Pediatric Radiology section meeting

February 12, 2019

THE DEVELOPMENTAL PROCESS OF PULMONARY ARTERIOVENOUS MALFORMATION IN HHT

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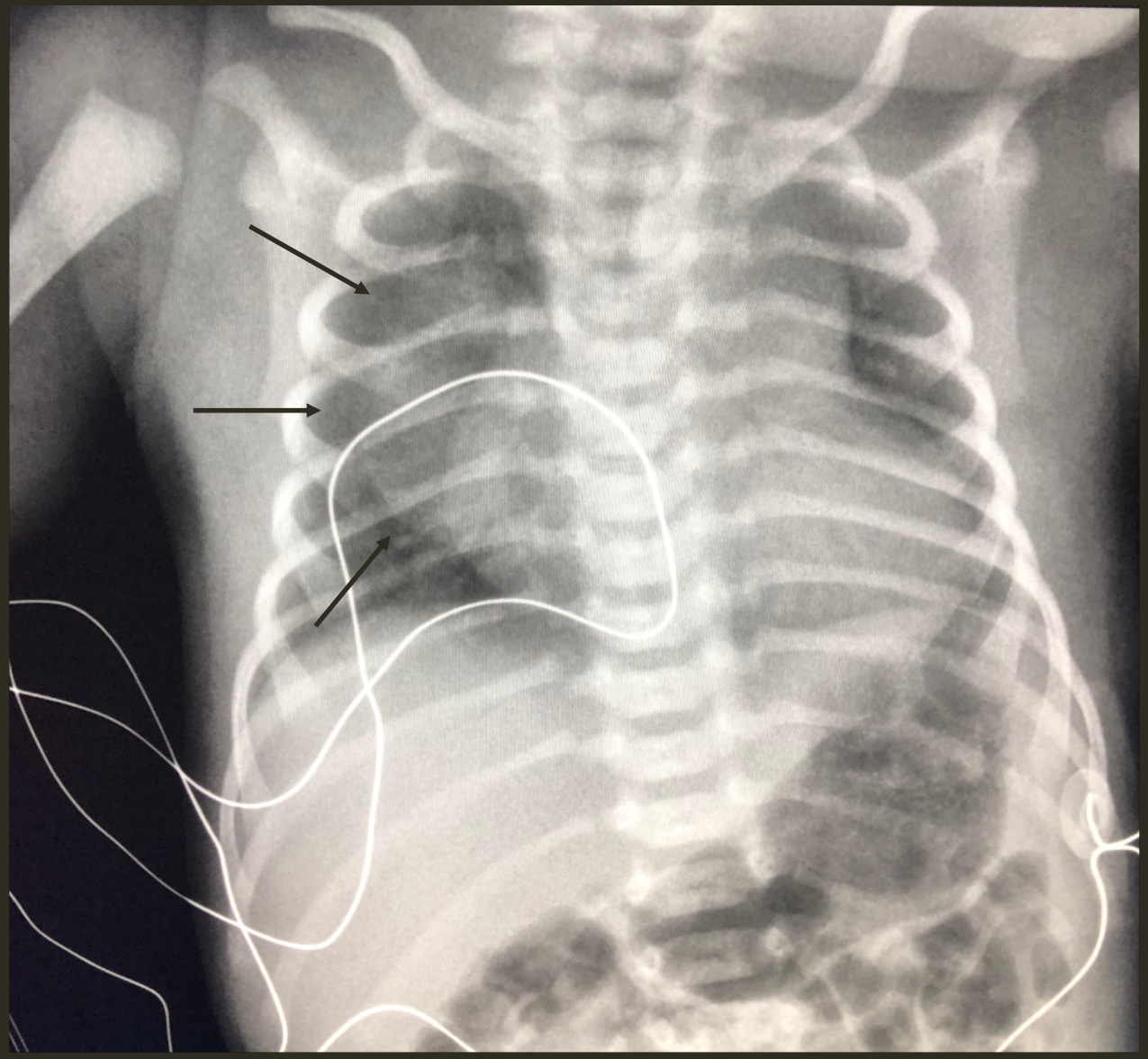
PATIENT 1

Boy,1day

Context of prematurity (36,2 weeks)

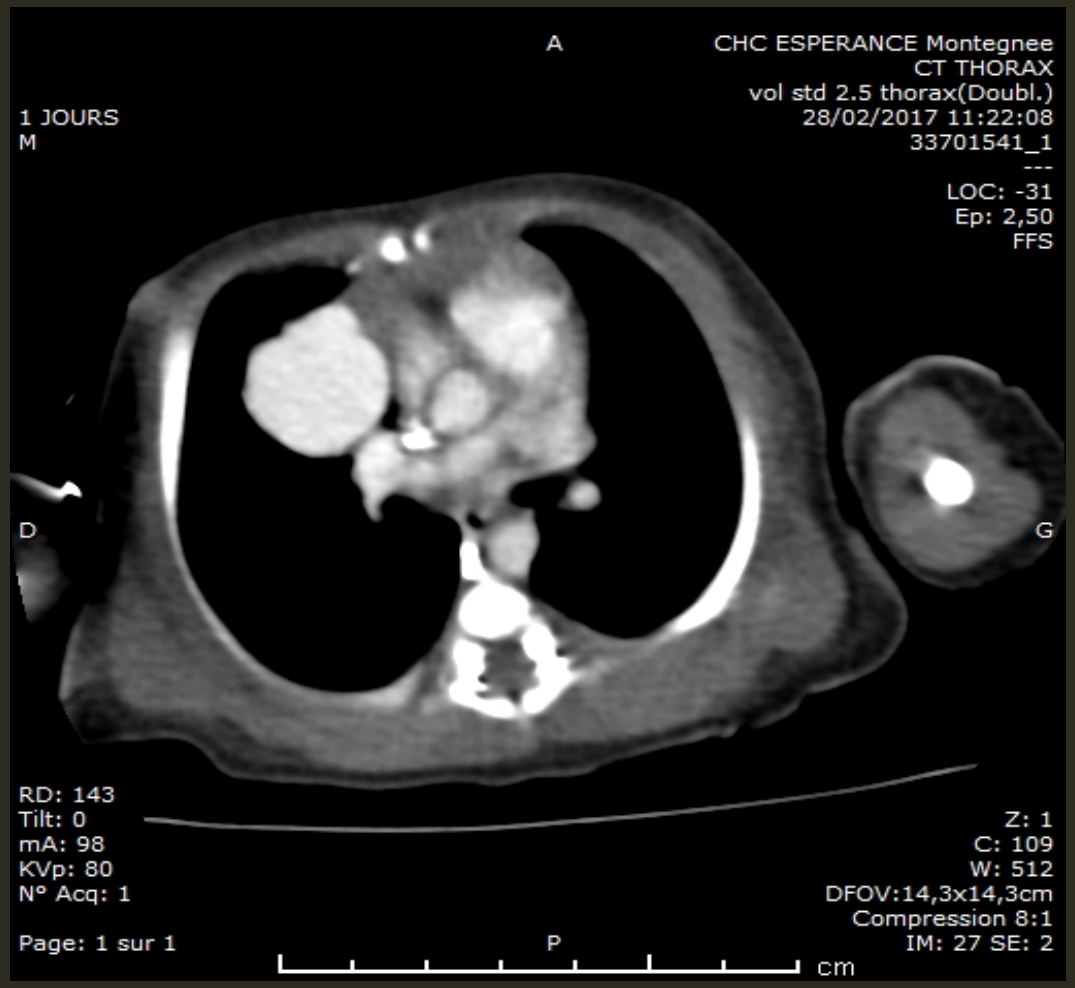
Desaturation without respiratory distress

