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ORIGINAL ARTIÇLE

Clinical characterization of patients with renal amyloidosis

Caracterización clínica de pacientes con amiloidosis renal

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ABSTRACT

Introduction: renal amyloidosis is a disease of great clinical importance due to the poor prognosis and short life expectancy of patients who present it.
Objectives: to identify the clinical characteristics of patients with renal amyloidosis seen at the "Dr. Abelardo Buch" Nephrology Institute, between 2010 and 2018.
Methods: observational, descriptive, retrospective cohort study, developed in a population of 26 patients with a diagnosis of renal amyloidosis, attended at the indicated health institution. The documentary review allowed the collection of information that led to the variables analyzed. Descriptive statistical methods were used for the analysis of the information obtained, and medical ethics were respected.
Results: patients in the age group 50-64 years old predominated (57,7 %), being the female sex the most representative (65,4 %). Edema and foamy urine were the main clinical manifestations, being nephrotic syndrome (92,3 %), the main reason for indication of renal biopsy. Renal function replacement therapy was not required in 88,5 % of patients, and 76,9 % of patients survived to six months. Survival was higher for the group of patients who did not require dialytic therapy compared to those who did, although no statistically significant differences were found (p=0,13).
Conclusions: the sample of patients with renal amyloidosis was characterized, evidencing the main clinical characteristics shown by them. A reduction in the survival rate was observed as the disease evolved over time.

Keywords: Amyloidosis; Amyloidogenic Proteins; Signs And Symptoms; Diagnosis; Renal Dialysis.

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RESUMEN

Introducción: la amiloidosis renal es una enfermedad que reviste gran importancia clínica debido al mal pronóstico y una corta esperanza de vida de los pacientes que la presentan.
Objetivos: identificar las características clínicas de los pacientes con amiloidosis renal atendidos en el Instituto de Nefrología "Dr. Abelardo Buch", entre 2010 y 2018.
Métodos: estudio observacional, descriptivo, de cohorte retrospectivo, desarrollado en población de 26 pacientes con diagnóstico de amiloidosis renal, atendidos en la institución sanitaria indicada. La revisión documental permitió la obtención de información que dio salida a las variables analizadas. Se emplearon para el análisis de la información obtenida fueron empleados métodos de estadística descriptiva, se respetó la ética médica.
Resultados: predominaron los pacientes del grupo etáreo 50-64 años (57,7 %), siendo el sexo femenino el más representativo (65,4 %). Los edemas y orinas espumosas fueron las principales manifestaciones clínicas, siendo el síndrome nefrótico (92,3 %), el principal motivo de indicación de biopsia renal. El 88,5 % de los pacientes no requería de tratamiento sustitutivo de la función renal, sobreviviendo a los seis meses el 76,9 % de los pacientes. Se apreció una supervivencia mayor para el grupo de pacientes que no requirieron de terapia dialítica en comparación a los que si lo requerían, aunque no se precisaron diferencias estadísticamente significativas (p=0,13).
Conclusiones: fue caracterizada la muestra de pacientes con amiloidosis renal, evidenciándose las principales características clínicas mostradas por ellos. Se constató una reducción en l tasa de supervivencia al evolucionar en el tiempo la enfermedad.

Palabras clave: Amiloidosis; Proteínas Amiloidogénicas; Signos y Síntomas; Diagnóstico; Hemodiálisis.

INTRODUCTION

Renal amyloidosis, a disease characterized by the accumulation of amyloid fibrils in renal tissues, has emerged in recent decades as a topic of growing clinical and scientific interest. The relevance of this disease lies in its ability to trigger progressive dysfunctions in the kidneys, affecting the quality of life and renal health of affected individuals. The main forms of systemic amyloidosis are primary (AL) and secondary (AA) amyloidosis.[1] In these systemic forms, the organs mainly involved are the kidneys, heart and gastrointestinal tract in the case of AL; and autonomic neuropathy, and osteoarticular involvement in AA.[2]

Amyloidosis has an estimated prevalence of 1 in 60,000 people and 0.8% at autopsy. It causes nephrotic syndrome in adults in approximately 5%, and is rarer in children. There is a slight male predominance for primary amyloidosis. In large series of native kidney biopsies from adult patients in the USA, amyloidosis was the histological diagnosis in 2% of cases, most of which were of the AL type. In contrast, in developing countries and in the Mediterranean, renal amyloidosis is more commonly of the AA type.[3,4,5]

