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Case Report

Burkitt's Lymphoma in A 14-Year-Old Boy with Jacob's Syndrome (47, XYY): A Rare Case Report

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Abstract

Background: Burkitt's Lymphoma (BL) is an aggressive B-cell non-Hodgkin lymphoma characterized by MYC gene rearrangement. Jacob's syndrome (47, XYY) is a rare chromosomal abnormality in males, typically presenting with tall stature, behavioral challenges, and learning difficulties. The coexistence of BL and 47, XYY syndrome is exceptionally uncommon.

Case presentation: We report a 14-year-old boy diagnosed with stage IV Burkitt's lymphoma presenting with paraplegia secondary to a large extradural spinal mass (D5-D7). Surgical excision confirmed BL with MYC and IGH rearrangements. Karyotyping identified a 47, XYY chromosomal pattern. The patient received treatment per the COG-ANHL1131 protocol and achieved complete metabolic remission.

Conclusion: This case highlights the rare association of Burkitt's lymphoma with Jacob's syndrome and underscores the importance of genetic evaluation in pediatric hematologic malignancies with atypical clinical or behavioral presentations.

Keywords: Burkitt's lymphoma; Jacob's syndrome; 47, XYY; Spinal cord compression; Pediatric lymphoma; MYC rearrangement

Introduction

Burkitt's Lymphoma (BL) is one of the fastest-growing human tumors, accounting for approximately 30% - 50% of childhood non-Hodgkin lymphomas. It is characterized by a chromosomal translocation involving the MYC oncogene, most commonly t (8;14) (q24; q32), leading to dysregulated cell proliferation. BL has three clinical subtypes: endemic, sporadic, and immunodeficiency-associated.

Jacob's syndrome (47, XYY) is a sex chromosome aneuploidy affecting approximately [1] in 1,000 males. It results from an extra Y chromosome and is often underdiagnosed due to its subtle phenotype. Clinical features include tall stature, acne, learning difficulties, and variable behavioral traits such as impulsivity or attention-deficit symptoms.

The coexistence of Burkitt's lymphoma and Jacob's syndrome (47, XYY) has not been previously reported in the literature, making this the first documented case to our knowledge [2]. This report presents a case of stage IV BL in a 14-year-old male with Jacob's syndrome, highlighting diagnostic, genetic, and therapeutic aspects.

Case Presentation

Clinical Presentation

A 14-year-old previously healthy boy presented in May 2024 with progressive lower limb weakness following a football injury. Over several days, he developed paraplegia, urinary retention, and constipation. Neurological examination revealed grade 2-3/5 lower limb power, sensory loss from T10 downward, and loss of anal sphincter tone [3].

Radiologic findings

Magnetic Resonance Imaging (MRI) of the spine demonstrated a large extradural mass at the D5-D7 levels, causing severe thecal sac compression. The lesion appeared highly vascular and adherent to the dura. Surgical excision on May 5, 2024, achieved complete resection.

Pathologic and genetic findings

Histopathology of the excised mass showed sheets of medium-sized atypical lymphoid cells with a "starry-sky" pattern.

Immunohistochemistry (IHC): Positive for CD45, CD20, CD10, BCL-6, and PAX-5; Ki-67 proliferation index ≈100%.

FISH analysis

- MYC rearrangement positive (18%)
- IGH rearrangement positive (30%)

Bone Marrow Cytogenetics: Karyotype 47, XYY (Jacob's syndrome) confirmed on June 4, 2024 (Figure 1).

CSF Cytology: No malignant cells detected.

Genetic and Developmental Assessment

During hospitalization, the patient's unusually tall stature (183 cm, above the 97th percentile for age) and episodes of impulsive behaviour prompted genetic evaluation. Karyotyping confirmed Jacob's syndrome (47, XYY). No intellectual disability was reported, though mild behavioral disturbances and emotional lability were noted.

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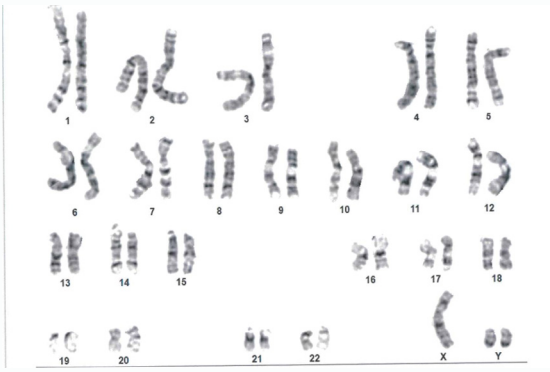

Figure 1: Magnetic Resonance Imaging (MRI) of the thoracic spine demonstrating a large extradural soft-tissue mass from D5-D7 causing severe spinal cord compression, consistent with lymphomatous involvement and correlating with acute paraplegia.

Treatment

The case was discussed in a multidisciplinary tumor board, and the patient was classified as Stage IV BL with CNS involvement. He was treated per the COG-ANHL1131 protocol, as follows:

- Rituximab: May 30, 2024
- R-CVP (Cyclophosphamide, Vincristine, Prednisone): June 5, 2024
- R-COPADM Cycles 1-2: June-July 2024
- Intrathecal Methotrexate and Cytarabine (R-IT-CYVE): June and July 2024
- Maintenance Phase (R-CYM): September-November 2024

Clinical course and follow-up

Postoperatively, the patient developed transient paraplegia secondary to reperfusion injury, which improved gradually. After chemotherapy, lower limb strength improved to grade 3-4/5, bladder function recovered, and bowel control partially returned.

PET/MRI (December 3, 2024)

- No hypermetabolic or FDG-avid lesions
- Complete metabolic remission

At the latest follow-up (October 2025), the patient remained in remission and ambulant without support.

Discussion

The co-occurrence of Burkitt's lymphoma and Jacob's syndrome is exceedingly rare. While the MYC rearrangement is the primary driver of BL, chromosomal abnormalities such as XYY may contribute to genomic instability, increasing susceptibility to oncogenesis [4,5].

The XYY karyotype is associated with increased height, acne, and mild neurodevelopmental and behavioral differences. Although once erroneously linked to aggression, recent data suggest that behavioral symptoms stem from social and cognitive challenges rather than inherent violent tendencies.

Few cases have described hematologic malignancies in 47, XYY individuals. Reported cases include acute myeloid leukemia, chronic myeloid leukemia, and rarely, lymphomas. The mechanism remains unclear but may involve altered hematopoietic stem cell function or secondary genomic instability from sex chromosome aneuploidy.

Our case demonstrates that comprehensive genetic testing can uncover underlying chromosomal anomalies influencing disease biology and patient management. In pediatric and adolescent patients with atypical stature or behavioral features, a karyotype may provide valuable insight.

Conclusion

This case highlights the rare coexistence of Burkitt's lymphoma and Jacob's syndrome (47, XYY) in a pediatric patient presenting with spinal cord compression. Early diagnosis, aggressive chemotherapy, and multidisciplinary care resulted in complete remission and neurological recovery. Clinicians should consider genetic evaluation in pediatric lymphoma cases with atypical physical or behavioral characteristics to enhance diagnostic precision and inform holistic care.

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