Turkish Neurosurgery 2009, Vol: 19, No: 2, 159-162

Amyloidoma of the Temporal Bone and Upper Cervical Spine; Presentation of a Rare Clinical Entity with a Brief Literature Review

Temporal Kemik ve Üst Servikal Vertebral Yerleşimli Amiloidoma: Nadir Bir Olgu Sunumu

Hakan ORUCKAPTAN[1]
Kader KARLI OĞUZ[2]
İlkay IŞIKAY[3]
Şevket RUACAN[4]

[1,3] Hacettepe University School of Medicine, Neurosurgery Department, Ankara, Turkey
[2] Hacettepe University School of Medicine, Radiology Department, Ankara, Turkey
[4] Hacettepe University School of Medicine, Pathology Department, Ankara, Turkey

Received : 06.02.2009
Accepted : 26.02.2009

Correspondence address:
Hakan ORUCKAPTAN
E-mail: oruckaptan@hacettepe.edu.tr

ABSTRACT

The amyloidoses comprise a heterogeneous group of diseases characterized by the extracellular deposition of an insoluble protein complex in various tissues. Amyloidomas in bone are most common in patients with systemic amyloidosis and plasma cell dyscrasias. Decreased clearance of ß2 microglobulin frequently causes excessive amyloid deposition in the musculoskeletal system in patients with a history of chronic renal failure and long-term dialysis treatment. Calvarial and/or upper cervical amyloid depositions are rarely seen in clinical practice; therefore the diagnosis requires high index of suspicion and special staining of the tissue. In this article, we present a patient with amyloidoma at the right temporal bone and upper cervical spine. The etiology, radiological findings and differential diagnosis were briefly discussed in the highlights of relevant literature. Amyloidomas should be particularly kept in mind in patients with a history of long-term dialysis therapy, plasma cell dyscrasias or long-standing inflammatory diseases. Differential diagnosis mostly encounters benign or malign mesenchymal neoplasms of the dura and skull base, metastatic tumors, plasmacytoma and brown tumor in the calvarium, as well as primary osseous tumors or metastatic lesions in the spine.

KEYWORDS: Amyloidosis, Amyloidoma, Calvarium, Hemodialysis, Spine

ÖZ

Amiloidosis protein komplekslerinin değişik dokularda ve özellikle ekstrasellüler mesafede anormal depolanmasıyla karakterize heterojen bir hastalık grubudur. Kemiklerde amiloid birikimi sistemik amiloidosis ve plazma hücre diskrezileri ile birlikte oldukça sık gözlenir. Kronik böbrek yetmezliği ve bu nedenle uzun süreli hemodiyaliz öyküsü olan hastalarda ß2 microglobulin klerensinin azalması kas-iskelet sisteminde aşırı amiloid birikimine neden olabilir. Kalvaryum ve/veya üst servikal vertebralarda amyloid birikimi klinik uygulamada oldukça nadir olup, tanı için sıklıkla kişisel öngörü ve özel histolojik boyama yöntemlerinin kullanılması gereklidir. Bu makalede sağ temporal kemik ve üst servikal vertebralarda amiloid birikimi olan bir olgu sunulmuş ve hastalığın etyolojisi, radyolojik bulguları ve ayırıcı tanısı ilgili literatür eşliğinde kısaca tartışılmıştır. Uzun süreli hemodiyaliz öyküsü olan veya plazma hücre diskrezileri ya da uzun süreli kronik enflamatuar hastalık geçiren kişilerde kas-iskelet sisteminde anormal amiloid birikimi öncelikle düşünülmelidir. Kafa kaidesinin benign veya malign mezenkimal tümörleri, kalvaryal metastatik tümörler, plazmasitomlar ve kahverengi tümörler, yine vertebraların birincil veya metastatik tümörleri ayırıcı tanıda yer alan başlıca lezyonlardır.

ANAHTAR SÖZCÜKLER: Amiloidosis, Amiloidoma, Kalvaryum, Hemodiyaliz, Omurga

INTRODUCTION

Amyloidoses are a group of diseases associated with the abnormal aggregation of proteins into insoluble amyloid fibrils in various tissues. Despite the important role of its local deposition in many diseases such as Alzheimer and Diabetes Mellitus, systemic amyloidosis can be classified into four major groups based on the type of involved precursor protein. Clinical manifestation may be due to visceral involvement and eventual organ dysfunction caused by disrupted tissue architecture (amyloidosis) or may be due to mass effect of its rarer local form (amyloidoma). Amyloid deposition in the cranium and/or the central nervous system (CNS) is a rare clinical entity and may be localized in the cerebral white matter, cerebellopontine angle, the pituitary gland, the skull base, the orbit and temporal bone (1).

