

Cardio-Vascular and Pulmonary Complications in Marfan Disease at the Angioscan: A Report of 3 Cases

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Abstract : We report three cases of Marfan disease, all of the same siblings, aged from 22 years, 29 years and 39 years, referred to our department for a thoracic and cerebral angioscan. Examinations were conducted with a 64-bar Siemens CT scanner with a dual-body automatic injector from Medrad. Various thoracic complications found featured: aneurysm of the ascending thoracic aorta and apical subpleural blebs. Stanford Type A aortic dissection was noted in two cases. A mild bilateral pneumothorax was found in one patient. To the brain assessment, only one patient presented aneurysm of left carotid siphon. Angioscan remains the reference exam for the assessment of cardio-vascular monitoring during Marfan disease. It allows identify serious complications such as thoracic aorta aneurysms that engages the vital prognosis.

Keywords: Marfan disease, aneurysm (aneurism), aortic dissection

1. Introduction

Marfan disease is an uncommon genetical disorder, autosomal dominant, caused by the mutation of the gene coding for Fibrillin type 1 (FBN1)[1]. Most common complications include cardiovascular, skeletal, ophthalmological and pulmonary [2]. Multi-detector CT scans development have highly contributed in the monitoring aspects and the management of these complications. Throughout three observations, we are going to study cardiovascular and pulmonary complications of Marfan disease.

2. Observation 1

A 29-year-old male patient working as a gardener without particular past medical history. A sudden death of the mother's patient (unreported cause) was identified as a family history. Thoracic and cerebral angioscan were performed in all patients with the same protocole.

Thoracic angioscan allowed to find for the 1st patient : an aortic dissection (the presence of an intimal tear : blue arrow) interesting the initial part of the ascending aorta range from the aortic arch to the descending thoracic aorta without pericardial concern. It was classified as Stanford Type A (figure 1 : A, C).

A loss of parallelism of the arterial wall related to a fusiform aneurysm of the initial part of the aorta with a maximal transverse diameter of 55mm was found. This aneurysm measures 65mm in height (figure 2).

The examination of the lungs revealed some apical subpleural blebs at the right side.

Cerebral and abdominal aorta angioscan and the echodoppler of lower limbs arteries were normal.

The therapeutic management specially consisted at Surgery by Bentall method.

A medical treatment based on betablockers, diuretic and anticoagulant was administrated.

3. Observation 2

The second patient is 22 years old in « terminal » level without past medical history. The consultation was motivated by the discover of Marfan syndrome in his elder brother (patient 1). He didn't reveal any symptom.

Thoracic angioscan revealed :

A fusiform aneurism of the sinusal portion of the thoracic aorta with an average diameter estimated at 40mm, without dissection

The presence of some emphysema blebs at the right subpleural apex in the pulmonary parenchyma.

A deformity of the rib cage with a pectus excavatum-like aspect (figure 3) was found.

Cerebral angioscan, the exploration of the abdominal aorta and the echodoppler of the lower limbs were normal.

He was under betablockers (avlocardyl).

4. Observation 3

Patient 3 was 39 years old, working as a fisherman without past medical history.

Thoracic angioscan showed : an aortic dissection classified Stanford Type A. It extended to the aortic arch and stopped at the descending thoracic aorta.

A fusiform aneurism of the initial aorta with a diameter of 55mm.

Bilateral mild pneumothorax at the costo-diaphragmatic gutter and pulmonary apex level unsticking the mediastinal pleura (figure 4 : A and B).

Cerebral angioscan found an aneurism of the left carotid siphon (figure 5).

5. Discussion

The incidence of thoracic aorta aneurisms is estimated at 4,5 cases for 100.000 [3]. Supra-valvular aorta aneurisms are less frequent than the aortic root aneurisms. They specially affect males (3 men/ 1 woman). Aortic root aneurisms affect younger patients (30-50 years old).

Twenty per cent of the patients affected by Marfan syndrome were operated for aortic root aneurism [4]. The three patients in our observation had an aneurism of the sinus portion of the thoracic aorta.

The study done by Chan Y C and all, found on 525 patients with Marfan disease, 112 cases (21,3%) of aneurism and/or an aorto-iliac dissection. These latters concerned the thoracic aorta in 66,7% of cases [5].

Even if they are rare, aneurisms could seat everywhere. A patient in this study (patient 3) presented an extra-aortic aneurism specially on the left carotid siphon.

The role of the imaging in aneurism is to help realise the anatomical assessment and some the etiological one.

It is reported in the literature a special association of bilateral popliteal aneurism and abdominal aneurism during Marfan disease [7].

By measuring the aortic diameters, it allows the screening of aneurismal enlargement and helps set up the surgical indication when the diameter of the aneurism catches 50mm.

Aortic dissection constitutes the most fearing complication during the evolution of Marfan disease. Its frequency is estimated at 4 to 10 for 100.000 people. Acute flaps occur on fragilized aortas. This is due to hereditary dystrophic conditions, first and foremost Marfan disease and familial annulo-aortic ectasias. We also find congenital affections or acquired diseases, most often atheromatous.

