAL NEOPLASIA AND GENETIC DISEASES: A SINGLE CENTER EXPERIENCE

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Cytogenetics is defined as the study of chromosome morphology, pathology function, and behavior. Cytogenetics is important to define clonality for bone marrow disorders and for diagnosing a lot of constitutional disorders. We analyzed a ten-year experience of bone marrow and peripheral blood cytogenetics for defining clonal and not clonal disorders at the Cancer Cytogenomics Laboratory of the Federal University of Ceará. All bone marrow and peripheral blood samples were submitted to cell culture (for 24 hours and 72 hours, respectively), after which all were processed, and G-banding performed using trypsin and Wright-Giemsa staining. The karyotype results of 2,205 patients were analyzed including additional information such as age, sex, requesting unit and suspected diagnosis. Among these, 1744 (79.1%) were bone marrow samples and 461 (20.9%) were peripheral blood samples used to investigate constitutional changes. The female gender (54.4%) stood out in the Bone Marrow samples, aged between 71 and 80 years (20.2%). Among the bone marrow results, 726 of 1744 (42%) were normal and 431 of them (24.7%) were abnormal karyotypes. The main etiological suspicion was Myelodysplastic Syndrome (54.2% of cases), and 42.1% were normal. Among the chromosomal abnormalities, the main alterations found were: 26% cases with isolated del (5q) and/or other alterations, 10% of cases were complex, 10% were isolated del(11q) and/or with other alterations, 9% of cases with del (7q) isolated and/or with other alterations and 7% cases with +8 isolated and/or with other alterations, 2% cases had del(17q) isolated and/or with other alterations, 1% of cases had del(17p) and one case with i(17q). A total of 127 cases were suspected as AML, with 34.6% altered karyotypes, 34.4% normal karyotypes. Among the abnormal karyotypes: 20,5% cases with t(15;17) and 13,6% were complex karyotypes, 6,8% were del(11q23), 4,5% cases with t(8;21), 1 case with del (17p) and another with i(17q). A total of 161 cases were suspected as CML, among which: 49,7% abnormal karyotype, 24.2% were normal. Among the chromosomal abnormalities, 65% were t(9;22) and 2,2% were t(9;22) and other alterations and/or variants of t(9;22). In this way, this study shows that cytogenetic was important for diagnostic purposes of patients principally for diseases such as Myelodysplastic Syndrome, Acute Myeloid Leukemia and Chronic Myeloid Leukemia. Cytogenetic analysis is also fundamental for analysis of constitutional and somatic genetic disorders. Regarding the constitutional results, 176/461 (38%) showed chromosomal abnormalities with 66 (37.5%) showing trisomy 21 followed by trisomy 18 (14.7%) and androgen insensitivity syndrome (11.3%). Identification of constitutional or malignance chromosomal anomalies can provide important information for genetic medical counseling. Cytogenetic analysis of hematological neoplasms and for constitutional disorders is helpful for diagnosis and monitoring the effects of treatment, being still very important in the new molecular era.

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TROMBOCITOSE ESSENCIAL COM PROGRESSÃO PARA LEUCEMIA AGUDA: RELATO DE CASO

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Introdução: A trombocitose essencial (TE) é uma síndrome mieloproliferativa caracterizada pela hiperproliferação dos megacariócitos na medula óssea e trombocitose no sangue periférico. Podem ocorrer eventos trombóticos, hemorrágicos ou sintomas vasomotores. A evolução para leucemia aguda é rara. **Relato:** Paciente feminina, 68 anos, hipertensa, tabagista (53 maços-ano) e com neuralgia do trigêmeo. Uso de antihipertensivos, ácido fólico, ácido acetilsalicílico 100mg/dia e

