Communication is key for rare disease: A case study in factor X deficiency

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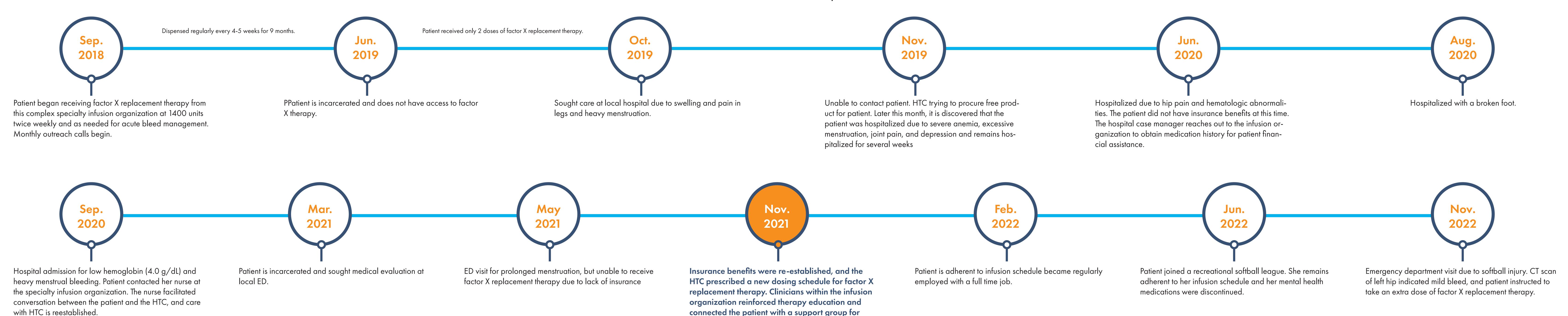


Objective

The purpose of this case study is to highlight how expertise in rare diseases and routine communication improved outcomes in a patient with factor X deficiency.

Methods

A multidisciplinary team conducted a retrospective review of medical documentation, including therapy-specific clinical assessments performed by the complex specialty infusion organization, hemophilia treatment center (HTC) visit notes, and hospital documentation from September 2018 to March 2023.



Summary

Patient is a 28 year-old female with severe factor X deficiency diagnosed at birth. She began receiving factor X replacement therapy from this complex specialty infusion organization in September 2018. The clinicians within the infusion organization performed monthly outreach calls for routine clinical assessments and medication dispensing activities. Frequent communication and education developed a rapport with the patient.

people living with factor X deficiency.

From June 2019 through October 2021, the patient's insurance coverage was inconsistent due to incarceration. Non-adherence to at-home factor X replacement therapy developed as a result, and she regularly sought care at local hospitals and emergency departments during the lapse of coverage. She suffered anemia with hemoglobin level as low as 4.0 g/dL, heavy and prolonged menstruation, and joint pain.

Routine outreach from the infusion organization still occurred despite inconsistent insurance coverage. Any communication between the infusion organization and the patient was conveyed to the HTC and the hospitals and emergency room departments where the patient sought out care. The infusion organization worked together with the HTC to communicate with and provide assistance to the patient in navigating insurance coverage after incarceration to re-gain access to outpatient therapy.

Insurance benefits were re-established in November 2021, and the HTC prescribed a new dosing schedule for factor X replacement therapy. Clinicians within the infusion organization reinforced therapy education and connected the patient with a support group for people living with factor X deficiency. In June 2022, the patient's mental health medications were discontinued, and she established regular employment and has been able to work 40 hours per week, she exercises regularly, and began playing sports.

Outcome	Before November 2021	After November 2021
Dispense rate	37% (14 dispenses over 38 months)	100% (18 dispenses over 17 months)
Hospitalizations/ED visits	7	
Wellness scale (Best: 5 – Worst: 25)	21	12
Hemoglobin level	4.0 g/dL (September 2020)	12.9 g/dL (December 2022)

Conclusion

Consistent patient contact coupled with clinical expertise positions complex specialty infusion organizations as instrumental healthcare partners in rare diseases, like factor X deficiency. Being a proactive member of the healthcare team and offering consistent support and disease education leads to positive patient outcomes.