In our study, two patients presented an aortic dissection type A of Stanford.

The dissection type B is the most widespread in Marfan disease.

Patients affected by the disease present an increased risk of aortic dissection at a younger age than others. In our study, the cases of dissections touched two patients less than 40 years old.

Tsai S H, shows 7 cases of aortic dissections developed on patients affected by the disease at an age less than 40 years old in 18 cases [8].

Mimoum L and all,found 37 cases of aortic dissection touching the ascending thoracic aorta for a total of 100 patients [9].

The risk if occurrence of aortic dissection is directly related to the size of the aneurism.

The two patients who presented aortic dissection, had an aneurism that measured 55mm.

Most authors estimate that a prophylactic repair is indicated when the aortic diameter is 45-50mm so that to avoid the occurrence of a tear. This risk is multiplied by 4 beyond 50mm of diameter. However these dissections were reported for diameters less than 50mm.

The indication of surgery should be systematic for aneurisms for which the largest diameter is more than 55 mm measured either by CT scan after injection of a contrast agent or by MRI.

Pulmonary affection features a high frequency of pneumothorax and thoracic deformations that could be severe (scoliosis, pectus excavatum) and sometimes associated with restrictive ventilatory condition.

The main thoracic deformity related to Marfan disease is the pectus excavatum, due to an abnormal growth of the ribs and sternum. Its functional repercussion is specially cardiac, the decrease of pulmonary volumes being not important mostly. In our study, one of the patients presents pectus excavatum (figure 3).

The presence of mild pneumothorax was also demonstrated.

During Marfan syndrome, the presence of pneumothorax is common associated with a pectus excavatum or carinatum. This type of deformation (pectus carinatum) was not found in patients with pneumothorax.

The second patient presented a pectus excavatum without pneumothorax.

A study performed in 100 patients affected by Marfan Syndrome has demonstrated 11% of pneumothorax. Eight patients out of eleven presented thoracic deformities with a high prevalence of pectus carinatum or excavatum [10]. Other factors favoured the occurrence of spontaneous pneumothorax during Marfan disease such as the presence of subpleural apical blebs in the two cases.

The occurrence of a pneumothorax constitutes an uncommon revelation mode of the disease, even in case of familial form. It's important to conjure up the diagnosis in a patient experiencing recurrent spontaneous pneumothorax or a thoracic deformity so that to address them to the specialist.

6. Conclusion

Angioscan remains the reference exam for the assessment of cardio-vascular monitoring during Marfan disease. It allows identify serious complications such as thoracic aorta aneurysms that engages the vital prognosis by the high risk of its dissection and rupture that they could provoke. The occurrence of aneurysm, dissection or recurrent spontaneous pneumothorax specially in a family context should make conjure up the diagnosis and its confirmation for an early and adapted management.

7. Conflicts of interest

No conflict of interest were proclaimed.

8. Contribution of authors

All the authors brought their contribution.

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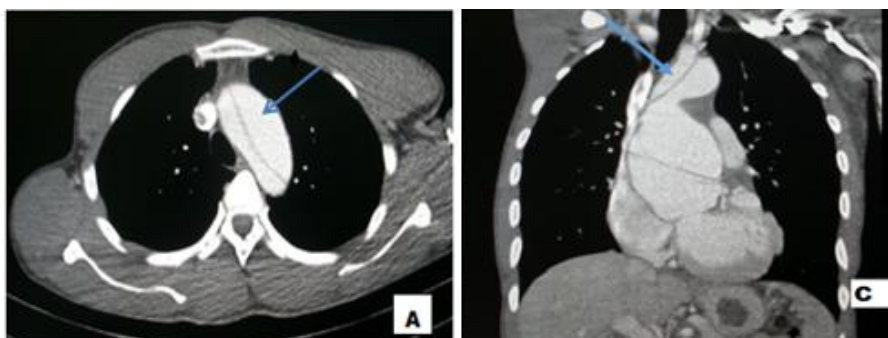


Figure 1 (A-C): Thoracic angioscan in mediastinal window : patient 1.

A : Axial section showing an aortic dissection starting at the aortic sinus and extending to the arch of the descending thoracic.

C : Coronal reconstruction showing the aortic dissection and its width.

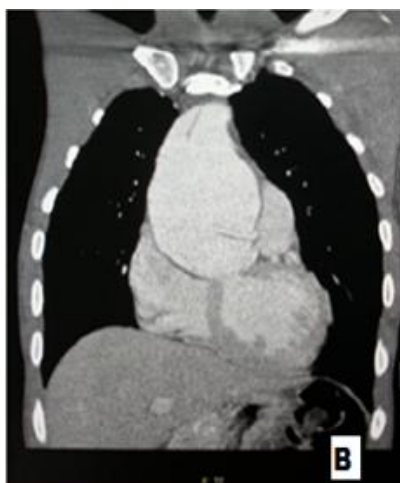


Figure 2 : thoracic angioscan in mediastinal window patient 1.

