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Case Report

Aneurysms and Aortic Insufficiency: Two Very Rare Complications of Wilson's Disease

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ABSTRACT

Wilson's disease is a disorder of copper metabolism that results in the accumulation of copper in various body tissues. The most common organs involved in this disease are the liver and the brain, with most of the clinical symptoms being related to these 2 organs. Albeit less affected, the cardiovascular system is also involved. In this paper, we aim to report a very rare cardiac complication of Wilson's disease. [1] ***(Iranian Heart Journal 2021; 22(3): 123-127)***

KEYWORDS: Aneurysm, Aortic insufficiency, Wilson's disease

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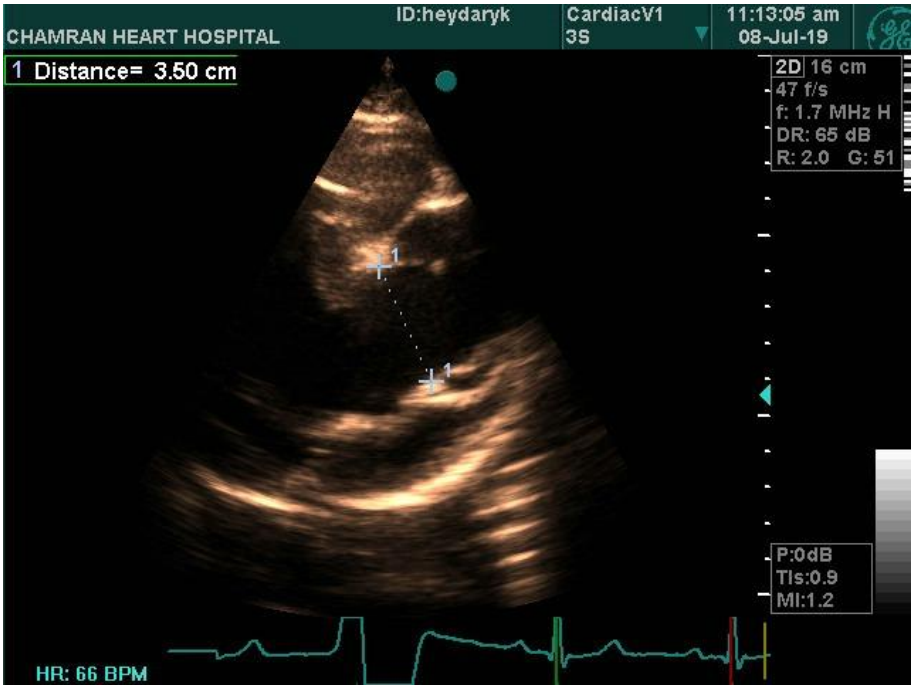
Wilson's disease is a disorder of copper metabolism that results in the accumulation of copper in various tissues of the body. The most common organs involved in this disease are the liver and the brain, and most of the clinical symptoms are related to these 2 organs. [1] The first case of Wilson's disease was introduced by Wilson in 1912. [2] Based on a report by the World Health Organization, the true incidence of Wilson's disease is 1/10000 to 1/30000. [3] The clinical manifestations of Wilson's disease include neurological symptoms such as impaired motor movements, involuntary movements such as tremors, cone dystrophy, and liver dysfunction manifesting itself as acute or chronic hepatitis, cirrhosis, or hepatic encephalopathy. [4, 5] The heart is another organ that can be affected by this disease. [6] In this paper, we present a very rare cardiac complication of Wilson's disease.

Case Report

Our patient was a 41-year-old man, known to have suffered from Wilson's disease since he was 12 years old. He had a brother and a sister, both of whom were affected by this disease. The patient's father was his mother's cousin. The patient's family history was positive, with the clinical manifestations of his Wilson's disease being convulsions. He also had neuromuscular problems, mood disorders, and mental retardation (Fig. 1). We performed a computed tomography scan, which revealed

brain atrophy. The patient was treated with 500 mg of D-penicillamine daily and 100 mg of phenobarbital twice a day. Routine tests were normal, and his vital signs were stable. On physical examination, he had a 5/6 diastolic murmur over the right second intercostal space. Additionally, neuromuscular problems and dysarthria were observed. Echocardiography revealed that the size of the aortic annulus was 3.50 cm, the size of the sinus of Valsalva was 5.06 cm, the size of the sinotubular junction was 4.56 cm, and the size of the ascending aorta was 3.48 cm. In addition, there were thickened aortic valves, severe aortic regurgitation, up-to-moderate mitral regurgitation, mild pulmonary insufficiency, and mild tricuspid regurgitation, but no aortic stenosis, mitral stenosis, tricuspid stenosis, and pulmonary stenosis was detected (Fig. 2).