CASE REPORT

A 47-year-old male patient was evaluated at an outside center with a complaint of headache. He was referred to our hospital with a pre-diagnosis of the right temporal bone neoplasm documented with a magnetic resonance imaging (MR) study. He had a history of chronic renal failure and peritoneal dialysis treatment for the last 14 years. His initial neurological examination was completely normal. The results of complete blood count and biochemistry showed good correlation with the presence of chronic renal failure and hypercalcemia. His MR study revealed an extraaxial mass lesion of bony origin measuring 28x47x45 millimeters in diameters. The lesion compressing the temporal parenchyma with no associated edema was hyperintense on T1-weighted (W) and isointense on T2W images peripherally. A profoundly dark center on both T1 and T2-weighted (W) images was also remarkable (Figure 1 A and B). The enhancement pattern was not identified since no contrast material had been given during the examination. The whole calvarium was thick and had increased trabeculations, which gave the typical salt and pepper appearance of secondary hyperparathyroidism on plain x-rays. The temporal lesion was radiodense with an irregular contour (Figure 1C). His computed tomography (CT) showed an expansile mass lesion with increased thickness of the inner and outer table of the squamous temporal bone. Dense calcification was observed in the tumor matrix (Figure 1D). Additionally, there were well-demarcated focal radiolucent lesions with mild expansion in the left lateral mass of the atlas and odontoid tip (Figure 1E).

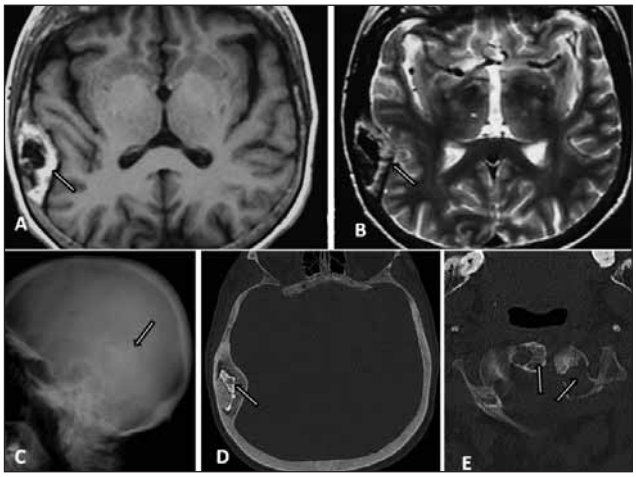

Figure 1: *(A-E). Axial T1-weighted (W) spin-echo (SE) (TR/TE: 600/20ms) (**A**) and T2-weighted (W) turbo SE (TR/TE:4000/100ms) (**B**) show an expansive bony lesion with hyperintense (T1-W) and isointense (T2-W) periphery and dark center (arrows). The lesion has a mass effect on the temporal parenchyma. On lateral plain film of the skull there is thickened calvarium with increased trabeculation (**C**). Irregular contoured calcification is also remarkable (arrow). An axial noncontrast computed tomography (CT) imaging reveals a fusiform lesion that involves inner and outer tables and shows heavily calcified matrix in the squamous part of the right temporal bone (**D**, arrow). Wel-demarcated focal radiolucent lesions in C1 and C2 vertebrae were additionally observed (**E**, arrows).*

Widespread vascular tramline calcification was observed in the soft tissues of the head and extremities.

He underwent a surgery for complete resection of the lesion and concomitant acrylic cranioplasty with a pre-diagnosis of brown tumor. His postoperative course was uneventful. Routine histopathological examination and additional studies with Congo red and crystal violet dyes verified the diagnosis of an amyloidoma (Figure 2). After histopathological confirmation, radiological and pathological screening of the patient with colonoscopy, bone marrow and rectal biopsies, Tc99m bone scintigraphy, chest and abdominal CT, echocardiogram and abdominopelvic ultrasonography revealed no evidence of systemic involvement other than the cervical lesions described above. Protein electrophoresis showed ß2microglobulin excess supporting the diagnosis of dialysis-related amyloidosis.