Systemic amyloidosis presents with a wide variety of signs and symptoms, with the kidney being an organ often affected, giving it a poor prognosis. Although it is a rare disease, it should always be considered in the differential diagnosis of the causes of progressive renal failure with or without proteinuria.[6,7,8,9]

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Renal involvement, in the context of systemic AA and AL amyloidosis, is practically constant (90%), with glomerular involvement being the most frequent. The main forms of renal presentation are: nephrotic syndrome with loss of albumin and edema that are difficult to manage, acute renal failure mainly due to tubulointerstitial involvement, when it affects vessels exclusively it presents with arterial hypertension, nephrogenic diabetes insipidus, infiltration of the collecting duct, Fanconi syndrome, light chain toxicity in the proximal convoluted tubule, type I tubular acidosis and rapidly progressive glomerulonephritis.[10,11,12]

For the diagnosis of amyloidosis, a high index of suspicion is required, with electron microscopy and protein electrophoresis being some of the methods used for this purpose.[13,14] Due to the high mortality rate leading to kidney failure in addition to severely compromising the function of other organs, the need to study this disease is understood. Taking into account the above, the present research was carried out, which aimed to identify the clinical characteristics of patients with renal amyloidosis treated at the "Dr. Abelardo Buch" Nephrology Institute, between 2010 and 2018.

METHODS

An observational, descriptive, retrospective cohort study was conducted. The study population consisted of 26 patients with a histological diagnosis of renal amyloidosis, treated at the Dr. Abelardo Buch Nephrology Institute, in the period from 2008 to 2018.

The information analyzed was collected from the individual patient's medical history, registered in the Institute's Archives department, and the renal biopsy reports from the Institute's Pathology department. For these purposes, all biopsy reports of the patients during the study period were reviewed, so that all patients with amyloidosis could be identified. All of this allowed obtaining information that led to the variables analyzed (age, sex, skin color, clinical manifestations, reason for renal biopsy, 24-hour proteinuria, total plasma proteins, albuminemia, glomerular filtration rate, need for renal function replacement therapy, survival at six and 12 months).

Statistical processing

Data processing was carried out using the statistical package SPSS, version 26 for Windows, using descriptive statistical methods to process the information (absolute frequencies and percentages were analyzed, as well as measures of central tendency and dispersion). The results were presented in tables.

Ethical aspects

The Scientific Council and the Ethics Committee of the institution were consulted for the execution of the study, and approval was received from both. Each participant was informed of the confidentiality to which the data and results obtained during the stages in which the study was developed would be subject. Each patient was also informed of the objectives and characteristics of the study, and their written informed consent was requested to participate in it.

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RESULTS

The study found a predominance of patients in the age group 50-64 years (57,7 %), with the female sex being the most representative (65,4 %), as well as white skin color (61,5 %).

Table 1. Sample distribution according to presence of clinical manifestations.

Clinical manifestations	No.	%
Edema	25	96,2
Foamy urine	25	96,2
Heart failure	9	34,6
Syncope	7	26,9
Hepatomegaly	6	23,1
Anemia	6	23,1
Arrhythmias	4	15,4
Tumor	3	11,5
Macroglossia	1	3,8
Orthostatic hypotension	1	3,8
Digestive bleeding	1	3,8
Petechiae	1	3,8
Pathological fractures	1	3,8

Table 1 shows the frequency of clinical manifestations presented by the patients studied, with the majority showing edema and foamy urine with 96,2 % in both cases. Other symptoms such as heart failure (34,6 %), syncope (26,9 %), hepatomegaly (23,1 %), and anemia (23,1 %) followed in order of frequency.

The presence of nephrotic syndrome (92,3 %) was the main reason for indicating a renal biopsy. On the other hand, 65,4 % of patients on admission showed a 24-hour proteinuria in a range of 3,5-6,9 grams. Regarding total proteins, 73,1 % showed a value less than 60 g/l; and an albuminemia less than 30 g/l.

Table 2. Sample distribution according to glomerular filtration rate.

Glomerular filtration rate (ml/min/1.73m2 SC)	**No.**	**%**
≥ 90	7	26,9
60-89	9	34,6
30-59	7	26,9
15-29	3	11,5

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Table 2 shows the value of the glomerular filtration rate, with patients with a filtration rate ranging between 60-89 ml/min/1,73m2 SC predominating (34,6 %), while 11,5 % had values between 15-29 ml/min/1,73m2 SC.

