

## Introduction

- 12-year-old girl seen via telehealth with a clinical history suggestive of noncardiac syncope.
- Deferred physical exam and screening diagnostics revealed undiagnosed partial atrioventricular (AV) canal requiring surgical repair.
- This case highlights the ease at which significant congenital heart disease (CHD) diagnosis may be missed if focused on clinical history alone.
- Physical examination and appropriate screening diagnostics are imperative for every congenital heart consult.

## Clinical Presentation and Management

- 12-year-old healthy female who presented via telehealth for initial evaluation by pediatric cardiology for two episodes of syncope.
- History obtained from patient and her mother reveals she had “passed out” twice over the past year while sitting on the toilet, straining to have a bowel movement.
- Both episodes were preceded by prodrome of palpitations, “vision going black,” and “ringing in her ears” with estimated duration of unconsciousness lasting 1 minute.
- Patient presented to the ED following most recent episode, was referred to pediatric cardiology when RVH was noted on ECG and murmur was present on physical exam. Mother reported a murmur had been auscultated at a check-up four years ago but it was not evaluated and had not been auscultated since.
- Based on history her episodes were most consistent with noncardiac syncope, likely due to increased vagal tone. Patient education was provided with plan for in clinic follow-up for repeat ECG and physical exam.
- On presentation to clinic she denied subsequent syncopal episodes but endorsed new onset dyspnea on exertion.
- Physical exam revealed a grade I/VI systolic ejection murmur at the left upper sternal border.
- Patient has since undergone surgical repair and is doing well.