hidroxiureia (HU) 1000 mg 2x/semana e 500 mg 5x/semana. Realizava acompanhamento ambulatorial externo devido TE desde 2010 com escore R-IPSET de alto risco, sem histórico de trombose, sem alterações de cariótipo e com uso de HU desde 2012. Transferida ao SUS em 2014 com exames locais: biópsia de medula com mieloproliferação e aumento de megacariócitos, mutação JAK V617F positiva, medulograma com vários grupamentos de plaquetas, sem blastos. Solicitou consulta ambulatorial em julho/22 por sintomas constitucionais (astenia, perda ponderal de 2kg) e hematomas nas extremidades. Exames demonstravam Hb 7,3 g/dL, leucócitos totais 4480/uL, plaquetas 38000/uL, creatinina 1,49, sem alterações ao exame físico, ECOG 0. Pela possibilidade de bicitopenia de causa medicamentosa foram suspensos AAS e HU e solicitado exames. Retorna após 7 dias com piora da astenia e com laboratoriais: Hb 7,9 g/dL, leucócitos totais 4760/uL (diferencial: 4% bastões - 190/uL, 20% blastos - 950/uL), plaquetas 38000/uL, LDH 558U/L, anisocitose, policromatofilia e eritrócitos em gota. Demais exames sem alterações. Internou para investigação e realizou medulograma com raros representantes do setor eritroide e megacariocítico e vários representantes do setor granulocítico sendo 22% de blastos. A imunofenotipagem demonstrou 18% de células imaturas positivas para: CD34, CD117, HLADR, CD13, CD123, CD33 fraco. Os clones mieloides têm aumento de mastócitos imaturos com expressão aberrante de CD25, expressão anômala de cMPO nos eosinófilos. Há ausência da mutação FLT3. Diagnóstico de leucemia mieloide aguda secundária a TE. Atualmente recebe tratamento com o protocolo 7+3. **Discussão:** Os estudos de follow-up mostram que 1 a 4% dos pacientes progridem em 7-10 anos. Os fatores de risco são: idade >60 anos, anemia, plaquetose > 1 milhão, duas ou mais mutações somáticas e uso de agentes alquilantes. A mutação JAK V617F positiva não interfere na sobrevida ou risco de progressão. Em um pequeno estudo, as alterações genéticas parecem apresentar ganho da mutação 2p e deleção do 5q, assim como mutação TP53 mesmo em pacientes com JAK2 negativos (pior prognóstico). O tratamento para pacientes considerados fit é quimioterápico (7+3). Devido ao CD33+ poderia ser associado à gemtuzumab ozogamicina se disponível no SUS. O prognóstico é desfavorável devido a idade e TE prévia. A sobrevida relativa média em 5 anos é de 30,5%. Pode-se realizar TCTH alogênico sem mieloablação porém com chance de recaída e mortalidade de até 50%.

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RETROSPECTIVE STUDY OF PATIENTS WITH MYELODYSPLASTIC NEOPLASM WITH RING SIDEROBLASTS AT UNIVERSITY CENTERS IN NORTHEAST OF BRAZIL

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The myelodysplastic neoplasms (MDS) consist of hematopoietic neoplasms originated in the bone marrow and characterized by dysfunctional hematopoiesis, cytopenias, and dysplasia, which is currently diagnosed using clinical, morphological and genetic parameters. A particular subtype of this disease, MDS with ring sideroblasts (MDS-RS), presents with an abnormal accumulation of perinuclear mitochondrial iron in erythroid precursors and a low risk of progression to acute myeloid leukemia. Therefore, a retrospective longitudinal observational study was conducted with 31 patients diagnosed with MDS-RS at two tertiary hospitals in Fortaleza, Ceará. The clinical characteristics and the results of karyotype, as well as the transfusion dependence of patients were evaluated. From these 31 patients, 16 (51.6%) are female and 15 (48.4%) are male. 18 had normal karyotype (58.1%), and 13 had altered karyotype (41,9%). As for cytogenetic abnormalities, it was observed that of these 13 patients, 2 patients had Y nullisomy (15.3%), 2 had chromosome 11 deletion (15.3%), 1 manifested chromosome 20 deletion (7.7%), 2 (15.3%) with chromosome 5 deletion, 1 exhibited chromosome 7 deletion (7.7%), 1 presented trisomy of chromosome 8 (7.7%), and 4 had other alterations (31%). In regard to risk stratification of these 30 patients, 29 had very low or low risk (93.5%) and 2 had intermediate risk (6.5%). Considering transfusion dependence, 11 were dependent (35.5%) and 20 were not (64.5%). From these 11 patients who underwent transfusion 2 (18.1%) had this terapy at diagnosis and 9 (81.9%) after first-line treatment. Low-risk MDS is a heterogeneous group of disorders – clinically, pathologically, and even molecularly. Despite being generally considered a low-risk subtype of MDS, with generally good prognosis, MDS-RS is an entity with diverse clinical presentations, characterized by transfusion dependence and a range of cytogenetic alterations as demonstrated in this study. High risk of developing transfusion dependence is frequently observed and refractoriness to erythropoietin is now considered formal indication to the new approved TGF-β inhibitor, luspatercept. Therefore, it is necessary to accurately evaluate patients, including iron staining in diagnosis workup, and to perform risk stratification before decision making. In conclusion, it can be stated that clinical and correct laboratory evaluation of individuals with MDS-RS is essential to ensure a better understanding of the patient's condition and to identify important prediction factor of response to therapy.

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SÍNDROME DE SWEET: RELATO DE CASO

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