Table 3. Survival of patients with amyloidosis according to the need for renal function replacement therapy.

Need for renal function replacement therapy	No.	%	6 months (%)		12 months (%)	
			S(t)	OF*	S(t)	OF*
Yeah	3	11,5	66,7	27,2	0,0	0,0
No	23	88,5	78,0	8,6	47,8	10,4
Total	26	100	76,9	8,3	42,3	9,7

*Standard deviation

Table 3 shows that 88,5 % of patients did not require renal function replacement therapy. When evaluating patient survival, overall, it can be seen that 76,9 % of patients survived six months after diagnosis, while only 42,3 % remained alive after one year. Regarding the need for treatment, survival was higher for the group of patients who did not require dialysis therapy compared to those who did [78,0 % versus 66,7 % in the first six months; while after 12 months, 47,8 % of those who did not require replacement therapy remained alive, with all those who required such replacement therapy dying. However, these differences were not statistically significant (p=0.13).

DISCUSSION

The high frequency of cases found in the study was in the age group between 50 and 64 years. According to the consulted bibliography, the age of the patients reflects the underlying clinical condition; patients with AL amyloidosis are typically adults, it is estimated that it affects five to 12 people per million per year, although autopsy studies suggest that the incidence could be higher and those with amyloidosis associated with familial Mediterranean fever are children. The clear predominance found in the female sex does not coincide with the consulted bibliography where we found that there is a slight male predominance for primary amyloidosis.[5]

The high presence of edema and foamy urine in almost all patients and symptoms such as heart failure, syncope, hepatomegaly, anemia, arrhythmias and association with tumors in less frequency coincide with the consulted bibliography. It is indicated that the infiltration of the myocardium by amyloid substance causes a functional alteration in the form of congestive heart failure, cardiomegaly and arrhythmias, with intense depression of the systolic and diastolic function, defining a typical pattern of restrictive cardiomyopathy. This infiltration represents the most important prognostic factor in systemic amyloidosis.[12]

International data recognize that gastrointestinal involvement is frequent, with macroglossia being one of the most characteristic alterations, which makes chewing and swallowing difficult. Malabsorption, diarrhea and digestive hemorrhage are frequently observed. Hepatic infiltration by amyloid substance is frequent in systemic forms and results in anicteric cholestatic syndrome and homogeneous hepatomegaly.[13]

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Nephrotic syndrome was the main indication for renal biopsy. This coincides with what has been reported in the literature, which also describes how nephrotic syndrome is one of the consequences of this disease, being present in approximately 5% of adults, being rarer in children.[7] In this regard, the values of nephrotic range proteinuria, hypoproteinemia, and hypoalbuminemia found in the study coincide with the bibliography consulted.(14)

In a study conducted by Esteve et al.,[6] of 76 cases, in 33 %, dialysis was not indicated due to poor clinical condition, short life expectancy and poor quality of life. The mean overall survival at diagnosis was 55 % and 40 % at 12 and 24 months (AL 58 % and 19 %; AA 55 % and 44 %). The mean survival from the start of dialysis was 30 % and 5 % at 12 and 24 months.

According to the literature consulted, the median survival of patients with AL is less than two years, and depends mainly on the associated syndrome. Thus, in patients with heart failure, the median survival is less than six months; while, when the associated syndrome is peripheral polyneuropathy, it is more than five years. Cardiac involvement is the cause of death in at least 50% of cases, while current survival at ten years from diagnosis is slightly less than 5 %. Other factors associated with poor prognosis in these patients include high doses of chemotherapy, the presence of symptomatic heart failure, the presence of failure in more than two organs, advanced age (>65 years) and those who are not candidates for transplant.[15]

BIBLIOGRAPHIC REFERENCES

1. Lachmann HJ, Hawkins PN. Systemic amyloidosis. Current opinión in pharmacology. [Internet]. 2006[citado 06/04/2024]; 6(2): 214-220. Disponible en: https://doi.org/10.1016/j.coph.2005.10.005

2. Browning M, Banks R, Tribe C, et al. Renal involvement in systemic amyloidosis. Proc Eur Dial Transplant Assoc[Internet]. 1983[citado 06/04/2024]; 20: 595-602. Disponible en: https://europepmc.org/article/med/6657678

