Evaluation of Gait Abnormalities in Patients with a History of Retinoblastoma

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INTRODUCTION

- Retinoblastoma (Rb) is the most common primary ocular malignancy of childhood, with a reported incidence of approximately 1 in 15,000. Rb can be inherited or develop insidiously and can be fatal if untreated. Due to advances in recognition and treatment, current survival rate is >95%.
- The incidence of metastatic Rb is rare in developed countries, occurring in about 5% of cases. This can make recognition and diagnosis of these cases challenging. The treatment outlook for patients with metastatic Rb is less promising than that of primary malignancy and varies depending on the extent of disease dissemination. Current studies estimate survival rates of 80-90% for patients with metastatic disease without central nervous system (CNS) involvement, but that falls to just 8% if the CNS is afflicted.

Stage	Extent of Disease Dissemination	Event-Free Survival 36 Months Post-Therapy
2 or 3	Regional extra-ocular disease	88%
4a	Overt distant metastasis (bone, bone marrow, and/or liver) not involving CNS	79%
4b	Patients with CNS disease	8%

Figure 1: Survival rates for 57 patients diagnosed with metastatic retinoblastoma based on extent of disease dissemination at initiation of treatment.

CASE PRESENTATION

- **Case 1**: 5-year-old female with a history of Group E, left unilateral Rb initially treated with intra-arterial chemotherapy (IAC), and ultimately enucleation.
  - Presented to her primary care physician seven weeks post-enucleation due to low back that caused her to limp a few days earlier. Clinical exam was unremarkable, so no further evaluation was recommended.
  - She returns to clinic six weeks later with recurrence of her low back pain and new-onset fever over the last 2-3 days. Limp was noted on clinical exam. Labs were ordered. Instructed to return to clinic in 3 days for lab results.
  - Upon return for lab results, the patient appears lethargic. Fever has persisted. Patient sent to Cook Children's Medical Center for urgent evaluation. CT and MRI suspicious for osseous metastatic disease of the spine, pelvis, and thoracic cage. Diagnosis of metastatic Rb confirmed with biopsy.

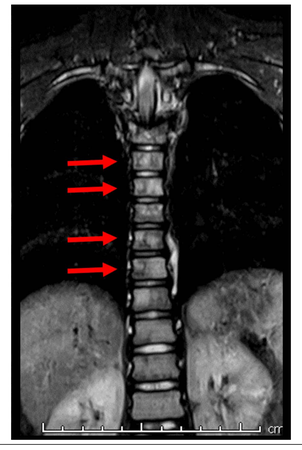
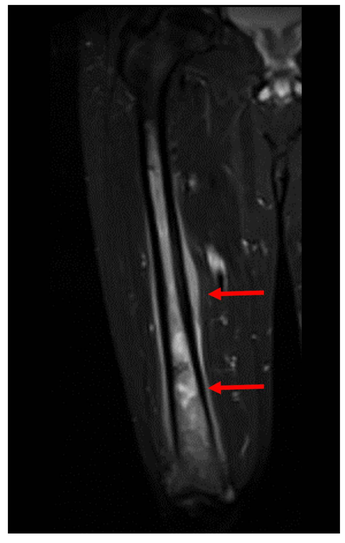


Figure 2: Coronal MRI image of the thoracic spine showing extensive osseous metastatic disease involving multiple vertebral bodies.

CASE PRESENTATION

- **Case 2**: 15-year-old male with a history of bilateral Rb that was diagnosed and treated in infancy.
  - Presented Emergency Department complaining of intense thigh and knee pain. X-rays of the thigh and knee were obtained and reported as normal. He was diagnosed with patellofemoral syndrome and no further evaluation was recommended.
  - Symptoms progressed over the following four months to include intermittent fevers and worsening leg pain.
  - Patient referred to orthopedic oncology where further imaging was performed. Diagnosis of metastatic Rb with dissemination to bone confirmed with biopsy.

Figure 3: Coronal MRI image of right femur showing abnormal signal and periosteal reaction.



CONCLUSION

- The prognosis of metastatic retinoblastoma correlates highly with the extent of disease dissemination. Therefore, early recognition and initiation of treatment is essential to enhance patient survival. Episodes of non-specific bone pain were reported as initial complaints in these two cases. This provoked gait abnormalities either reported by the patients or their families. These findings indicate that an insidious onset of gait abnormalities in patients with a history of Rb may be an early indicator of metastatic disease and warrants prompt evaluation.

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