Figure 1. The patient has neuromuscular problems, mood disorders, and mental retardation.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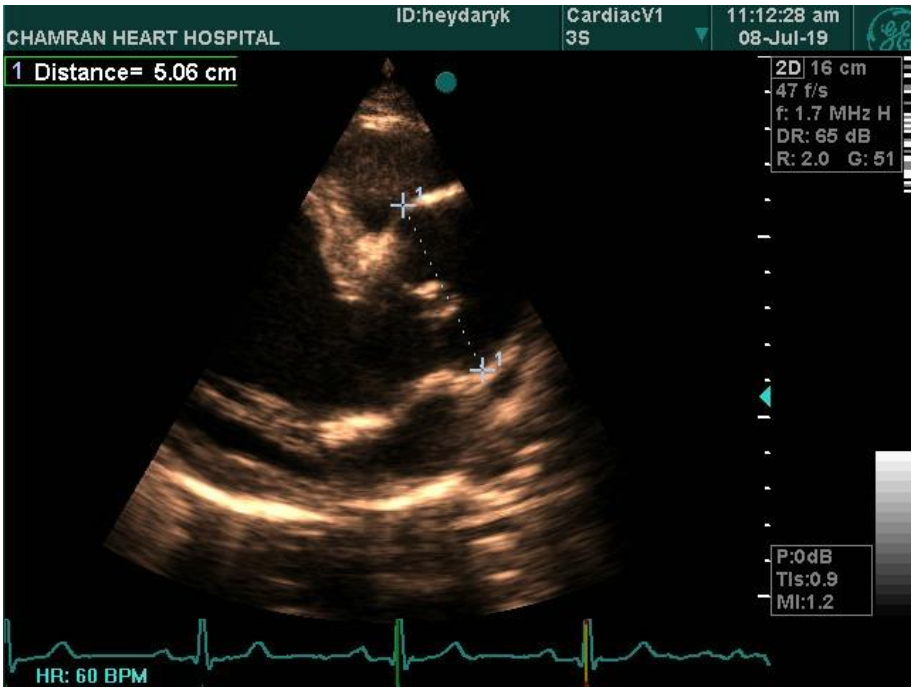
A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B

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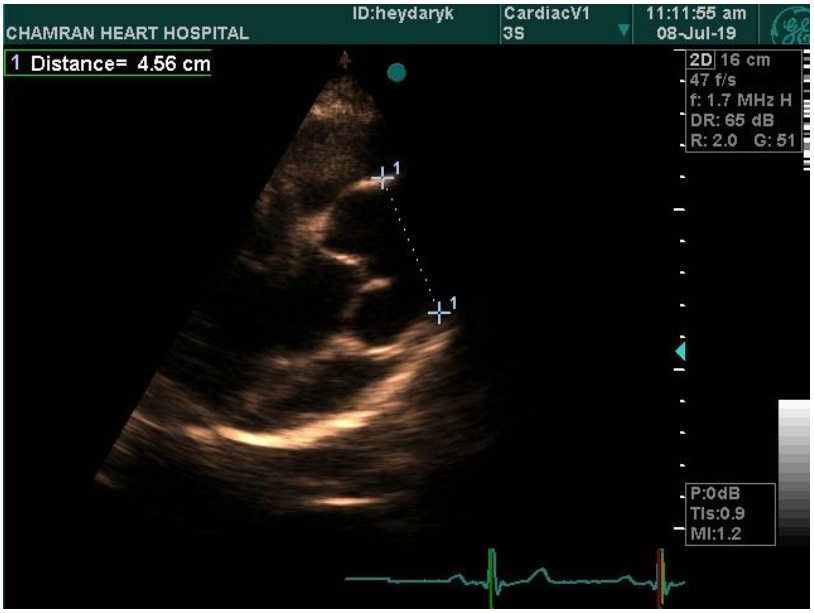

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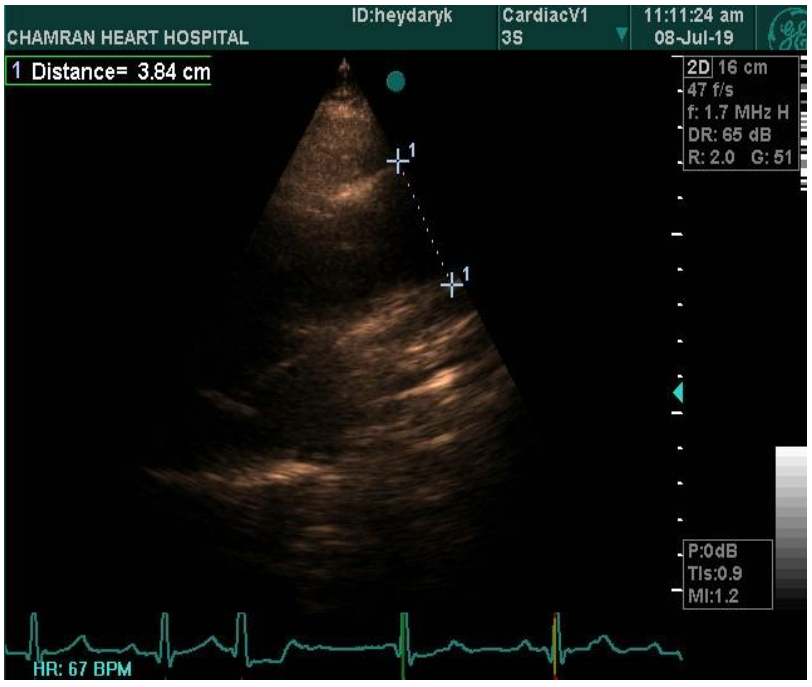

