Case Report

Vertebral solitary bone plasmacytoma in a young adult with Trisomy 21: A case report

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Context: Solitary bone plasmacytoma (SBP) are rare lesions, accounting for less than 5% of all plasma cell proliferations. We describe a case of a 21-year-old female with Trisomy 21 presenting with cauda equina compression from an SBP.

Findings: Solitary bone plasmacytoma (SBP) is a rare primary bone tumor. It is characterized by monoclonal proliferation of malignant plasma cells localized to a bone segment, without signs of systemic invasion. The vertebral location is the most common. It preferentially affects men during their 5th or 6th decade.

Clinical relevance: We report the first association between solitary bone plasmacytoma and Trisomy 21.

Keywords: Trisomy 21, Plasmacytoma, Extramedullary plasmacytoma, Solitary bone plasmacytoma, Cauda equina syndrome

Introduction

Solitary bone plasmacytoma (SBP) is a rare clinical entity, arising from the malignant proliferation of plasma cells. It accounts for approximately 3–5% of all plasma cell malignancies.¹ SBP is characterized by the presence of a single bone lesion. SBP is more common in males and is rarely reported before 40 years of age.²

We describe the case of a 21-year-old female with Trisomy 21 presenting with cauda equina compression from an SBP. To the extent of our knowledge, this is the first time this association has been described in the literature.

Case report

A 21-year-old female with Trisomy 21 was admitted to our neurology department with acute weakness and paresthesias in the two lower limbs. No bowel or bladder dysfunction was reported. Neurological examination elicited mild intellectual disability, flaccid paraparesis, posterior cord syndrome identified by loss of proprioceptive sensation and diminished vibration sense in the lower extremities. No sensory level was found. We noted localized lumbar

paraspinal muscles contraction with provoked pain in palpation of spinous processes. Cauda equina compression was suspected. A lumbar spine MRI was emergently performed. It showed a tumor mass invading the vertebral body and the posterior arch of the L4 vertebra, associated with invasion of the epidural space extending 55 mm in height. This process invaded almost the entire canal lumen and the cauda equina. The lesion was hypointense on T1 and T2 sequences, and enhanced intensely after injection of Gadolinium contrast (Fig. 1).

Our patient underwent emergent lumbar 3–5 laminectomy with careful debulking of the tumor. The gross appearance was that of a grayish epidural tumor with some hemorrhagic changes and bone infiltration. Histological examination showed a dense and diffuse proliferation of round, monomorphic tumor cells with plasmacytoid appearance (Fig. 2(A)). Tumor cells were strongly positive for CD138 and negative for keratin on immunohistochemistry. Keratin negativity excluded epithelial origin and plasmacytoma was retained [Fig. 2(B)].

Laboratory analysis showed hypoalbuminemia at 46.7 g/l, an elevated globulin protein without a monoclonal gamma spike on serum protein electrophoresis. The immunoelectrophoresis of plasma proteins revealed an IgA monoclonal spike with light chain kappa. Blood count, renal function, 24 h proteinuria and Bence Jones

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Figure 1 Lumbo-sacral MRI: (a) T1 Sagittal section, (b) T2 sagittal section, (c) T1 Gadolinium-enhanced sagittal section, (d,e) T1 Gadolinium-enhanced axial section, (f) T2 axial section showing tumor mass invading the vertebral body and the posterior arch of L4. The lesion is hypointense on T1 and T2 sequences, and enhances intensely after injection of Gadolinium contrast.

proteinuria were normal. The erythrocyte sedimentation rate (ESR) was elevated at 55. The phosphocalcic balance was normal. Radiographs of the flat bones: (skull, pelvis, rib cage, femur, vertebrae, clavicle and the humerus) were normal with the exception of a destruction of L4 right pedicle.

To differentiate a multiple myeloma from an isolated plasmacytoma, we completed a bone marrow biopsy. It showed plasma cell infiltration equal to 3%. The diagnosis of vertebral SBP with the extensive epidural extension was retained. Our patient received, one month after surgery, local radiation with a dose of 40 Gy in 20 fractions. Target volume included 2 healthy vertebral levels above and below L4 (L2 to S1). She remained disease free from completion of radiation therapy. Clinically neurological examination was normal with no motor deficit nor sensitive disturbance, laboratory

analysis showed the absence of monoclonal gammopathy after radiation. Control MRI was not practiced. Our patient has not yet reached 1 year after radiation and the follow-up period was 8 months.

