# Inpatient Rehab Course of a Patient with Rare Variant of Klinefelter Syndrome after Stroke: A Case Report

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## Introduction

- A 19-year-old male with a rare variant of Klinefelter syndrome (49,XXXXY) presented with slurred speech, right-sided weakness, and difficulty walking.
- Prior to admission, although patient had some cognitive delay due to Klinefelter syndrome, he was 80-90% independent with ADLs and ambulated with a wide based gait but did not require any assistive device. He was also able to ride his bicycle independently between his family members' houses on his family property.
- Patient was found to have an acute left internal capsular infarct with right hemiparesis, and hypercoagulable work-up showed elevated ANA and partially positive antiphospholipid panel.

## Case Report

- The patient was transferred to the acute inpatient rehabilitation unit, where he made significant progress from minimal to total assist with ADLs and ambulation on admission to set-up to minimal assist by discharge.
- A multidisciplinary approach was emphasized with a Child Life Specialist, Neurology, Rheumatology, and Hematology also following the patient.
- Post-discharge, he continued with outpatient therapies and has made almost full recovery of his strength and speech.
- He also continues to be monitored for central nervous system vasculitis associated with antiphospholipid antibody syndrome, and was started on immunosuppressive therapy (with mycophenolate mofetil) and long-term anticoagulant therapy.

## Results

***Figure 1:*** *Karyotype demonstrating 49,XXXXY variant of Klinefelter syndrome. Image credit: pathology.washington.edu.*

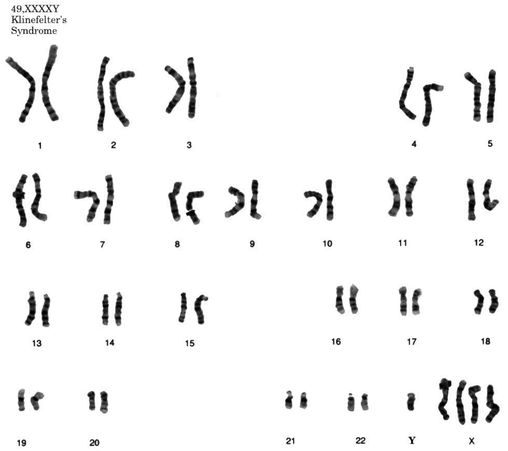
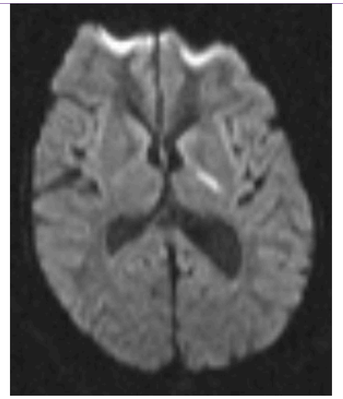



***Figure 2:*** *MRI brain showing acute infarction involving the posterior limb of the left internal capsule.*



## Discussion

Klinefelter syndrome is the most common sex chromosome abnormality, but the 49,XXXXY variant is the most rare type with a frequency of approximately 1 in 85,000 to 100,000 newborn boys. Compared to the classic 47,XXY phenotype, the 49,XXXXY variant presents with shorter stature and higher likelihood of craniofacial, cardiac, endocrine, and genitourinary abnormalities. Patients also have more severe learning disabilities and speech and motor delays. The mean age of diagnosis is 4 months by chromosome analysis. There are previous reports of the concurrence of Klinefelter syndrome with rheumatic and autoimmune diseases, in addition to higher risk of venous thromboembolism events, but few (if any) include patients with the 49,XXXXY variant. Rehabilitation and a multidisciplinary approach are essential for early recognition and management of complex comorbidities and complications.

## Conclusion

Klinefelter syndrome patients and especially those with the 49,XXXXY variant benefit from early intensive rehabilitation due to more severe congenital abnormalities and comorbidities including stroke.

## References

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