3. Aroca Martínez G, García Tolosa R, Cadena A, Anaya Taboada M, Niño LM. Amiloidosis Renal: Reporte de un Caso y Revisión de Tema. Rev colomb. Nefrol[Internet]. 2014[citado 06/04/2024]; 1(supl 1): 134–142. Disponible en: https://revistanefrologia.org/index.php/rcn/article/view/139

4. Bohle A, et al. The long term prognosis of AA and AL renal amyloidosis and the Pathogenesis of Chronic Renal Failure in Renal Amyloidosis. Pathol Res Pract[Internet]. 1993[citado 06/04/2024]; 189(3): 316-31. Disponible en: https://doi.org/10.1016/S0344-0338(11)80516-9

5. Cazalets C, et al. Epidemiological description of amyloidosis diagnosed at University Hospital of Rennes from 1995 to 1999. Rev Med Interne[Internet]. 2003[citado 06/04/2024]; 24(7): 424-30. Disponible en: https://doi.org/10.1016/s0248-8663(03)00136-x

6. Esteve J, Ponz N, García l, et al. Afectación renal en la Amiloidosis. Características clínicas, evolución y supervivencia. nefrología[Internet]. 2006[citado 06/04/2024]; 26(2): 157-289. Disponible en: https://revistanefrologia.com/es-afectacin-renal-en-la-amiloidosis-caractersticas-clnicas-evolucin-y-supervivencia-articulo-X0211699506019651

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Rev Ciencias Médicas. 2025; 28(2025): e6627

7. alcalá Salgado MA, Hernández-Estrada S, Abraham MV, Torres-Pastrana J. Diagnóstico diferencial de amiloidosis primaria y familiar en un paciente con psoriasis. Reporte de caso. Nefrol Latinoam*[Internet].* 2017*[citado 06/04/2024];* 14(2): 79-82. *Disponible en:* https://www.elsevier.es/es-revista-nefrologia-latinoamericana-265-articulo-diagnostico-diferencial-amiloidosis-primaria-familiar-S2444903217300227

8. Hamburger J, Crosnier J, Grunfeld JP. Nefrologia. Ciudad de la Habana: Editorial Científico Técnica; 1987.

9. Karam S, Haidous M, Royal V, Leung N. Renal AA amyloidosis: presentation, diagnosis, and current therapeutic options: a review. kidney International[Internet]. 2022[citado 06/04/2024]; 103(3): 473-484 Disponible en: https://www.sciencedirect.com/science/article/abs/pii/S0085253822010122

10. Hachulla E, Grateau G. Diagnostic tools for amyloidosis. Joint Bone Spine[Internet]. 2002[citado 06/04/2024]; 69(6): 538-45. Disponible en: https://doi.org/10.1016/S1297-319X(02)00449-9

11. Grogan M, Dspenzieri A, Gertz MA. Light-chain cardiac amyloidosis: strategies to promote early diagnosis and cardiac response. Hear[Internet]. 2017[citado 06/04/2024]; 103(14). Disponible en: https://doi.org/10.1136/heartjnl-2016-310704

12. Kyle RA, Greipp PR. Amyloidosis (AL). Clinical and laboratory features in 229 cases. Mayo Clin Proc[Internet]. 1983[citado 06/04/2024]; 58(10): 665-83. Disponible en: https://pubmed.ncbi.nlm.nih.gov/6353084/

13. Cowan AJ, et al. Amyloidosis of the gastrointestinal tract: a 13-year, single-center, referral experience. Hematologica[Internet]. 2013[citado 06/04/2024]; 98(1): 141-146. Disponible en: https://doi.org/10.3324/haematol.2012.068155

14. Sanchorawala V. Light-Chain (AL) Amyloidosis: Diagnosis and Treatment. Clin J Am Soc Nephrol[Internet]; 2016[citado 06/04/2024]. Disponible en: https://doi.org/10.2215/CJN.02740806

15. Amiloidosis de cadenas ligeras. Amyloidosis Research Consortium[Internet]; 2024[citado 06/04/2024]. Disponible en: https://arci.org/wp-content/uploads/2024/07/Spanish-AL-INTRODUCCION-A-LA-ENFFERMEDAD-WEB.pdf