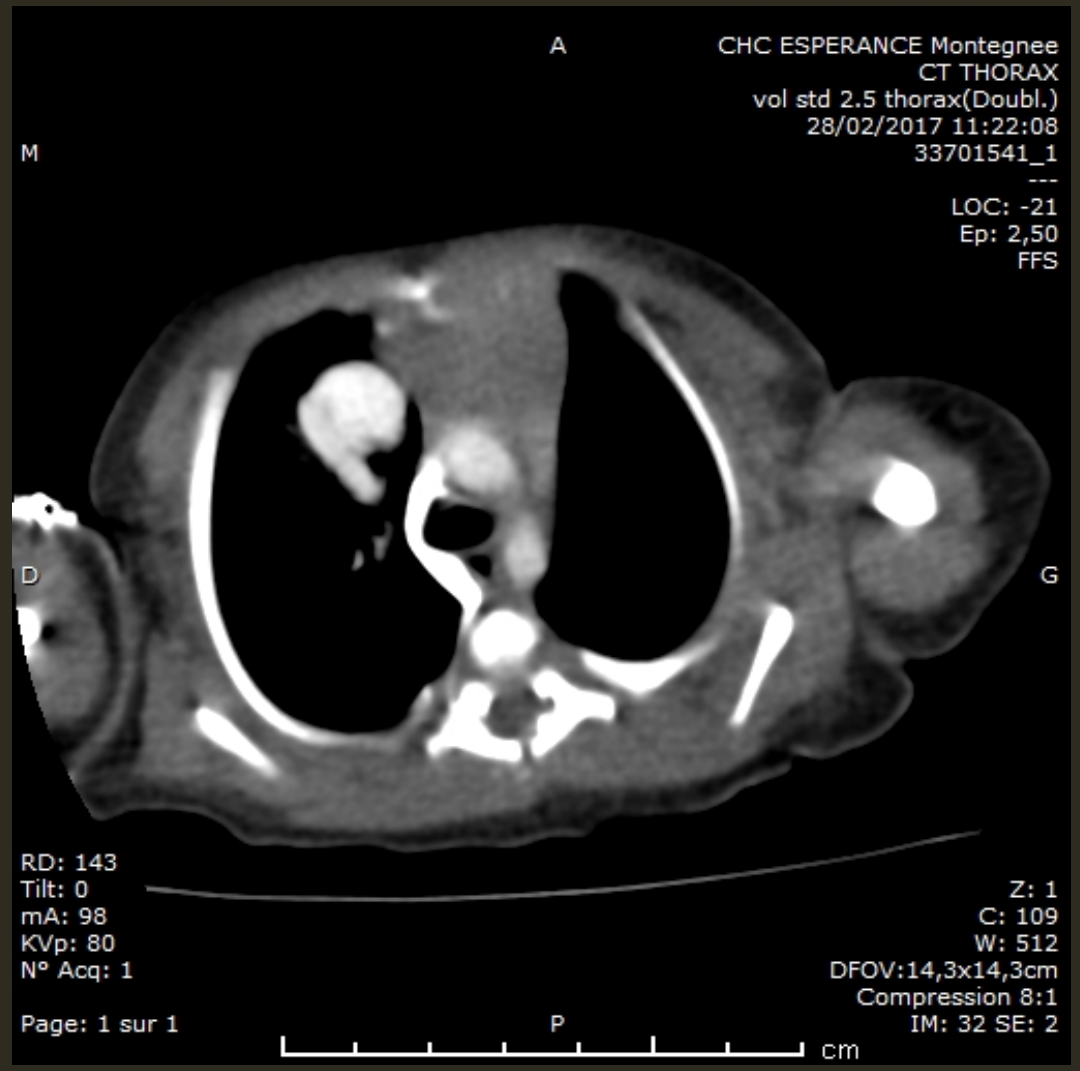
PATIENT 1



Chest X-ray

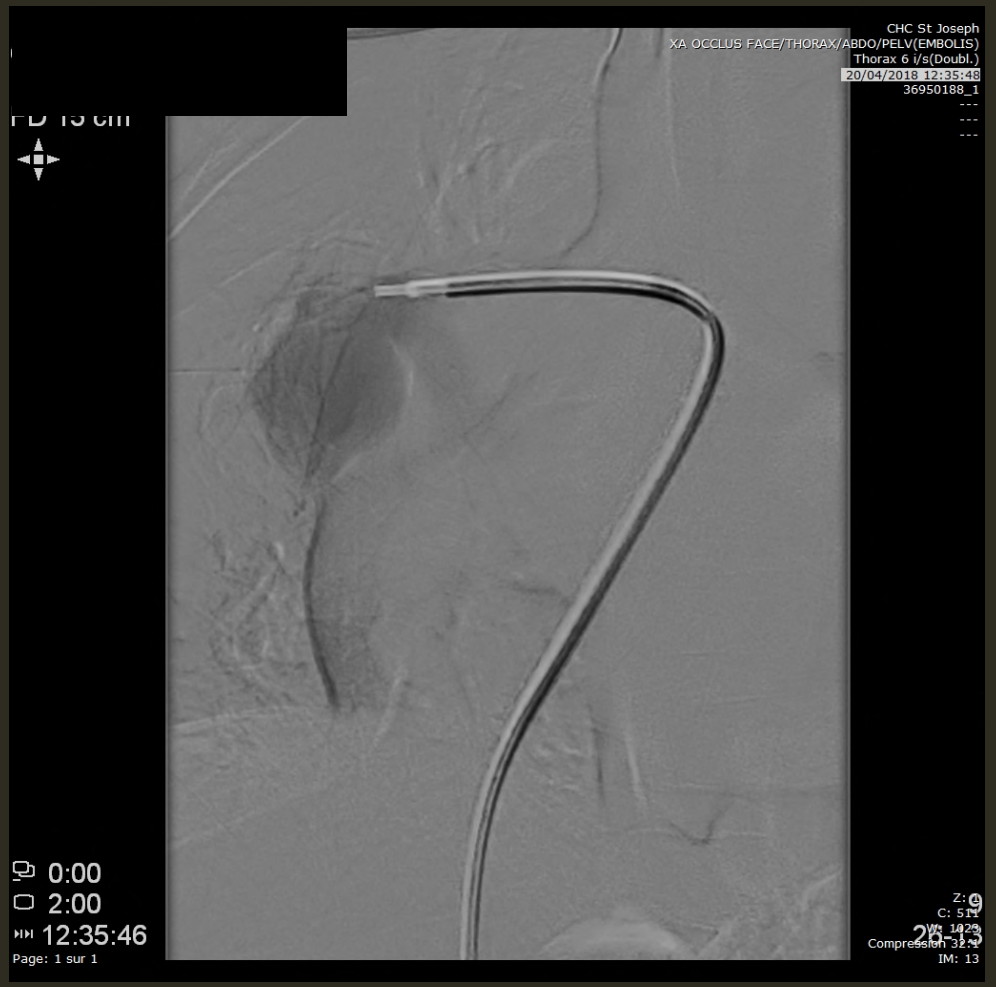
PATIENT 1

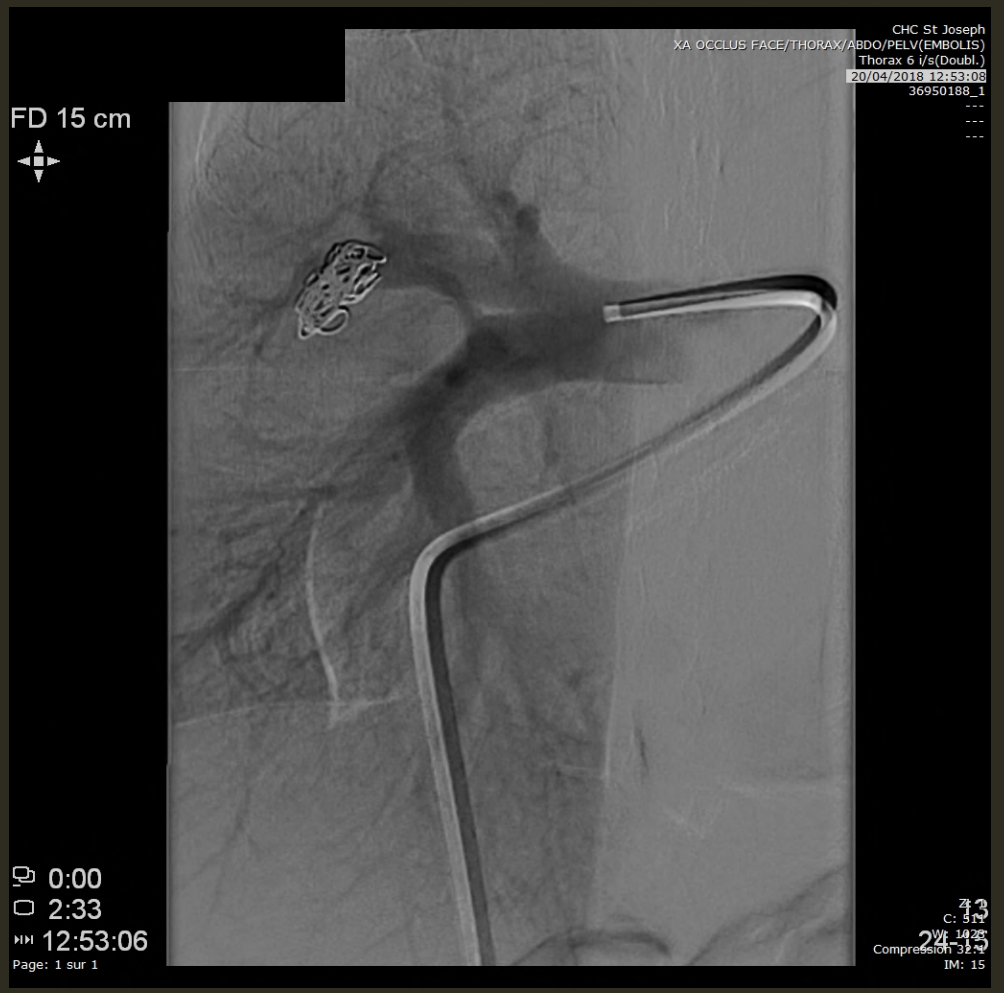