B: Coronal reconstruction showing a fusiform aneurysm of the thoracic

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Figure 3 : Sagittal reconstruction in bone window showing a pectus excavatum aspect of the thorax.

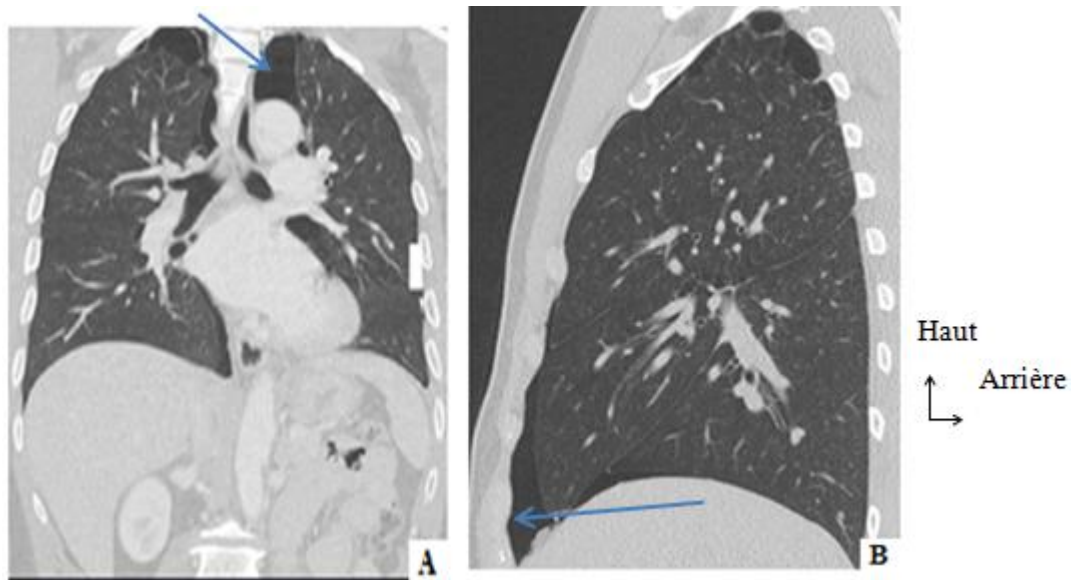


Figure 4 : (A and B)

A : Coronal reconstruction showing moderate bilateral pneumothorax at the apex.

B : Sagittalreconstruction in parenchymal window showing a pneumothorax of the costo-diaphragmatic gutter (blue arrow)

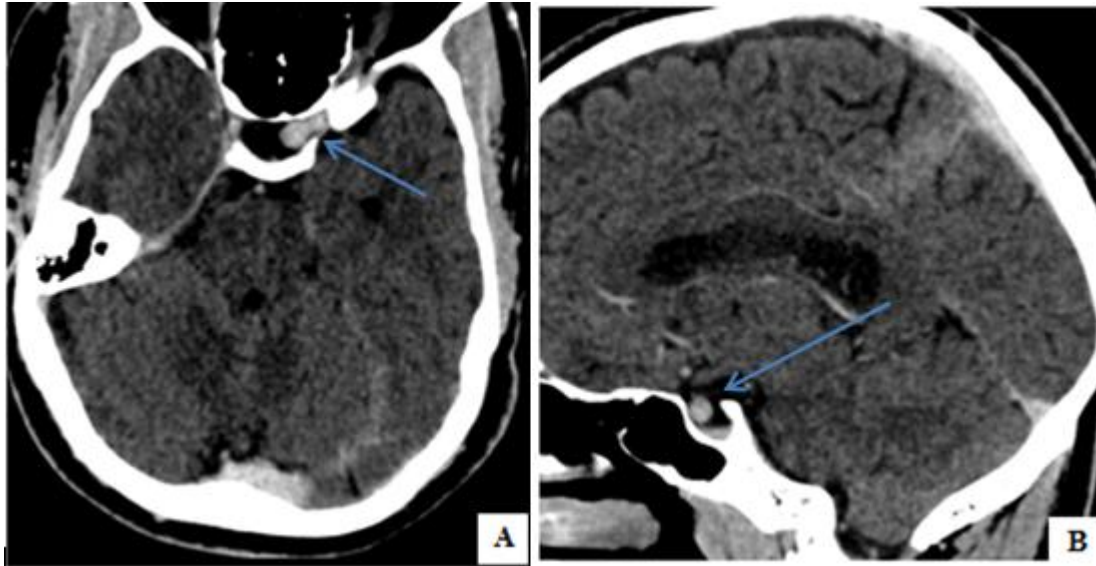


Figure 5 (A and B) : cerebral angioscanner patient 3

A : Cerebral axial section in parenchymal window after injection of contrast agent showing an aneurysmal enlargement of the left carotid siphon (blue arrow).

B : Sagittal reconstruction in parenchymal window after injection of the contrast agent showing an aneurysmal enlargement of the left carotid siphon (blue arrow).