DISCUSSION

Amyloidosis forms a heterogeneous group of disorders characterized by the extracellular deposition of an insoluble protein complex. Serum amyloid A (SAA) protein, monoclonal immunoglobulin light

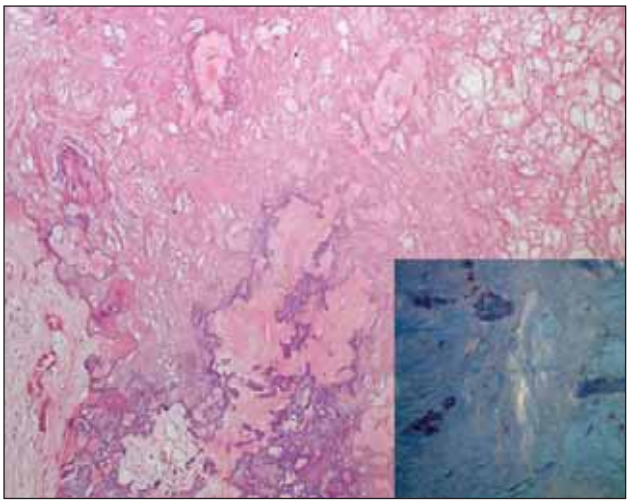

Figure 2: *The amyloid deposits appear orangiophilic with Congo red stain (x100). Congo red stained sections give typical apple-green birefringence under polarized light (lower-right corner).*

chains, transthyretin and Aß2 microglobulin are the constitutive proteins of four major types of systemic amyloidosis (4). Besides, there are suggestive data for the promoting role of serum amyloid P, interaction with collagen and/or charged glucosaminoglycans and decreased proteolytic activity in the formation of insoluble amyloid fibrils from these protein complexes in vivo (8). SAA protein is the constitutive protein of reactive amyloidosis that was formerly known as secondary amyloidosis. Its accumulation, as an acute phase reactant, is usually seen after longstanding inflammatory processes such as rheumatoid arthritis. AL amyloidosis, formerly known as primary amyloidosis, is caused by monoclonal plasma cell dyscrasia and develops from accumulation of immunoglobulin lambda light chains. Hereditary (ATTR) amyloidosis is derived by point mutations of transthyretin. These three major types of amyloidosis usually manifest with systemic organ involvement, especially of the liver, heart, kidney, respiratory and gastrointestinal system (1,4).

Aß2 amyloidosis is typically seen in patients with the history of chronic renal failure and long-term dialysis therapy. The exact mechanism is not known, however decreased clearance of ß2 microglobulin is believed to be the main responsible factor in the pathogenesis. Excessive deposition of insoluble, ß2m typically affects the musculoskeletal system and causes destructive spondyloarthropathy. Carpal tunnel syndrome, scapulohumeral and/or atlantoaxial arthropathy and bursitis are frequently encountered disorders in these patients(10). Bone cysts around joints and thoracic vertebrae were also reported, and can cause pathological fractures(4). Visceral involvement is possible, but are a less frequent finding in Aß2 amyloidosis than the other types of systemic amyloidosis.

Intracranial focal amyloid deposition is extremely rare in clinical practice and may be seen in the brain parenchyma, cerebellopontine angle, pituitary gland and skull base. Parenchymal involvement is usually seen in AL amyloidosis, and may resemble a primary brain lymphoma or malignant glioma (6). Although rare, the amyloid deposits in the calvarium have a predilection for the skull base. They have been described as a destructive bony lesion usually with scattered areas of calcification (11). Amyloid fibrils frequently contain large amounts of divalent sodium and calcium-bound proteins that give its characteristic calcified appearance on plain film and CT scans. The main determinant of MR characteristics is the heterogeneous composition of the lesion. The MR signal characteristics of solitary amyloidomas in the head and neck region have been previously described as being similar to those of skeletal muscle (3). Low signal intensity of the central area in both T1 and T2-W MR images corresponds to the inner calcified matrix that was verified with CT scan in the present case. Surrounding hyperintensity in T1- and iso or hypointensity in T2-weighted images may be due to poor fluid structure and different diamagnetic sensitivities of collagen and amyloid fibrils (2). Although it was not done in the present case, various degrees of enhancement of the mass were reported in the literature (5,11). These lesions show an expansile growing pattern frequently limited in the inner and outer tables. The absence of meningeal involvement and associated edema are also important radiological findings supporting the diagnosis. Chondroma and/or chondrosarcoma, metastatic tumors of the skull, brown tumor, hemangiomas, histiocytosis, plasmacytoma or other benign mesenchymal neoplasms of the dura and skull base are the most frequently encountered lesions in differential diagnosis. Chondroid tumors are expansive, lobulated, soft tissue masses with endosteal bone resorption, and have a tendency to involve the skull base. These lesions have high signal intensity on T2-W images. Heterogeneous contrast enhancement gives them a typical appearance of a honeycomb pattern in MR images (11). Brown tumors are seen as local destructive lesions in CT and MR studies, and the

