

PORTABLE DEVICE AS A DIAGNOSTIC TOOL FOR MARFAN'S SYNDROME IN A REMOTE AREA: A CASE REPORT

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Abstract: The Marfan's syndrome is an autosomal dominant genetic disease that affects connective tissue, with an incidence of approximately one case for every 5000 individuals. Cardiovascular involvement often occurs in adulthood. As the most common manifestation of the syndrome, ascending aortic aneurysm frequently leads individuals to conditions of clinical emergencies, the emergency room and intensive care unit are a common sites for diagnosing cardiac disorders in patients with Marfan. In this article we find the case report of a 41-year-old patient with a history of chest pain, signs and symptoms of heart failure, admitted to the ICU, with phenotypic manifestations of the disease, whose aortic aneurysm was diagnosed during the initial examination of POCUS. As it is a relatively rare diagnosis, in a rural patient who

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until now was unaware of the diagnosis itself, the result of which was identified in a routine examination in the ICU, performed with a portable device, the ability to change the outcome of a disease with high morbidity and mortality with a simple exam, easy to access and with low training requirements in a highly complex unit, even in distant care units in a population with low socioeconomic status. This article aims to show how a simple examination, POCUS, performed with a handheld device, is able to identify complex, high-impact diseases.

Keywords: Marfan's Syndrome; POCUS; Aortic Aneurysm; Handheld Ultrasound;

A 41-year-old, male, rural worker, admitted to the ICU of the Hospital Medicina Humana, Candeias-Ba, Brazil, 08 of

June 2020, with a history of non-ventilatory chest pain, associated with progressive dyspnoea, worsening in the last 6 months, now with minimal effort, admitted using a mask with reservoir, tachypneic, with 98% peripheral saturation. Tall patient, with micrognathism, arachnodactyly, flat feet and pectus carinatum (Supplementary material 1). During physical examination, with symmetrical edema in the lower limbs was observed, associated with bilateral basal crackles and murmur in aortic focus. Also observed ogival palate. It was not possible to observe a ocular lens lesions. Underwent POCUS, a cardiac image on paraesternal long-axis, showed signs of systolic alteration of the left ventricle (LV) was observed, an important increase in the aortic sinus, measuring 7.15 cm in diameter, with a slight valve flap, compatible with



an ascending aortic aneurysm with probable dissection. The examination was performed with a portable, hand-held ultrasound device, Medscan model WSB 123, Konted, WSM Electronic

Products (Beijing) Co., with software applied to the Samsung Note9 smartphone.

FIGURE 1: increase in the aortic sinus, leading to one deforming along left cameras, demonstrating a aortic aneurism