Chest Angio-CT

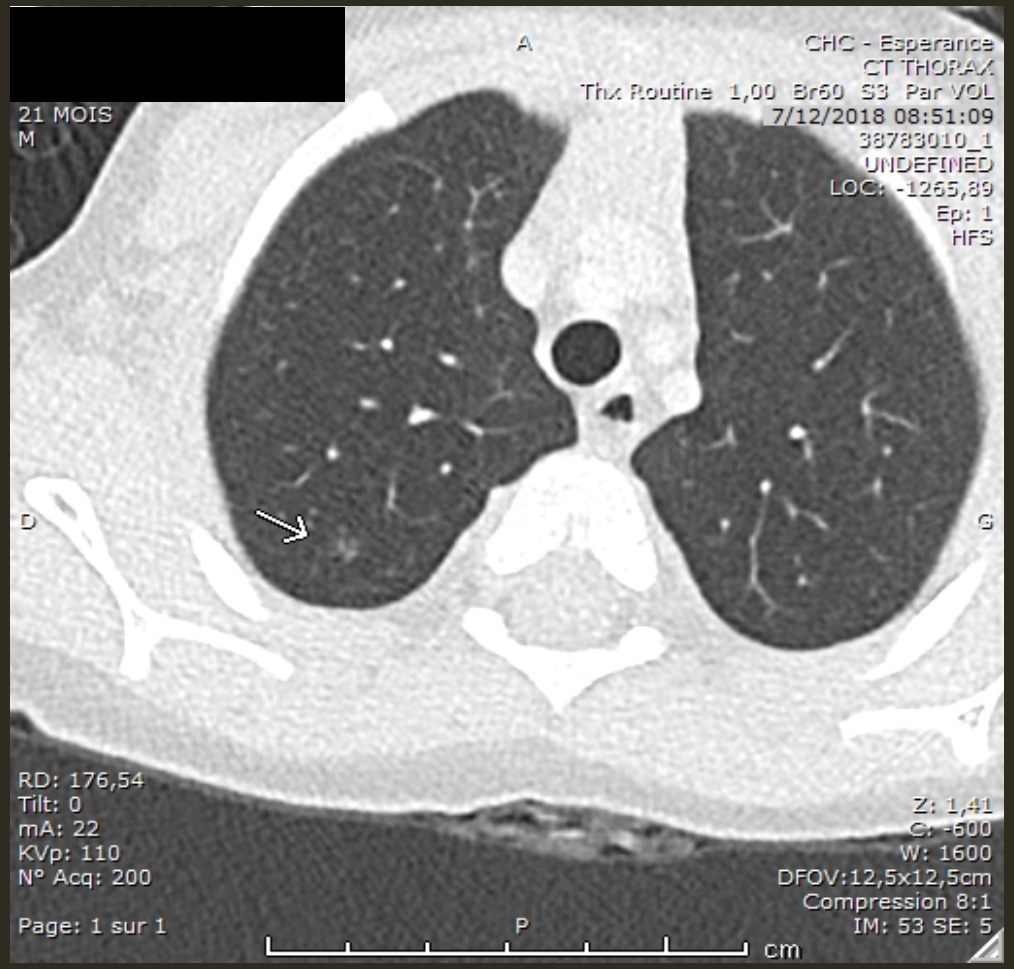
PATIENT 1

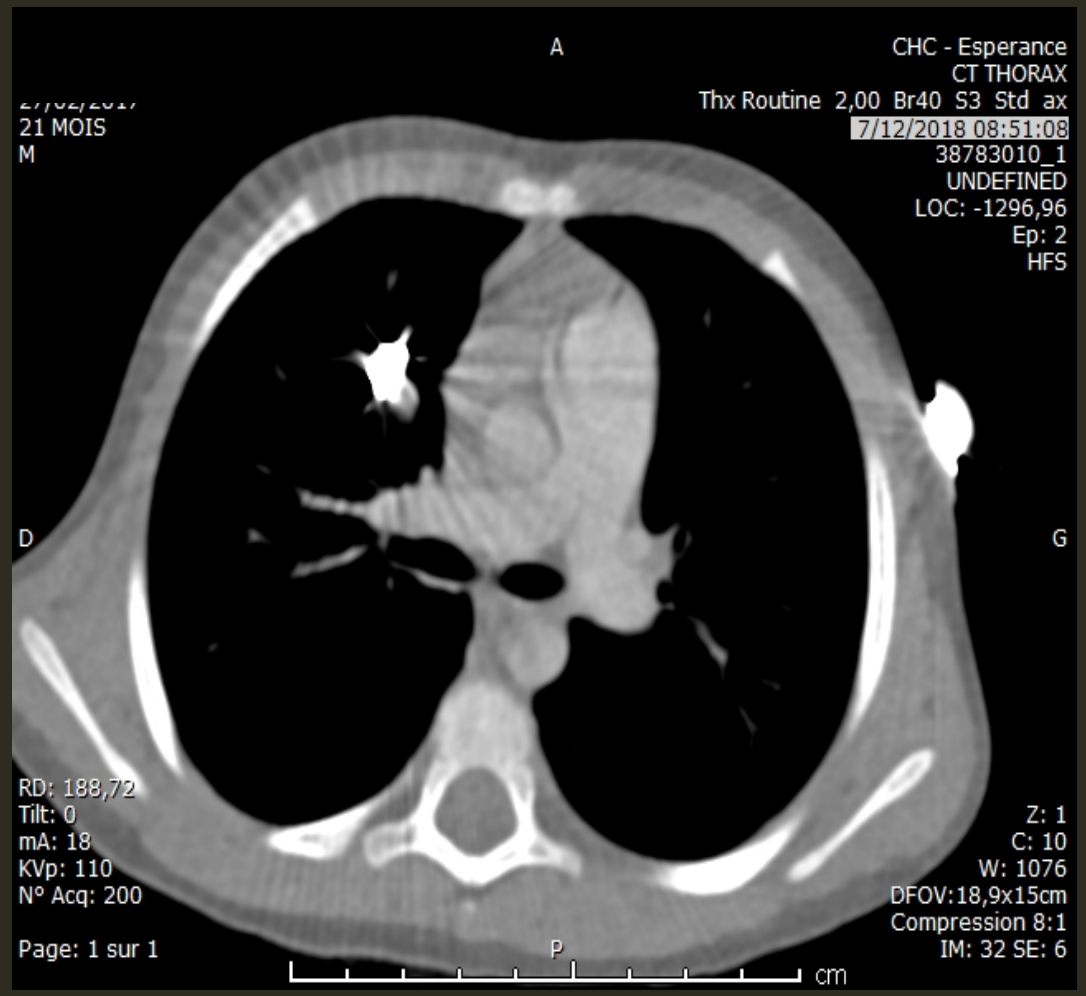




Percutaneous transcatheter embolization (courtesy: Dr Brisbois)