presence of associated hyperparathyroidism is a characteristic finding. Histiocytosis is a granulomatous lesion frequently involving the skull and brain. Radiological findings include the presence of a well-demarcated lytic lesion with an adjacent enhancing mass. Solitary plasmacytoma and/or multiple myeloma are plasma cell dyscrasias and often show dissemination at the time of diagnosis. Osteomas are usually asymptomatic dense lesions of the outer or inner table of the skull. The absence of lysis and diploic involvement is typical (9). Amyloidomas of the spine may be primary or secondary, and can cause compression fractures leading to paraplegia (7). These lesions have a predilection for the thoracic and less frequently cervical region in the spine. They are frequently seen as enhancing radiolucent lesions in CT and MR studies.

Diagnosis requires histological examination with specific staining of the tissue. Histological examination shows fibrillar protein deposition, which is characteristically stained with Congo-red dye and consequent apple-green birefringence under polarized light. After histological verification, the second step must be to elucidate presence of systemic amyloidosis. Radiological and hematological screening and abdominal or rectal fat biopsy are mandatory for evaluation. When systemic involvement is established, identification of precursor protein is crucial for determination of the appropriate therapy (4). As opposed to systemic amyloidosis, solitary amyloidomas are accepted as benign, slowly growing lesions, and the treatment is usually limited to elimination of underlying primary factors and surgical decompression if necessary. In Aß2 amyloidosis, renal transplant is the best approach to avoid long-termed dialysis. The use of high performance dialysis membranes may delay the progression of excessive amyloid deposition and its associated symptoms in these patients (10).

The diagnosis requires a high index of suspicion due to the diverse radiological characteristics. A history of long-term dialysis therapy, plasma cell dyscrasias or long-standing inflammatory diseases may help to direct the physician towards a possible amyloidoma diagnosis in cases with radiological findings similar to those presented here.

REFERENCES

1. Caerts B, Mol V, Sainte T, Wilms G, Van Den Bergh V, Sressens L: CT and MRI of amyloidoma of the CNS. Eur Radiol 7:474-476, 1997
2. Gandhi D, Wee R, Goyal M: CT and MR imaging of intracerebral amyloidoma: Case report and review of the literature. Am J Neuroradiol 24:519-522, 2003
3. Gean-Marton AD, Kirsch CFE, Vezina LG, Weber AL: Focal amyloidosis of the head and neck: Evaluation with CT and MR imaging. Radiology 181:521–525, 1991
4. Hazenberg BP, van Gameren II, Bizjet J, Jager PL, van Rijswijk MH: Diagnostic and therapeutic approach of systemic amyloidosis. Neth J Med 62:121-128, 2004
5. Hidalgo F, Aguilera C, Monfort JL, Rene´ M, Muntane´ A, Pons LC: Amyloidoma of the skull: Plain radiographs, CT and MRI. Neuroradiology 38:44–46, 1996
6. Lee J, Krol G, Rosenblum M: Primary Amyloidoma of the brain: CT and MR presentation. AJNR 16:712–714, 1995
7. Mizuno J, Nakagawa H, Tsuji Y, Yamada T: Primary Amyloidoma of the thoracic spine presenting with acute paraplegia. Surg Neurol 55(6):378-381, 2001
8. Morten IJ, Gosal WS, Radford SE, Hewitt EW: Investigation into the role of macrophages in the formation and degradation of ß2-microglobulin amyloid fibrils. J Biol Chem 282:29691-29700, 2007
9. Osborn AG, Blaser SI, Salzman KL: Diagnostic Imaging Brain. Salt Lake City: Amirsys, 2004: (II:4) 48-83
10. Saito A., Gejyo F: Current clinical aspects of dialysis-related amyloidosis in chronic dialysis patients. Ther Apher Dial 10:316-320, 2006
11. Simoens WA, van den Hauwe L, Van Hedent E, Warson F, Demaeseneer R, Williams D, De Schepper AM: Amyloidoma of the skull base. Am J Neuroradiol 21:1559–1562, 2000