Discussion

Trisomy 21 occurs in 1 out of every 700–1000 live births.³ Individuals with Trisomy 21 have an increased risk of developing acute leukemia. It has been estimated to occur 20–30 times more frequently than in the unaffected population.⁴ The distribution of cancers in patients with Trisomy 21 is unique, with a very high risk of leukemia in childhood and young adulthood, and a decrease in the risk of solid tumors at all ages.⁵ An association between Trisomy 21 and plasmacytoma has not been previously described, and we believe our case to be the first in publication.

Figure 2 (A) Histological examination: showing proliferation of round and monomorphic tumor cells with a plasmacytoid appearance. Tumor proliferation invades bone () (Hematoxylin and Eosin; 200x magnification). (B) Immunohistochemical study: strong positive staining of tumor cells for CD138 (200x magnification).

Plasma cell disorders account for 1–2% of all human neoplasms, with solitary plasmacytoma (SP) constituting less than 10% of these.⁶ These tumors can be categorized into two groups: SBP (also called intra-medullary plasmacytoma), and extramedullary plasmacytoma. Bone forms of solitary plasmacytomas are the more frequent.⁷ The most common affected bones are those with active bone marrow hematopoiesis such as the vertebrae, ribs, skull, pelvis, femur, clavicle and scapula.

The spine is a frequent location for SBP, representing 25–60% of the cases.⁸ SBP is defined as a focal lesion composed of malignant plasma cells but without diffuse medullary invasion, which distinguishes it from multiple myeloma. The diagnostic criteria for SBP include: histological evidence of the absence of other lesions on skeletal radiographs, the absence of bone marrow invasion (medullary plasmocytosis less than 10%), and no evidence of disseminated disease.⁷ Clinical symptoms are dominated by bone pain, neurological signs, and pathological fractures. SBP preferentially affects men during their 5th or 6th decade.⁹

In our case, SBP was diagnosed as the cause of spinal cord compression in a 21-year-old woman. The age of patients with these diseases is 10 years younger than those with multiple myeloma. According to the literature the incidence of spinal cord compression has been reported in 71% of the cases of SBP, in contrast to 7.4–16% in cases of multiple myeloma. A monoclonal component is present at diagnosis in 43–55% of cases.⁷ Our case demonstrated monoclonal expansion.

The standard treatment for SBP is radiotherapy at doses between 40 and 50 Gy, with a local control rate between 70% and 100%. The radiation must cover the entire tumor mass with safety margins of at least 2 centimeters of healthy tissue.¹⁰ In vertebral locations, the target volume should interest one or two healthy vertebral levels above and below the lesion level.¹¹ Surgical treatment may be indicated for diagnostic purposes, in the case of a neurological complication such as spinal cord compression as in of our patient, or in order to treat or prevent a pathological fracture on a bone weakened by tumor osteolysis.

The progression of SBP is represented by local recurrence, lymph node involvement, and/or multiple myeloma transformation. The average time from onset to recurrence is 2 years, and all previously described recurrences occur during the first five years after treatment.⁷ The follow-up period of our patient was 8 months after radiotherapy, which is a limitation of our study. The main predictors of recurrence are tumor size and dose of radiotherapy delivered, with tumors larger than 5 cm and tumors treated with a radiotherapy dose of 35 Gy or less being more likely to recur. Regarding transformation into multiple myeloma, the most important predictors in the literature are age greater than 63 years, the persistence of a monoclonal gammopathy after radiotherapy, intramedullary variety, vertebral location, insufficient dose of radiotherapy and tumor size greater than 5 cm.^{6,12}

The factors of poor prognosis in our patient are represented by the vertebral location of SP and the intramedullary variety. Young age is usually considered a favorable prognostic factor in respect to progression in overt multiple myeloma.¹³

Trisomy 21 is a chromosomal abnormality that has been known to predispose to hematological malignancies, mainly leukemias. In reviewing the literature, no cases associating SBP and Trisomy 21 were described.

Conclusion

SBP is a rare malignant tumor that often reaches the axial skeleton. Our case is the first described in the literature associating Trisomy 21 and the young age of onset with SBP. Radiotherapy is the standard treatment,

although surgical treatment may be needed in cases of spinal instability or neurological compression. The prognosis is affected by the progression to multiple myeloma, which justifies rigorous monitoring after treatment.

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Conflicts of interest Authors have no conflicts of interest to declare.

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