PATIENT 1





Chest Angio-CT
J 9 months

PATIENT 2

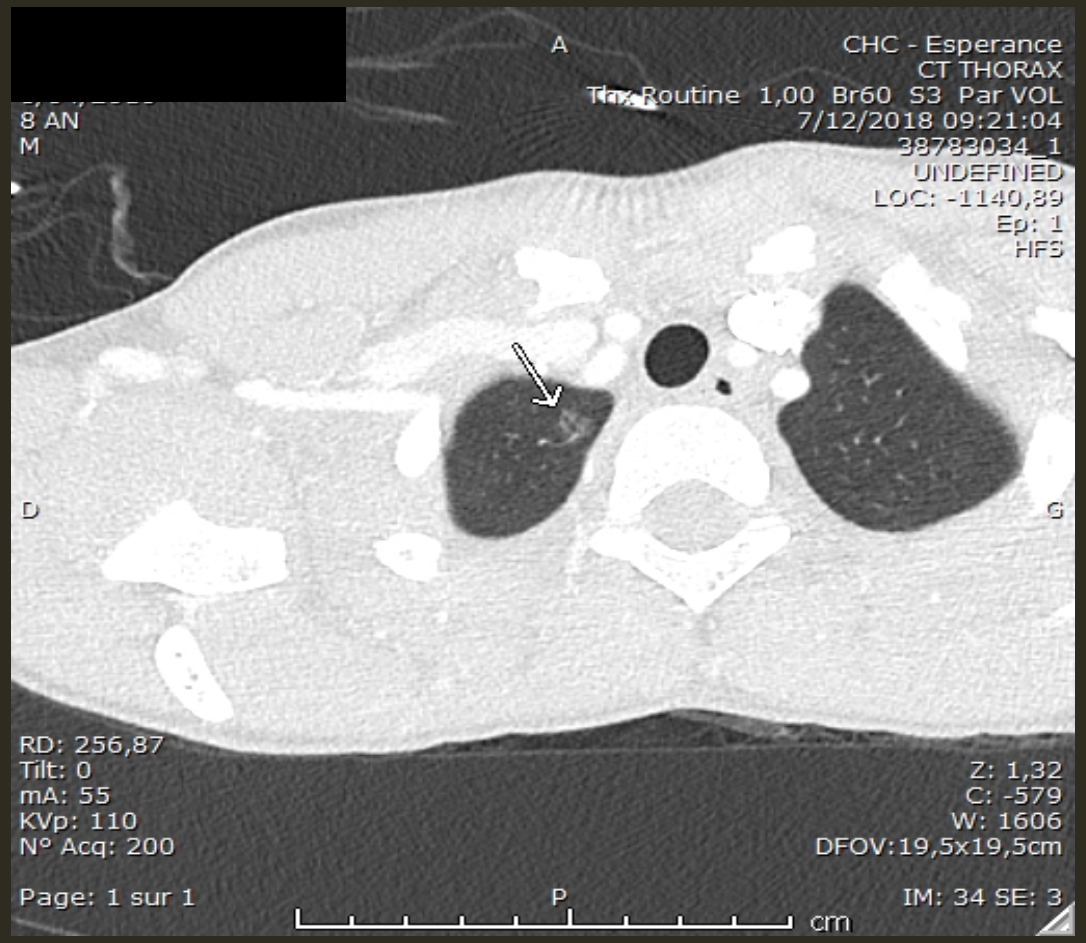
Boy, 8 y.o

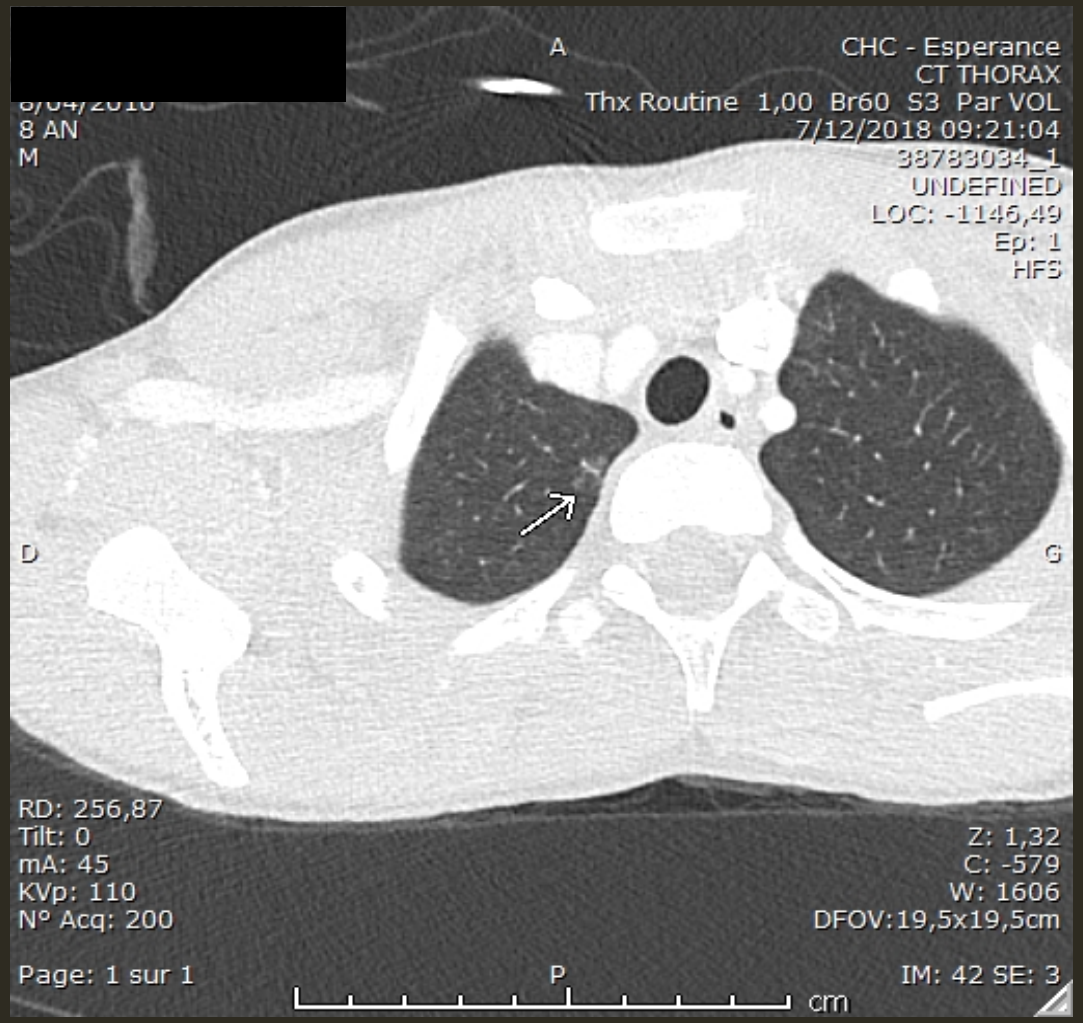
Epistaxis

Step brother

Family history of HHT: Screening → ENG +

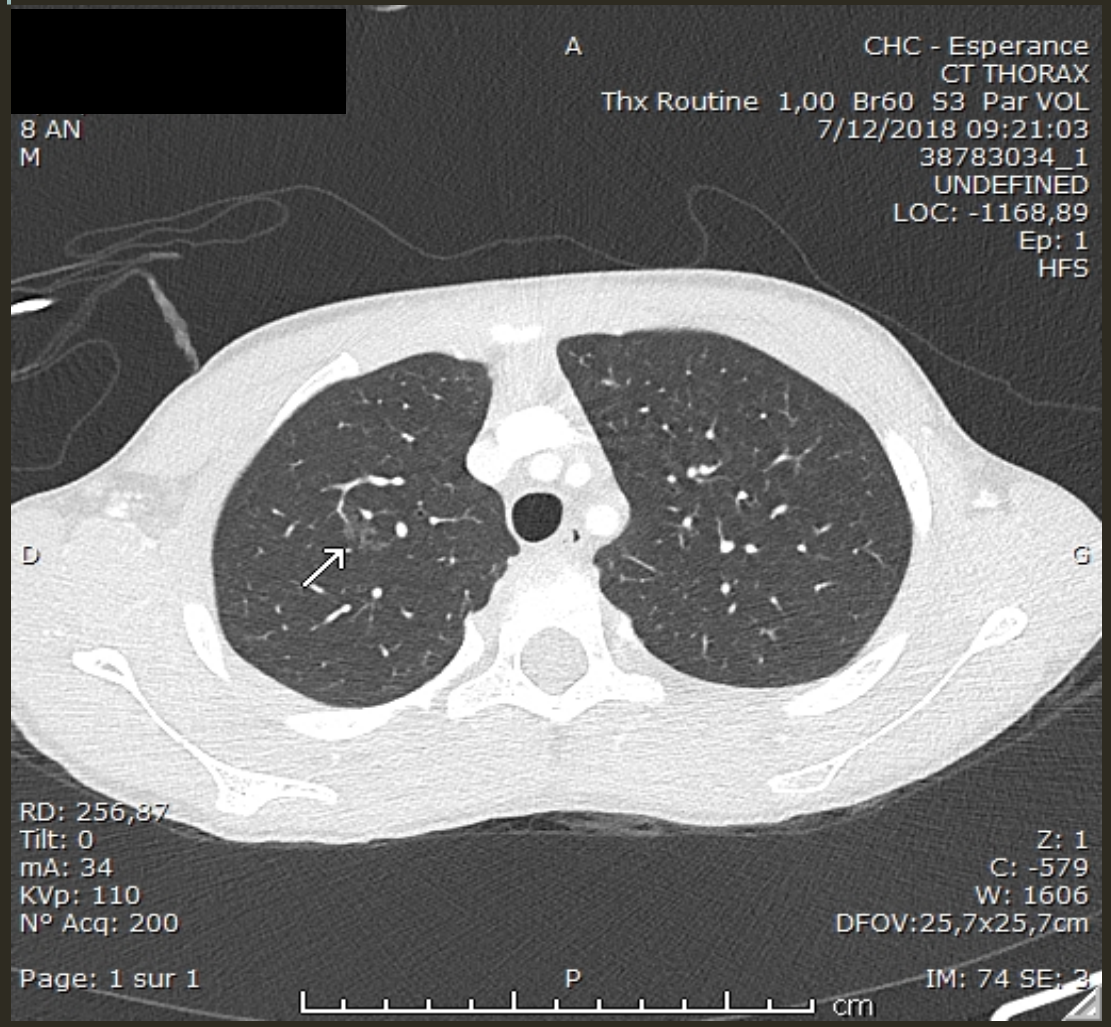
PATIENT 2

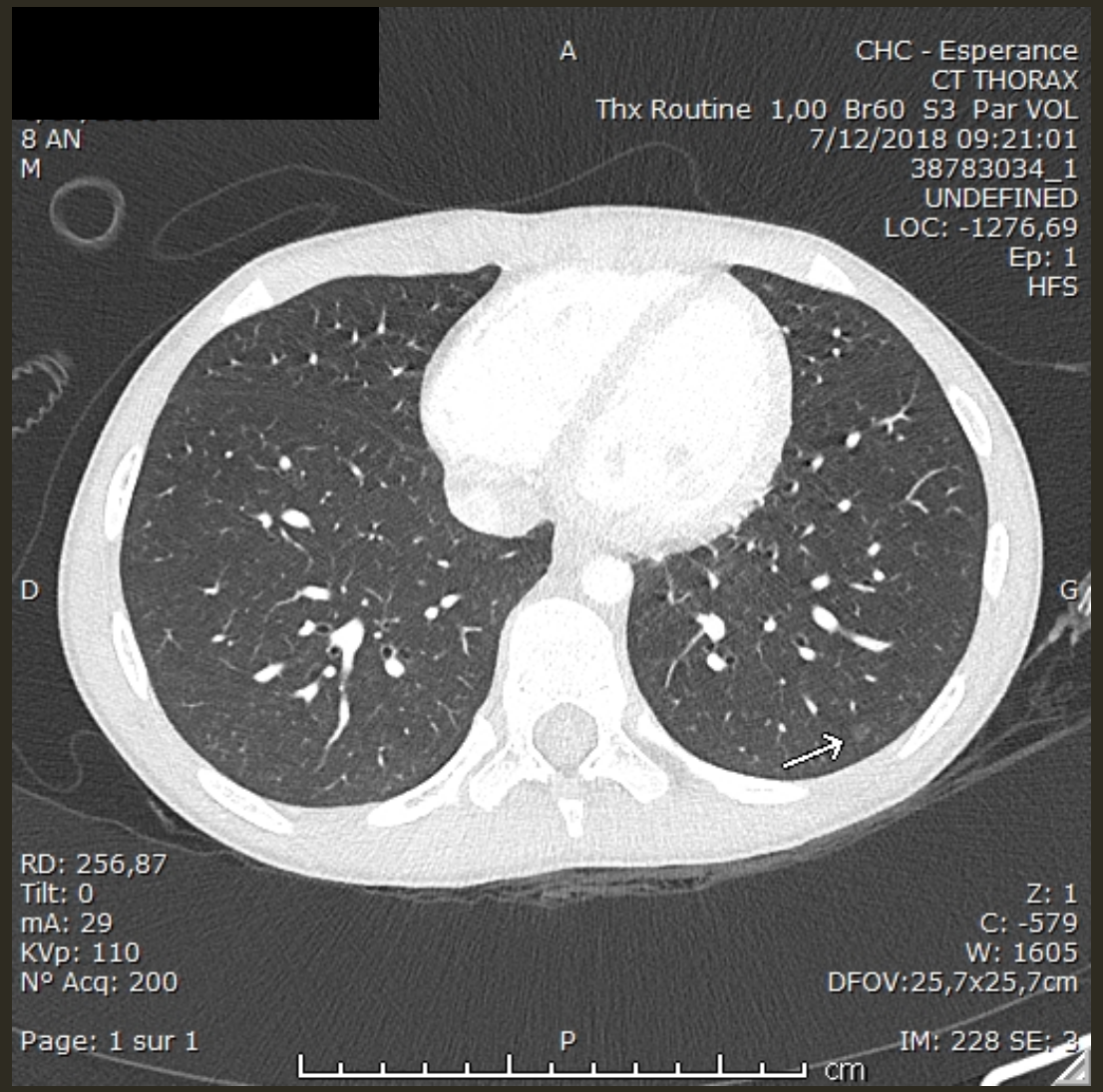




Chest Angio-CT

PATIENT 2

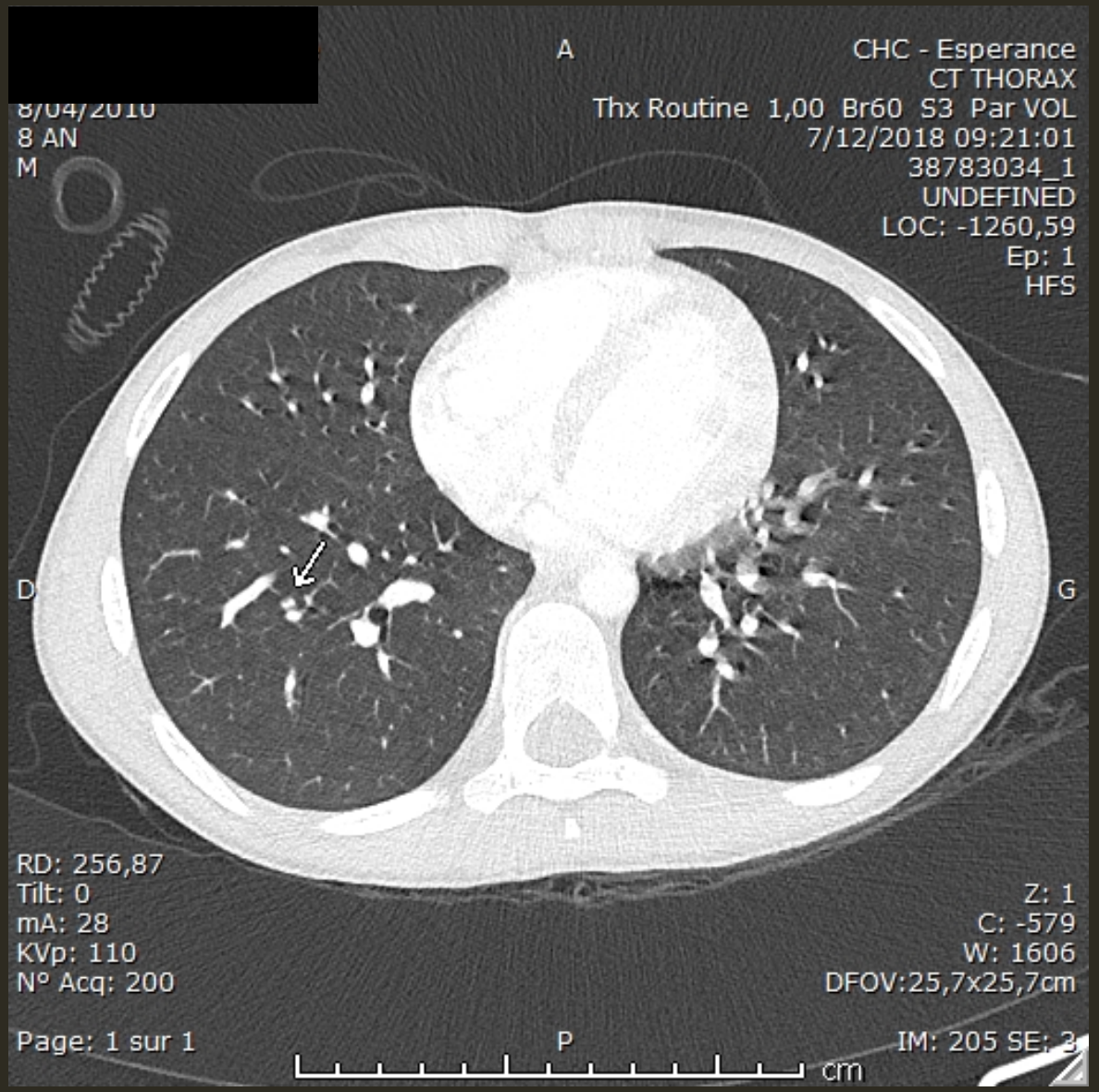




Chest Angio-CT

PATIENT 2

Chest Angio-CT



LITERATURE REVIEW:
PULMONARY ARTERIOVENOUS MALFORMATIONS IN HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)

HHT

- *General features*

PAVM

- *Definition , epidemiology and anatomical considerations*
- *Diagnosis and screening in the HHT population*
- *Clinical manifestations and complications*
- *Management*

GENERAL FEATURES (1)

Hereditary hemorrhagic telangiectasia(HHT)

= Rendu-Osler-Weber disorder

Autosomal dominant disease,

1/5000 – 1/10000

Characterized by mucocutaneous telangiectasia and arteriovenous malformations (AVMs) in several organs

Symptoms : epistaxis (mucosal telangiectasia), dyspnea (patients with large or multiple PAVMs) and hemoptysis

Definite HHT	If 3 are present
Possible or suspected HHT	If 2 are present
Unlikely HHT	If fewer than two are present
Criteria	
Epistaxis	Spontaneous recurrent nose bleeds. Night time bleeds are more suspicious
Telangiectasia	Multiple at characteristic sites—lips, oral cavity, fingers, nose
Visceral lesions	Gastrointestinal telangiectasia (with or without bleeding, PAVM, Hepatic AVM, Cerebral AVM, spinal AVM)
Family history	A first degree relative with HHT according to these criteria

Curaçao criteria:

GENERAL FEATURES (2)