D

Figure 2. Echocardiography findings show that A) the size of the aortic annulus is 3.50 cm, B) the size of the sinus of Valsalva is 5.06 cm, C) the size of the sinotubular junction is 4.56 cm, and D) the size of the ascending aorta is 3.48 cm.

DISCUSSION

Wilson's disease is an autosomal recessive disorder characterized by neurological and hepatic involvement. [4, 5] There have been many reports around the world since Wilson introduced the disease. [5] Although the cardiovascular system is less affected,

cardiovascular involvement has been reported. For instance, Hlubocká et al [6] reported that patients with Wilson's disease had increased thickness of the interventricular septum and the left ventricular posterior wall. Kuan et al [7] identified the cardiac manifestations of Wilson's disease as cardiac arrhythmias, cardiomyopathy, cardiac death, and autonomic dysfunction. In a case report, Bajaj et al [8] identified a case of second-degree heart block incidentally in a young woman with Wilson's disease. In addition, recently, Grandis et al [9] showed that patients suffering from Wilson's disease were at a higher risk of heart failure and cardiac fibrillation. In an imaging trial, Quick et al [10] assessed the cardiac involvement of Wilson's disease by magnetic resonance imaging and demonstrated that cardiac fibrosis might affect the prognosis in Wilson's disease. We herein reported aneurysms and aortic insufficiency as 2 very rare cardiac complications of Wilson's disease. To our knowledge, this is the first report of aneurysms and aortic insufficiency as a cardiac manifestation of Wilson's disease. The mechanism of these 2 cardiac manifestations is unknown, but the proposed hypothesis is that the excessive deposition of copper in the aorta tissue may justify these complications. The salient point in this case report is that aneurysms and aortic insufficiency are associated with Wilson's disease; nonetheless, whether this association is causal is unclear. Thus, further causal investigations are needed to elucidate this issue.

REFERENCES

1. Barbosa ER, Machado AA, Cançado EL, Deguti MM, Scaff M. Wilson's disease: a case report and a historical review. Arquivos de neuro-psiquiatria. 2009 Jun; 67(2B):539-43
2. Bandmann O, Weiss KH, Kaler SG. Wilson's disease and other neurological copper disorders. The Lancet Neurology. 2015 Jan 1; 14(1):103-13.
3. Coffey AJ, Durkie M, Hague S, McLay K, Emmerson J, Lo C, Klaffke S, Joyce CJ, Dhawan A, Hadzic N, Mieli-Vergani G. A genetic study of Wilson's disease in the United Kingdom. Brain. 2013 May 1; 136(5):1476-87.
4. Zhang YC. Research of hepatolenticular degeneration in the past 10 years. Zhonghua Shen Jing Jing Shen Ke Za Zhi. 1959; 5:326-7
5. Liu J, Luan J, Zhou X, Cui Y, Han J. Epidemiology, diagnosis, and treatment of Wilson's disease. Intractable & rare diseases research. 2017 Nov 30; 6(4):249-55.
6. Hlubocka Z, Mareček Z, Linhart A, Kejřová E, Pospíšilová L, Martasek P, Aschermann M. Cardiac involvement in Wilson disease. Journal of inherited metabolic disease. 2002 Jun; 25(4):269-77.
7. Kuan P. Cardiac Wilson's disease. Chest. 1987 Apr 1; 91(4):579-83.
8. Bajaj BK, Wadhwa A, Singh R, Gupta S. Cardiac arrhythmia in Wilson's disease: An oversighted and overlooked entity!. Journal of neurosciences in rural practice. 2016 Apr; 7(04):587-9.
9. Grandis DJ, Nah G, Whitman IR, Vittinghoff E, Dewland TA, Olgin JE, Marcus GM. Wilson's disease and cardiac myopathy. The American journal of cardiology. 2017 Dec 1; 120(11):2056-60.
10. Quick S, Reuner U, Weidauer M, Hempel C, Waessnig N, Mues C, Sveric K, Heidrich F, Reichmann H, Strasser R, Speiser U. 4096Cardiac manifestation in wilson disease. The CARMA Wilson study. A comprehensive clinical and cardiac imaging trial. European Heart Journal. 2017 Aug 1; 38(suppl_1)