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Clinical History

A 39 year-old female with Turner Syndrome completely asymptomatic in the previous years, presented to our department with recent onset of dizziness and fatigue. A chest X-ray (Figure 1) demonstrated an hypoplastic right lung with dextroposition of the heart and enlarged cardiac contour. In the right hemithorax, a tubular opacity descending toward the diaphragm adjacent to the right cardiac contour was noted.

Figure 1. Chest X-ray demonstrated an hypoplastic right lung with dextroposition of the heart and enlarged cardiac contour. In the right hemithorax, a tubular opacity (arrows) adjacent to the right cardiac contour was noted.

MRI was requested for further assessment of anatomy. MR angiography images (Figures 2 and 3) showed an anomalous vertically oriented venous structure which drained the right upper, middle and lower lobe pulmonary veins into the inferior vena cava.
Figure 2. MR-angiography showed an anomalous oriented venous structure (arrows) which drained the right upper, middle and lower lobe pulmonary veins into the inferior vena cava.
Figure 3. MR-angiography showed the anomalous venous structure associated with aortic coarctation.

The vertical vein connects to the inferior vena cava at approximately 2.5 cm below the cavo-atrial junction. The left upper and lower lobe pulmonary veins drained normally into the left atrium. Right atrium was enlarged. Mild narrowing at the level of the aortic isthmus with post-stenotic dilatation consistent with aortic coarctation was also noted (Figure 4).
Figure 4. MR Angiography revealed an aortic coarctation (arrow) at the level of the isthmus with post-stenotic dilatation.

Diagnosis
Scimitar Syndrome

Discussion
Scimitar syndrome is a rare congenital anomaly characterized by an anomalous connection of the pulmonary veins with the inferior vena cava. It represents 3-5 percent of all partial anomalous pulmonary venous return (PAPVR) and it is commonly associated with hypoplasia of the right lung, pulmonary sequestration, persisting left superior vena cava and dextroposition of the heart.\textsuperscript{1} PAPVR is seen in approximately 13 percent of patients with Turner syndrome. Another cardiovascular anomaly that commonly affects patients with Turner Syndrome is aortic coarctation, being present in about 15 percent of cases.\textsuperscript{2}

The age of presentation of Scimitar Syndrome is variable, with a mean age of diagnosis of seven
months. Generally the infantile onset of symptoms like tachipnea, recurrent pneumonia and heart failure are associated with a worse prognosis. When patients are asymptomatic, diagnosis is usually made incidentally during adulthood. Clinically, it is an acyanotic left to right shunt: the anomalous pulmonary vein drains blood from the right lung into the IVC resulting in right heart volume overload and risk of heart failure. Imaging methods are needed to confirm the diagnosis, evaluate the severity of the right-to-left shunt and evaluate the impact on cardiac function.

The diagnosis of scimitar syndrome is usually initially made on chest X-ray. The most common non-invasive methods used to confirm the diagnosis are transthoracic or transesophageal echocardiography, CT or MR angiography. Scimitar syndrome typically presents with the “scimitar sign” on the postero-anterior chest radiograph, which consists of an aberrant venous return that resembles a curved sword named scimitar. The syndrome also includes dextroposition of the heart due to right lung hypoplasia. To confirm the diagnosis and to assess the severity of the shunt, further assessment with non-invasive techniques such as CT or MR angiography is recommended.

By using MR angiography, cine-MR and velocity-encoded cine (VEC-MR) sequences, MRI represents the main non-invasive imaging technique to provide a complete anatomical assessment and shunt quantification. VEC-MRI allows for precise quantification of pulmonary-to-systemic flow ratio (Qp/Qs), which determines the hemodynamic significance of the shunt. ECG-gated CT angiography may be an alternative imaging technique to identify and characterize the connection of the anomalous pulmonary vein.

In addition, associated abnormalities such as aortic coartaction, lung sequestration, persisting left superior vena cava, cardiac valvular abnormalities, lung hypoplasia and other cardiac or pulmonary abnormalities may be complementary assessed by CT and MRI. Conservative management is generally appropriate, although surgical treatment may be considered in symptomatic patients, with other associated cardiac abnormalities or with a Qp/Qs higher than 1.7. In our patient Qp/Qs was calculated as 1.2 and aortic coarctation was mild, therefore medical management was undertaken.

The diagnosis of scimitar syndrome can be made on chest x-ray. In adults may be often asymptomatic, but follow-up is needed to monitor consequences such as pulmonary hypertension and right ventricular heart failure.

Cardiac magnetic resonance is the technique of choice to confirm the diagnosis especially in young patients thanks to the absence of ionizing radiation. It is a comprehensive method that allows for anatomic depiction, shunt quantification and cardiac function assessment.

References:

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