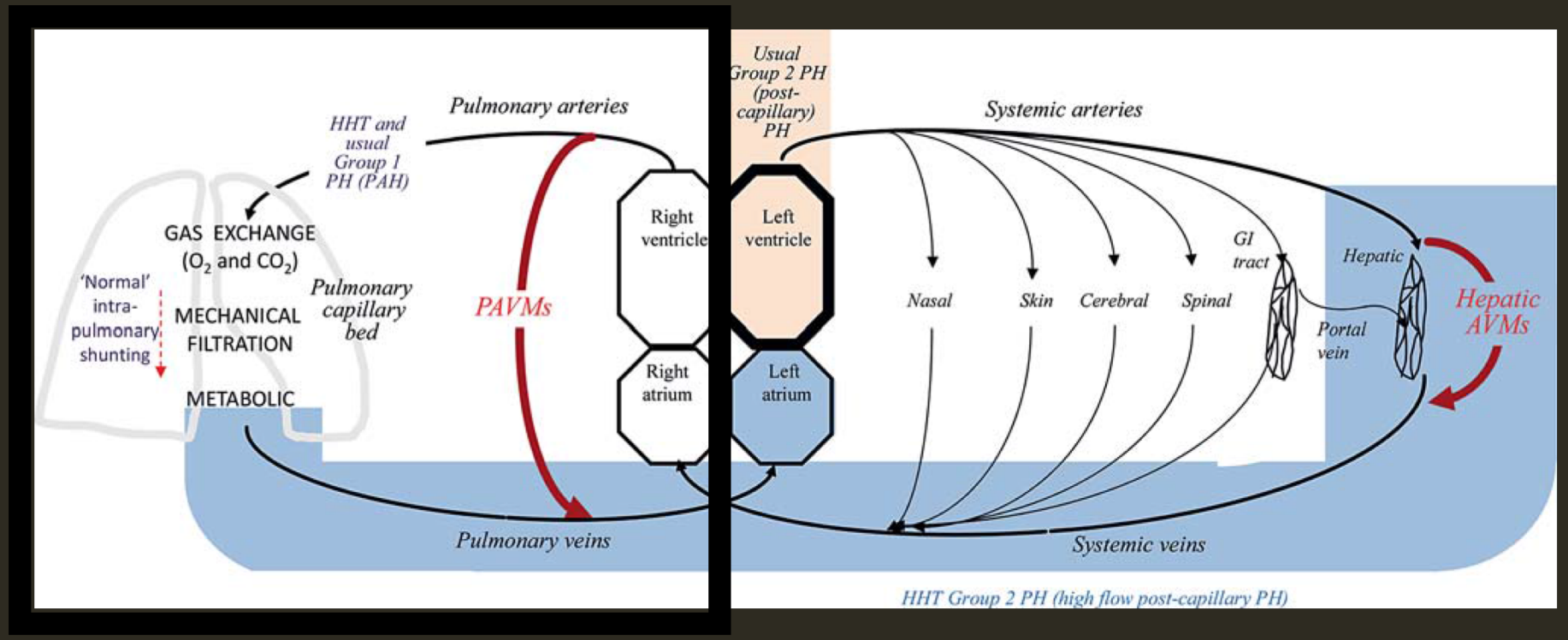
The majority of cases of HHT are due to germline mutations

- Endoglin (ENG, chr 9) ***HHT type 1*** → PAVMs and cerebral AVMs (X3-6)
- Activin receptor-like kinase1 (ALK1, chr 12) ***HHT type 2*** → hepatic AVMs (X3-6)
- SMAD family 4 (SMAD4, chr 18) ***HHT type 3***

80%

PAVM
DEFINITION

Pulmonary arteriovenous malformation (PAVM) is an abnormal communication between the pulmonary artery and pulmonary vein without an intervening capillary communication



PAVM
EPIDEMIOLOGY

About 80–90% of patients with PAVMs eventually may present with Hereditary Hemorrhagic Telangiectasia (HHT), while others are sporadic

Associated PAVMs to HHT are often multiple (50%) , bilateral, with a predominance of the lower lobes (60–95%)

Approximately, 15–35% of HHT patients may present with PAVMs

PAVM
ANATOMICAL CONSIDERATIONS(1)

3 components : one or more feeder arteries (long or very short), an aneurysm sac and one or more drainage veins

« *Simple* » *PAVM* (approximately 80%) : one or more feeder arteries from a single segmental pulmonary artery

« *Complex* » *PAVM* : several afferent feeder arteries from several segmental arteries

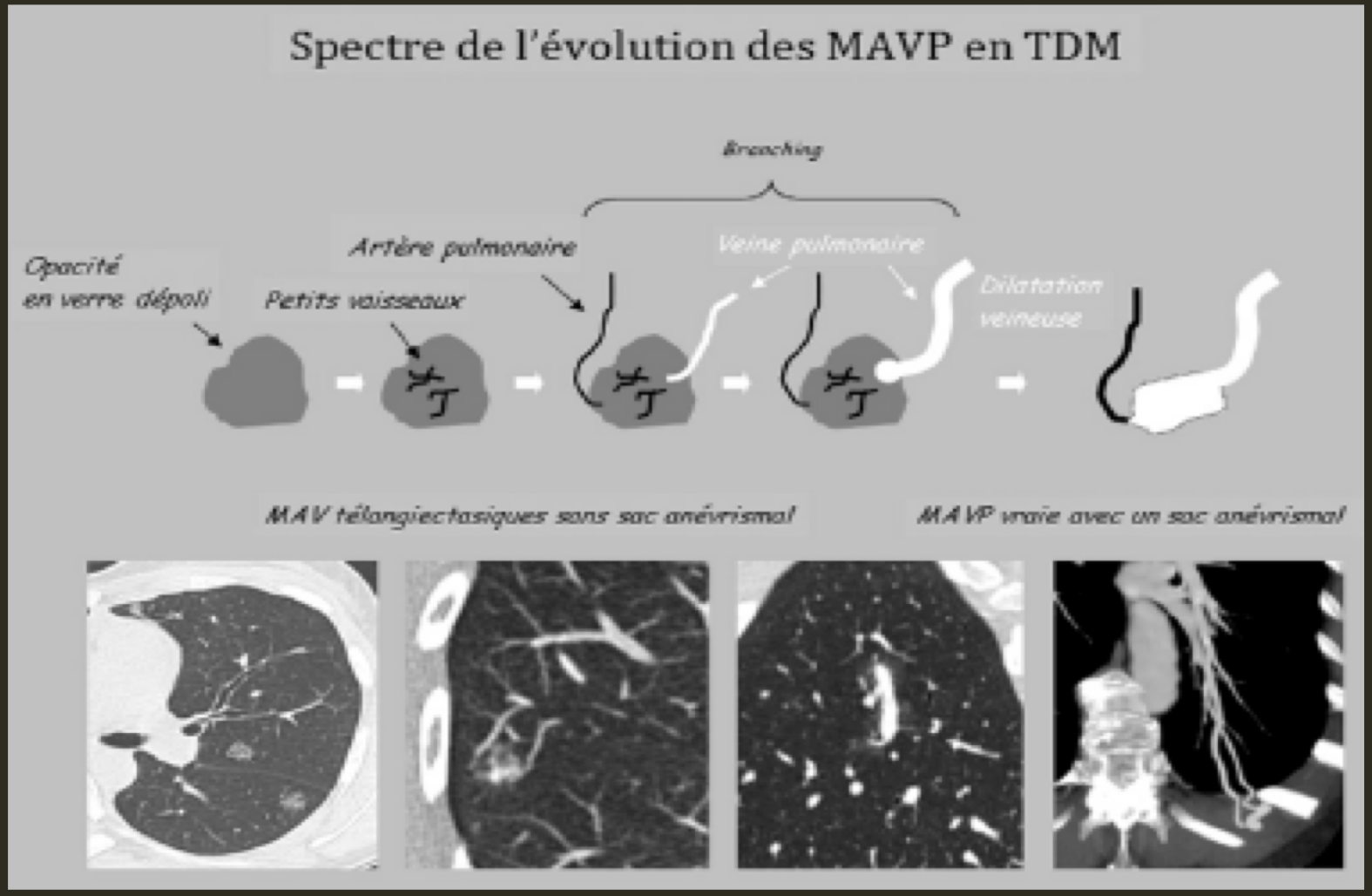
PAVM
ANATOMICAL CONSIDERATIONS(2)