## Imaging

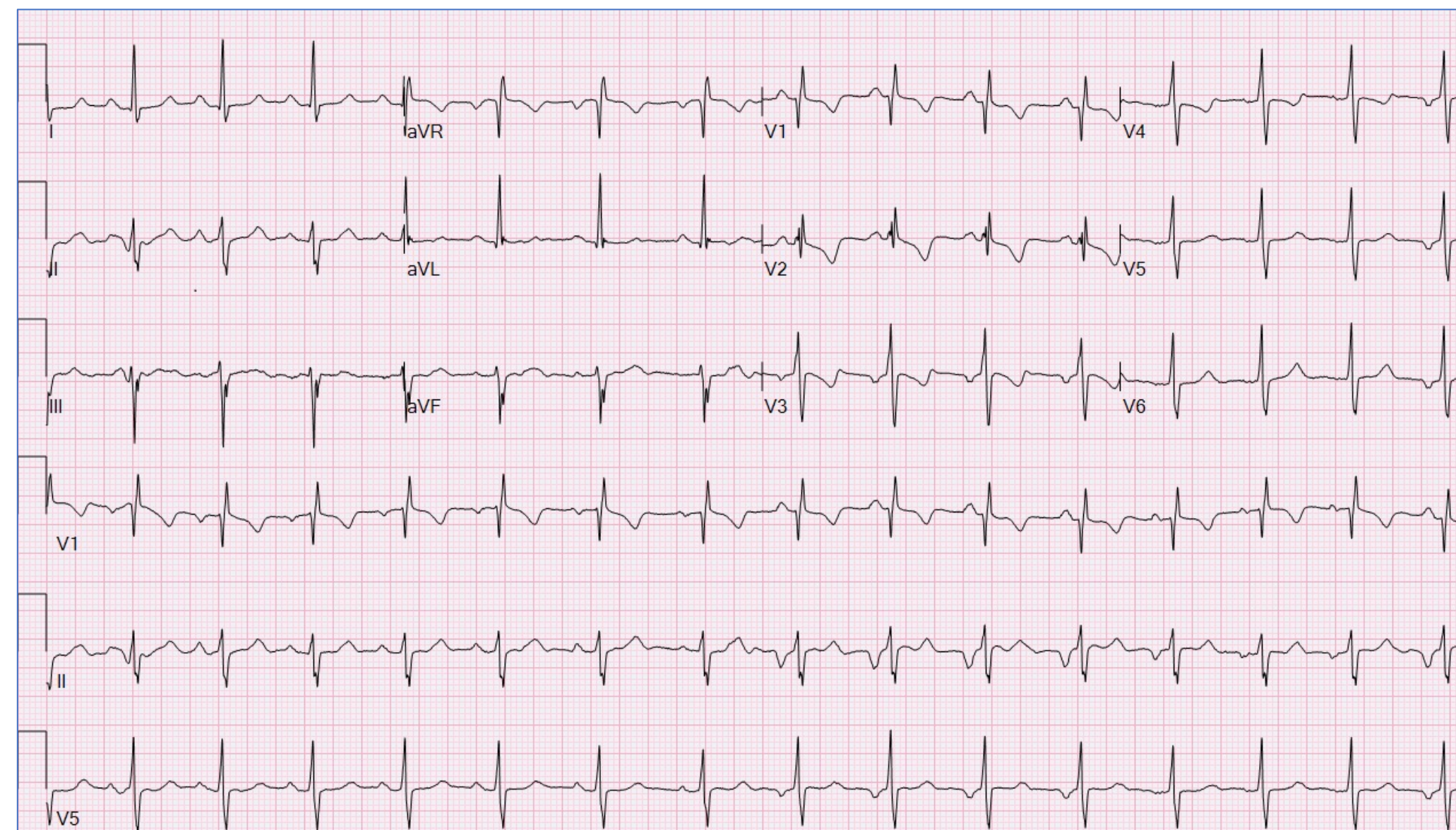


Figure 1. ECG demonstrating normal sinus rhythm with a northwest axis and right ventricular hypertrophy for age.

