Recurrent Extraspinal Myxopapillary Ependymoma with NTRK Fusion: A Case Report

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Background

Myxopapillary ependymoma (MPE) is a rare, slow-growing tumor typically arising in the filum terminale. Extraspinal manifestations are uncommon and can pose diagnostic and therapeutic challenges. Here, we present the case of a male patient with a gluteal mass diagnosed as MPE, followed by recurrence and systemic progression despite multimodal treatment, including surgery, radiotherapy, and targeted therapy

Case Presentation

A 45-year-old male presented with a progressively enlarging, painless mass in the right gluteal region. Physical examination revealed a firm, non-tender subcutaneous lesion. The mass was surgically excised, and histopathological examination confirmed the diagnosis of myxopapillary ependymoma (WHO grade I).

Follow-up imaging with whole-spine and brain MRI at eight months revealed local recurrence in the gluteal and sacral regions. A whole-body FDG-PET scan demonstrated high metabolic activity in the sacrum and inguinal lymph nodes. Due to significant post-surgical scarring and functional concerns, surgical resection of the recurrent sacral and gluteal lesions was deemed inappropriate. Instead, the patient underwent inguinal lymphadenectomy, with histopathological confirmation of recurrent myxopapillary ependymoma.

Given the recurrent nature of the disease, radiotherapy was initiated. Next-generation sequencing (NGS) analysis identified an NTRK gene fusion, leading to the initiation of targeted therapy with larotrectinib. The patient demonstrated stable disease under treatment. However, after eight months, follow-up imaging detected disease progression with new pulmonary metastases.

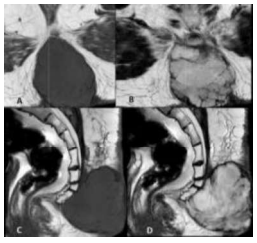

Fig1 It documents the presence of a large extra-axial expansive lesion located in the subcutaneous tissues of the gluteal region, denoted by a high signal in T2 images (B-D) which extends in depth to touch the sacru

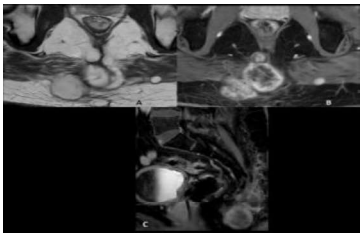

Fig2 *After surgery, multiple nodules of lesions regrew along the surgical incision, scattered both in the subcutaneous tissue and in the context of the gluteal muscles. Recurrent nodules are characterized by high signal on T2-weighted images (A) and by inhomogeneous contrast enhancement (B-C).*

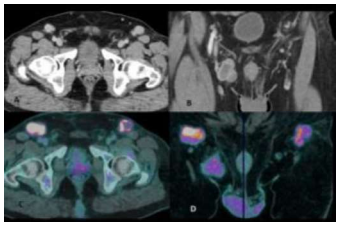

Fig3 *Pathological lymph nodes are identified, necrotic and dimensionally increased on CT* ***(A-B)*** *and hypermetabolic on PET-CT scan* ***(C-D).***

Conclusion This case highlights the challenges of treating recurrent extraspinal myxopapillary ependymoma, the potential role of targeted therapy, and the need for close disease monitoring. The presence of NTRK fusion guided treatment decisions, but eventual disease progression underscores the necessity for further research into novel therapeutic strategies for recurrent and metastatic MPE.

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