

intersticiais e perivasculares com cadeias leves kappa e lambda de imunoglobulina apresentando relação dentro dos limites da normalidade. Afastada a presença de mieloma múltiplo, a paciente iniciou terapia com Dexametasona 4 mg (10 comprimidos ao dia por 4 dias), Melfalana 2 mg por 4 dias e profilaxia de Pneumocistose com tratamento com Sulfametoxazol + trimetoprima 800+160 mg 3x por semana. Após tratamento não houve queda nas cadeias leves livres e nem melhora das funções cardíaca e renal e foi encaminhada para o serviço de transplante de medula óssea. **Discussão:** A ACL é uma doença rara caracterizada pelo depósito de cadeias leves monoclonais em diversos órgãos, mais prevalente em homens e diagnosticada em média aos 58 anos. Plasmócitos clonais produzem excessivamente cadeias kappa ou lambda, com metade dos casos associada ao mieloma múltiplo e 20% à gamopatia monoclonal de significado indeterminado. A disfunção renal e a síndrome nefrótica são comuns, enquanto envolvimento hepático e cardíaco são menos frequentes, incluindo cardiomiopatia e insuficiência cardíaca. O diagnóstico é confirmado por biópsia com coloração pelo vermelho do congo e espectroscopia de massa. O tratamento visa suprimir a produção de cadeias leves, destacando-se o regime com daratumumabe, bortezomibe, ciclofosfamida e dexametasona e o transplante autólogo de células-tronco, que apresenta boa eficácia e taxas baixas de mortalidade. A resposta hematológica é um importante preditor de sobrevida, com taxa de 53% em dez anos para pacientes com resposta completa ao tratamento. **Conclusão:** A ACL apresenta desafios diagnósticos e terapêuticos substanciais. Este caso ilustra a importância de um diagnóstico precoce e de um tratamento intensivo, visando aumentar a sobrevida e o bem-estar do paciente, mesmo diante de um prognóstico reservado.

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MAPPING AMYLOID SUBTYPES IN BRAZIL THROUGH MASS SPECTROMETRY: RESULTS OF A DIAGNOSTIC SUPPORT PROGRAM

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Objectives: Amyloidosis results of tissue deposition of misfolded proteins aggregates, leading to localized or systemic forms. Over 40 proteins are amyloidogenic, the main 2 being immunoglobulin light chain (AL) and transthyretin (ATTR). Mass spectrometry (MS) is the gold-standard for subtyping amyloid. Despite MS commercial availability in Brazil since 2019, amyloidosis remains underdiagnosed with nearly a years delay, underscoring the need for ongoing medical education and specialized referral centers. Limited access to MS due to regional disparities and health system heterogeneity led to the implementation of a diagnostic support program (DSP)

operated by a pharmaceutical company. This study aims to describe the occurrence of amyloid subtypes in Brazil using DSP data. **Materials and methods:** This observational, cross-sectional study analyzes data from DSP database between January 2022 and May 2024. Eligible patients were ≥18 years, with biopsy-proven amyloidosis confirmed by Congo red staining and consented to data collection/publication. Amyloid was extracted by laser microdissection, processed by protein denaturation, trypsin digestion and resulting peptides analyzed by MS (QExactiveHF-X mass spectrometer coupled with NanoUltimate liquid chromatography). Protein identification used bioinformatics (MaxQuant v.4.2.1, FragPipe v.13 platforms against UniProt/SwissProt database). The techniques were conducted at Grupo Fleury Laboratory. Sociodemographic characteristics, biopsy sites and MS results were analyzed using descriptive statistics. **Results:** 403 patients were included. Median age was 65 years (56-74), 52% were male, and 65% from the southeast region. AL was the predominant subtype in Brazil (76%), followed by ATTR (7%), both present in all regions of the country. AA and AHL types were reported only in the southeast region. Only one case of AA amyloidosis was observed, in the gastrointestinal tract. Abdominal fat and kidney were the most common sites of biopsy (23% and 22%), followed by bone marrow (15%). This pattern was consistent across most regions, except in the south, where abdominal fat and bone marrow were equally used (24.1% each), and heart biopsies were the third most common (17.2%). AL amyloidosis was mainly identified in the kidney and bone marrow (29%; 14%), and ATTR in abdominal fat, and heart (35%; 30%). **Discussion:** This study is the first to analyze a Brazilian national cohort with amyloidosis, providing a comprehensive overview of amyloidosis subtypes and their geographical distribution across different country regions. AL is the most common subtype in Brazil, followed by ATTR, consistent with findings in Europe and the United States. Abdominal fat is the main biopsy site nationwide, following international recommendations for diagnosing amyloidosis and avoiding invasive organ biopsies. **Conclusion:** The widespread use of MS on a national scale, facilitated by the creation of a DSP, has proven to be a relevant tool in the diagnosis of amyloidosis in Brazil. It allows for accurate amyloid subtyping, contributes to epidemiological information, improves the diagnostic process and enables the correct treatment of patients. Additionally, it strengthens the establishment of a reference laboratory with expertise in the field in Brazil, with the potential to become a reference center in Latin America. **Financial disclosure:** The development of this study was funded by Janssen-Cilag Ltda

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PRACTICAL CHALLENGES IN AMYLOID SUBTYPING IN BRAZIL: A COMPARATIVE STUDY BETWEEN MASS SPECTROMETRY AND A CLINICAL-LABORATORY MODEL

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