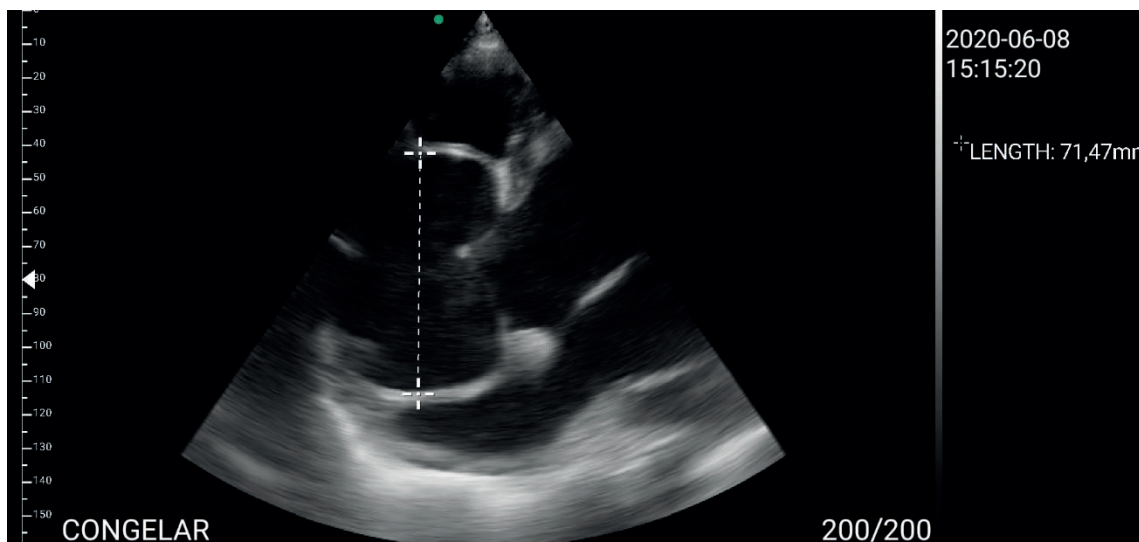
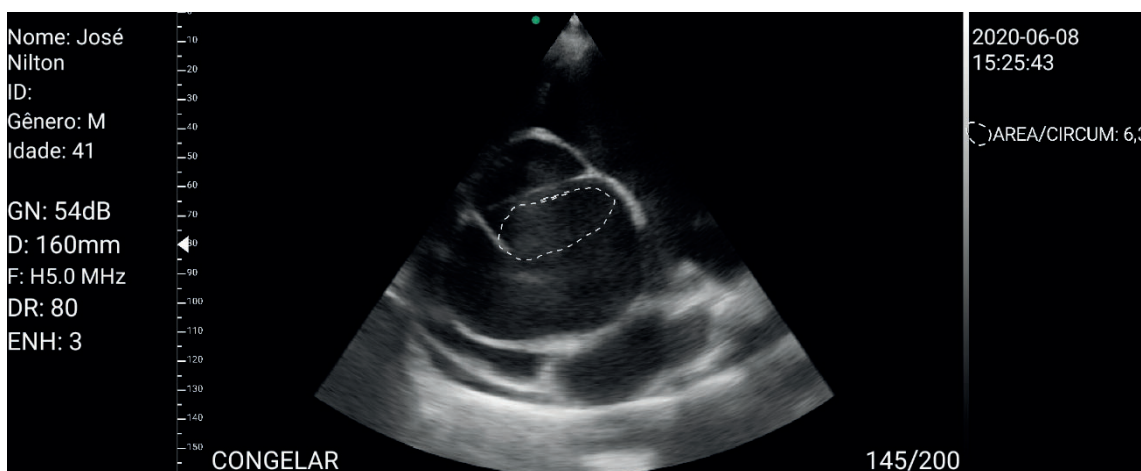


FIGURE 2: cross section of the aortic root at the level of the valves: We notice the "flap" during the opening in the systolic phase



DISCUSSION

Marfan syndrome is a relatively rare genetic disease, but it is one of the major diagnostic suspicions in young patients with aneurysmal involvement of its root. With autosomal dominant transmission. It presents an incidence of one case per 5000 individuals (Ho NCY, Tran JR, Bektas A, 2005). The clinical diagnosis was established in 1996 by the Ghent criteria, which use criteria of genetic, family, clinical and imaging findings. The criteria were updated in 2010 (Loeys BL, Dietz HC, Braverman AC, et al, 2010) (For more information see table in supplementary materials).

The diagnosis of MS in childhood is often difficult, given that certain clinical manifestations only become evident with age. The ascending aorta is the

portion of the arterial tree most affected by aneurysmatic development (Gomes SC, Almeida AG, 2020). The patient in this case have six children of which two have similar physical aspect – informed during interview.

Transthoracic ultrasound (ETT) in the context of POCUS plays an important role because it is noninvasive exam and can be performed in the context of emergencies with very close to time the onset of symptoms and assisting in the differential diagnosis of chest pain (Nishigami K, 2018). The diagnosis of ascending aortic aneurysm can be made with ultrasound examination with transthoracic approach with high sensitivity. The increase in the diameter of its root at the sinus level, above 5.5 cm, the one with the greatest predictive value (Jondeau G, Michel JB, Boileau C, 2011). The patient in



question presented a diameter of 7.0 cm on examination, confirmed later with echocardiography performed by the specialist (see others images in supplementary materials). Clinical criteria and examination findings confirmed the disease, previously unknown to the patient.

CONCLUSION

The routine examination in the ICU, elaborated in a simple and systematic way, was able to diagnose a disease of high morbidity and mortality, corroborating the importance of the point of care ultrasound within the differential diagnoses in the ICU, and, in the specific context, changing the course of disease within a family with few resources and access to the health system.

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