A Missed Connection: Right-Sided Heart Failure as Noted in Congenital Diaphragmatic Hernia in a Patient with Turner Syndrome

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Introduction

Congenital diaphragmatic hernias (CDH) are both rare and surgically correctable when detected and managed at an early stage of development. Right-sided CDH when compared to left-sided CDH frequently is misdiagnosed as either elevation of the hemidiaphragm or as a mass rather than a herniation.

Patients with liver herniation secondary to CDH have an increased mortality rate when compared to other classifications of CDH, particularly when combined with additional anomalies or genetic concerns prognosis tends to decline. CDH is rare in cases of aneuploidy as seen in Turner Syndrome. Turner Syndrome is the most common chromosome abnormality occurring in approximately 1/2000 births causing loss of all or part of the X chromosome. At baseline, patients have a three-fold increase in mortality when compared to the general population; thus, with additional complications as seen with CDH, mortality increases with added cardiac and pulmonary complications.

Case Presentation

A 46 y/o female with history of HTN, Turner Syndrome, obesity, and chronic bilateral lower extremity (BLE) edema, status-post coarctation of aorta repair at age 10 presented to the ED with concerns of 2 weeks history of worsening BLE, nonproductive cough, dyspnea on exertion, and orthopnea. On presentation, she was tachypneic and tachycardic, normotensive, and was saturating 88-89% on room air. She had elevated Pro-BNP at 981 with a negative troponin. Patient was treated with Furosemide PRN along with head elevation and oxygenation via NC. A CXR and CT chest showed right hemi-diaphragmatic hernia with liver extending into the thoracic cavity (Figure 2 and Figure 3). Upon further discussion the patient's mother revealed that the patient has had a right hemi-diaphragmatic hernia with noted liver protrusion without surgical correction since 9 months of age (Figure 1).

Upon further work-up, echocardiogram revealed severely reduced RV function with normal LV function. Cardiac MRI showed RV hypokinesis and RV ejection fraction (EF) of 34%. Through chronic compression within the thoracic cavity, compensation was required to meet O2 demands, the heart failure that the patient was exhibiting was likely secondary to the genetic defect of the diaphragm with associated herniation. Further complication was noted secondary to patient's body habitus that was both impacted by the underlying Turner's syndrome in conjunction with the patient's obesity.

CT surgery was consulted, and the decision was made to not proceed with surgical intervention due to increased risk of mortality. Patient was discharged to home without any intervention. The patient has been re-admitted to the general medicine floor and medical ICU multiple times for a similar presentation since she was initially admitted and discharged from the Family Medicine service.

Imaging

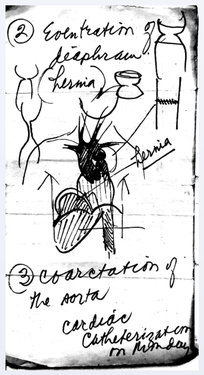

Figure 1

Figure 1: Handwritten note by the pediatrician depicting the patient's diaphragmatic hernia since 9 months of age with noted liver protrusion.

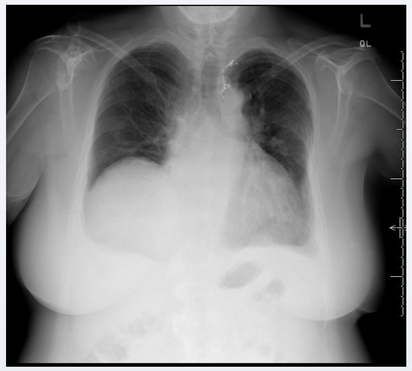

Figure 2a:

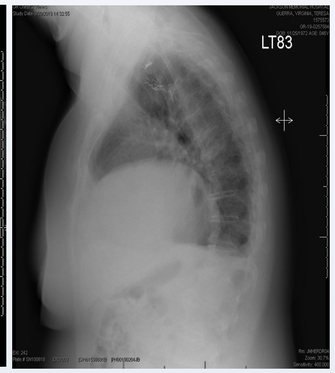

Figure 2b:

Figure 2a and 2b: CXR images showing right hemi-diaphragmatic hernia with liver extending into the thoracic cavity.

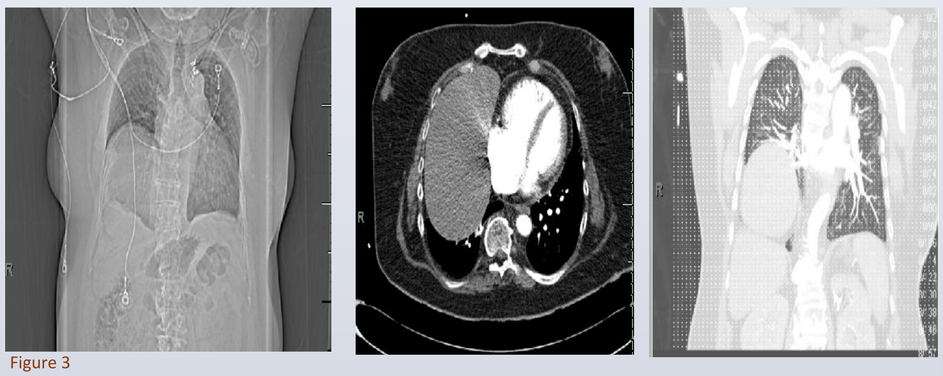

Figure 3

Figure 3: CT chest showed right hemi-diaphragmatic hernia with liver extending into the thoracic cavity

Discussion

The CDC estimates that CDH occurs in about 1/3600 births in the United States. The etiology is unknown, though it is believed to be due to abnormal fusion of pleuroperitoneal membranes during gestation. Surgery is necessary to repair the hernia soon after birth to avoid further complication. The CDH study group reported 63% survival rate in 1995-1996. It is known that there is a high rate of postanal death associated with CDH due to pulmonary hypoplasia and/or hypertension, the postnatal death rate is significantly higher if there is intrathoracic herniation of the liver.

Patients can remain asymptomatic during childhood and become symptomatic in adulthood with more negative outcomes, which was demonstrated by our current case. With worsening herniation, there is a risk of increased pulmonary hypoplasia and associated heart failure. Though with chronic interpretation of heart failure presentation without further investigation of underlying pathology, a chronic diagnostic delay had occurred.

Conclusion

As family physicians, early intervention and continuity of care during early diagnosis of Turner Syndrome complicated by CDH is recommended and is an integral part in allowing our patients to have the best possible prognosis . Though rare, detailed inspection of studies and hypervigilance in underlying genetic predisposition is crucial in management. Primary care physicians should anticipate that the patient will likely become symptomatic in the future with additional complications including those as debilitating as heart failure with little opportunity for correction and management outside of palliative and comfort interventions. In the case of our patient, loss to clinical follow up and poor health literacy resulted in multiple hospital readmissions and poor quality of life.

Key Points

- In patients with Turner Syndrome presenting with signs of heart failure, it is crucial to keep a broad differential including CDH.
- Primary care physician should anticipate that patients with Turner Syndrome with known hemi-diaphragmatic herniation will likely become symptomatic in the future. Both early intervention and continuity of care during childhood is recommended.
- Early intervention and continuity of care in children diagnosed with turner syndrome is necessary, and a multispecialty approach is key especially in the presence of additional anomalies.

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