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New Onset Murmur or Hamman's Sign: Mid-systolic click in a Suspected case of Connective Tissue Disorder

Abstract

We present the case of a 15 y/o previously healthy male who presented to CHMC from an OSH with a 3 day history of shortness of breath, audible clicking noise, and chest discomfort after playing at the batting cages 4 days prior. No history of trauma. He was transferred to CHMC for further evaluation after identification of a pneumothorax on chest CT performed at OSH.

With respect to family history, the patient is one of 8 children. Of note, per discussion with family, his older siblings are all tall and have some connective tissue anomalies. Two siblings were noted to have developmental delay.

Physical exam was significant for the following: 4/6 mid-systolic click heard best at the left 5th intercostal when lying supine or left lateral decubitus, resolves with sitting up. Positive for 7 of the criteria for connective tissue disorder per the Ghent revised criteria including: wingspan to height ratio was 78" to height 76" (ratio 1.02), Pubic symphysis to floor 45", pectus excavatum, bilateral positive wrist sign, left thumb sign, hindfoot deformity, skin striae evident on his back and posterior auricular aspect of his neck bilaterally.

Initial management included 15L 100% FiO₂ per oxy mask. Genetics was consulted with concern for underlying connective tissue disorder. Cardiology was consulted with concern for new onset murmur and an ECHO was performed. Serial imaging of his pneumothorax did not show improvement but as he was clinically stable and asymptomatic, the patient was discharged with follow-up with pediatric surgery scheduled for 2 weeks later. Follow-up with ophthalmology revealed no evidence of myopia or subluxation of the intraocular lens in either eye.

Keywords

Hamman sign, connective tissue disorder, spontaneous pneumothorax

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New Onset Murmur or Hamman's Sign: Mid-Systolic Click in a Suspected Case of Connective Tissue Disorder

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Abstract

We present the case of a 15-year-old previously healthy male who presented to Children's Hospital and Medical Center from an outside hospital with a three-day history of shortness of breath, audible clicking noise, and chest discomfort after playing at the batting cages four days prior. Physical exam was significant for a 4/6 audible mid-systolic click heard best at the left fifth intercostal when lying supine or in the left lateral decubitus position. The murmur was not audible in the sitting position. The patient tested positive for seven of the Ghent revised criteria used for diagnosing connective tissue disorders. In consideration of a patient with a potential connective tissue disorder, it is imperative to take a good history and perform a thorough physical exam.

Case

We present the case of a 15-year-old previously healthy male who presented to our institution from an outside hospital with a three-day history of increasing shortness of breath and discomfort after playing at the batting cages. He was transferred to our institution for further evaluation after identification of a pneumothorax on chest computer tomography. Consent was obtained from the patient's parents.

The patient reported that four days prior to admission, he went to the batting cages with his dad. The next morning, he noticed an audible clicking noise in his chest. He also noted shortness of breath and chest discomfort. There was no history of trauma or injury. On the day of admission, his parent noticed a loud whooshing noise associated with his heartbeat, which prompted them to seek medical attention.

Per discussion with the patient and his family, the patient heard an audible clicking noise when lying supine or in the left lateral decubitus position. The family history was significant for tall stature and connective tissue abnormalities on both maternal and paternal sides. The patient is one of eight children. In addition, two siblings were also noted to have developmental delays.

The physical exam was significant for the following: The patient had a 3/6 audible mid-systolic click heard best at the left fifth

intercostal when lying supine or in the left lateral decubitus position. The murmur was not audible in the sitting position. On further evaluation, the patient tested positive for seven of the Ghent revised criteria used to diagnose connective tissue disorders, which include: wingspan to height ratio was 78" to height 76" (ratio 1.02), pubic symphysis to floor 45", pectus excavatum, bilateral positive wrist sign, left thumb sign, hind-foot deformity, skin striae evident on his back and posterior auricular aspect of his neck bilaterally.¹

Our initial management included administering 15L 100% FiO₂ per oxygen mask. Incentive spirometry was encouraged. Genetics was consulted with concern for underlying connective tissue disorder. Cardiology was consulted with concern for new onset murmur and an echocardiogram was performed. Serial imaging of his pneumothorax did not show improvement, but as he was clinically stable and asymptomatic, the patient was discharged with follow-up to pediatric surgery two weeks after discharge. Evaluation by ophthalmology revealed that there was no evidence of myopia or subluxation of the intraocular lens in either eye.