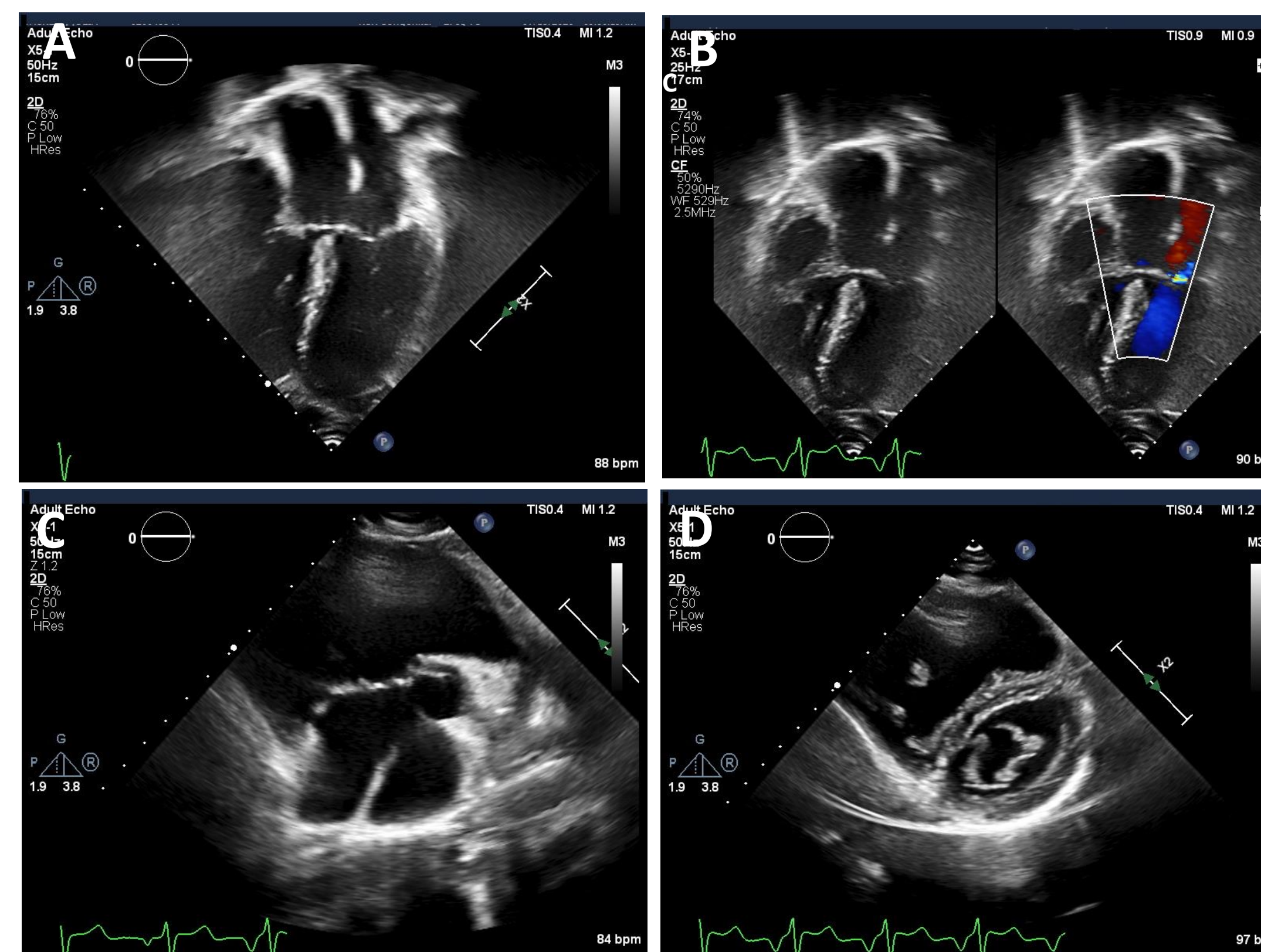


Figure 2. A) and B) show Partial Atrioventricular Canal with Primum ASD and cleft mitral valve, no VSD via apical imaging. C) Parasternal imaging of primum ASD, D) Parasternal short depicting mitral valve cleft.

## Discussion

- Even following the discovery of a hemodynamically significant congenital defect, we believe the patient's syncopal episodes to have been vasovagal in origin given their consistency with the classic description.
- This case of incidentally found partial AV canal could have continued to elude detection if reliance had been placed solely on a telehealth consultation without supportive physical exam or ECG, a course of action which could have easily seemed appropriate for a case of classic vasovagal syncope.
- Diagnostic delay occurs in 8.9% of CHD with 35% requiring immediate intervention because of compromised hemodynamic status <sup>1,2</sup>
- In all cases of late diagnosis, clinical cardiac findings were present that should have alerted examining physicians to the possible presence of underlying CHD <sup>1,2</sup>
- Diligence to clinical work-up will be imperative to prevent the increased use of telehealth from contributing to prevalence of missed diagnosis.
- Consulting providers must recognize the potential pitfalls of anchoring and confirmation bias in their medical decision making. Any attempt at diagnosis via telehealth is especially vulnerable to such errors.
- The preliminary impression based on the patient's chief complaint may not be “the right answer” when considering the entire body of evidence following full clinical work-up.

## Conclusion

- Telehealth has become a useful venue for evaluation in many patients
- New consults cannot be considered to have a full evaluation without supportive diagnostic tools such as physical exam, ECG and if appropriate echocardiogram.
- Incidental cardiac pathology may be missed if focused on the patient's chief complaint by history alone.

## References

1. Pfammatter, J.-P., & Stocker, F. P. (n.d.). *Delayed recognition of haemodynamically relevant congenital heart disease.*
2. Massin, M. M., & Dessy, H. (2006). Delayed recognition of congenital heart disease. *Postgraduate Medical Journal*, 82(969), 468–470. <https://doi.org/10.1136/pgmj.2005.044495>

## Acknowledgments

The authors would like to thank the patient and her family for allowing us to share her experience. We also thank our clinical staff at the University of Kentucky Congenital Heart Clinic for their effort and flexibility to ensure our patients could receive appropriate care during a time of great change in medicine during the beginning of the COVID-19 pandemic.