The patient returned to our institution less than two weeks after discharge with increasing shortness of breath. Repeat chest x-ray at that time revealed that the pneumothorax had not resolved for which a chest tube was placed. He was transitioned to water seal 24 hours after placement of the chest tube.

Imaging: Initial chest x-ray showed minimal thoracic curvature and a left apical pneumothorax measuring 5.0 cm in thickness within the left apex. Computer tomography of the chest with contrast showed the following findings: left hydropneumothorax with moderate pneumothorax and a small pleural fluid component, as well as 3-4 mm tiny blebs in the apical portion of the left lung. An echocardiogram demonstrated a normal sized aortic root without any evidence of mitral valve regurgitation or prolapse.

Discussion

Marfan syndrome is an autosomal-dominant condition, associated with fibrillin mutation that has a prevalence of one in 5000.² There can be variable presentations of Marfan syndrome. The most common gene mutation associated is Fibrillin 1 (FBN1). Ghent criteria is used to assist with the clinical diagnosis of patients with Marfan syndrome.³ A score of greater than seven suggests systemic involvement.

In this case, the patient very likely has a connective tissue disorder. Due to his lack of aortic and eye involvement, he does not meet criteria for Marfan syndrome. However, there are other connective tissue disorders that have overlapping phenotypes. Differential diagnosis includes but is not limited to: non-specific connective tissue disorder, potential Marfan syndrome, Mitral valve prolapse, Aortic root diameter at upper limits of normal for body size, Stretch marks of the skin, and Skeletal conditions similar to Marfan syndrome (MASS) phenotype, Loeys-Dietz syndrome, congenital contractural arachnodactyly, and mitral valve prolapse syndrome. With respect to our patient, the family is still awaiting insurance approval for further genetic evaluation.

Spontaneous pneumothorax (Diagnosis and Management): A pneumothorax is a collection of air between the visceral and the parietal pleura. Males between the ages of 16-24 have a higher incidence of spontaneous primary pneumothorax. Patients will present with dyspnea and may have pleuritic chest pain. On exam, there may be hyper-resonant percussion on the affected side, decreased lung sounds, and the patient can appear cyanotic. Diagnosis is confirmed with chest x-ray.

Supplemental oxygen is given to help with reabsorption of air in the extra-pleural space. If the air leak fails to resolve, a chest tube can be placed to drain the air collection. For our patient, conservative management with supplemental oxygen did not completely resolve his pneumothorax, and a chest tube was placed for resolution.

Discussion Regarding Hamman Sign: Hamman sign is classically understood as a loud synchronous precordial sound.³ This case is unique as the patient had a Hamman sign

that sounded like the murmur traditionally heard with mitral valve prolapse. There have been other documented cases in the literature of unique Hamman signs; however, our case also had the additional physical exam findings consistent with connective tissue disorder.⁴

Mitral valve prolapse, the presence and displacement of thickened mitral leaflets into the left atrium during systole, has been associated with mutations in FBN1.⁵ Mitral valve prolapse has been classically associated with connective tissue disorders; however, this has been challenging to identify statistically.⁵ With concern for the significant implications associated with mitral valve prolapse in a pediatric patient with new onset murmur, echocardiography is a non-invasive modality that can provide additional information regarding a patient's respective cardiac anatomy and function.

Conclusion

Our patient presented with a spontaneous pneumothorax and a unique Hamman sign that was concerning for underlying cardiac pathology. With concern for connective tissue disorders, it is imperative to take a good history and to perform a thorough physical exam to exclude underlying potential cardiac abnormalities. ■

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