In patients with HHT, the pulmonary distribution of PAVMs can be either random or diffuse (5-7%)

« Diffuse PAVM » varies depending on whether the disease affects all the segmental arteries or all the sub-segmental arteries of at least one lobe

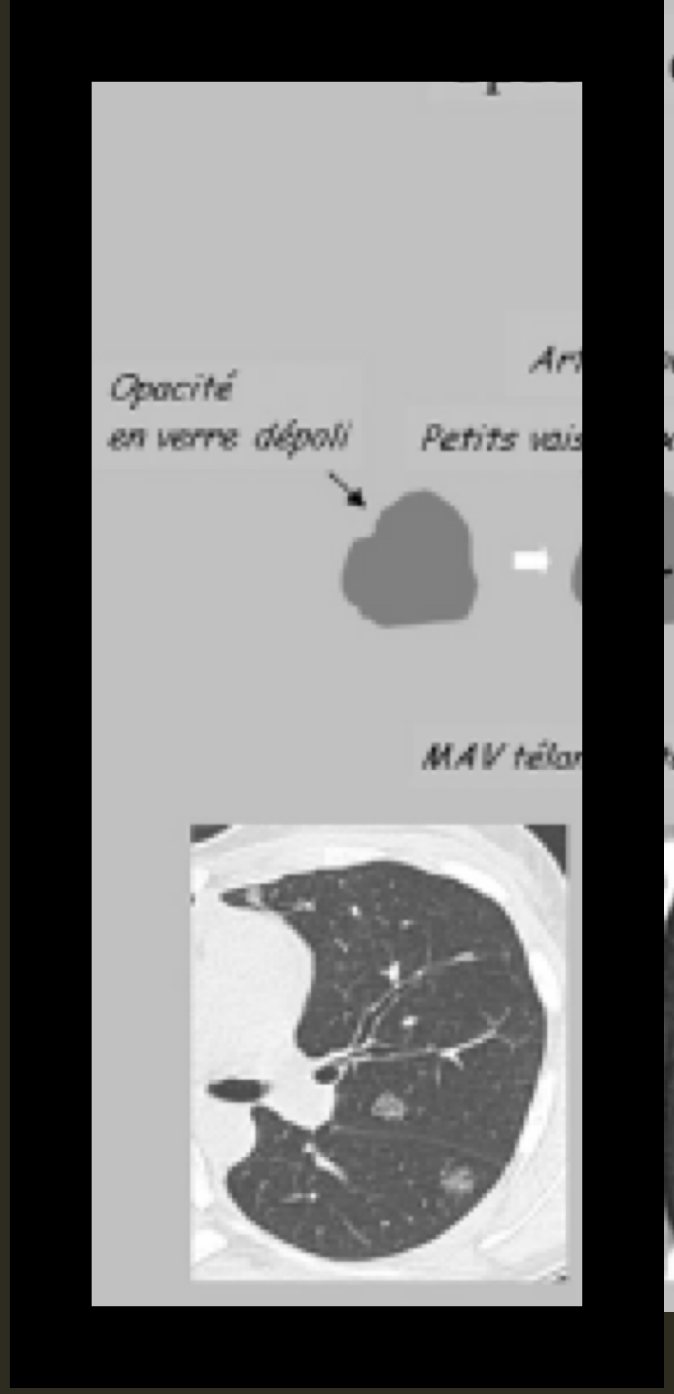
PAVM
ANATOMICAL CONSIDERATIONS(3)

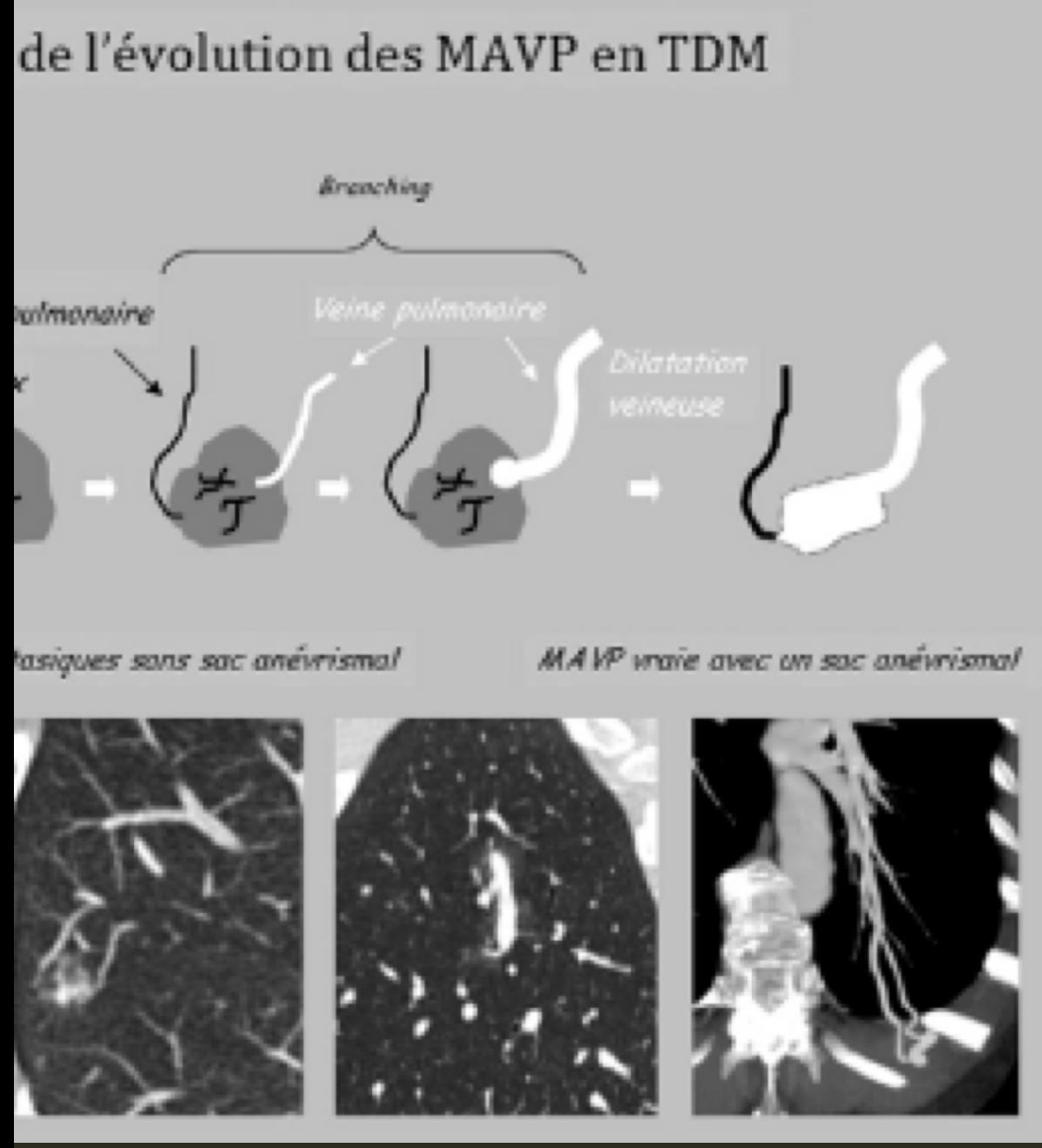
PAVM growth follows 4 main stages



STEP 1

A ground glass nodule corresponds to the initial dilatation post-capillary venules associated with an inflammatory cell infiltrate





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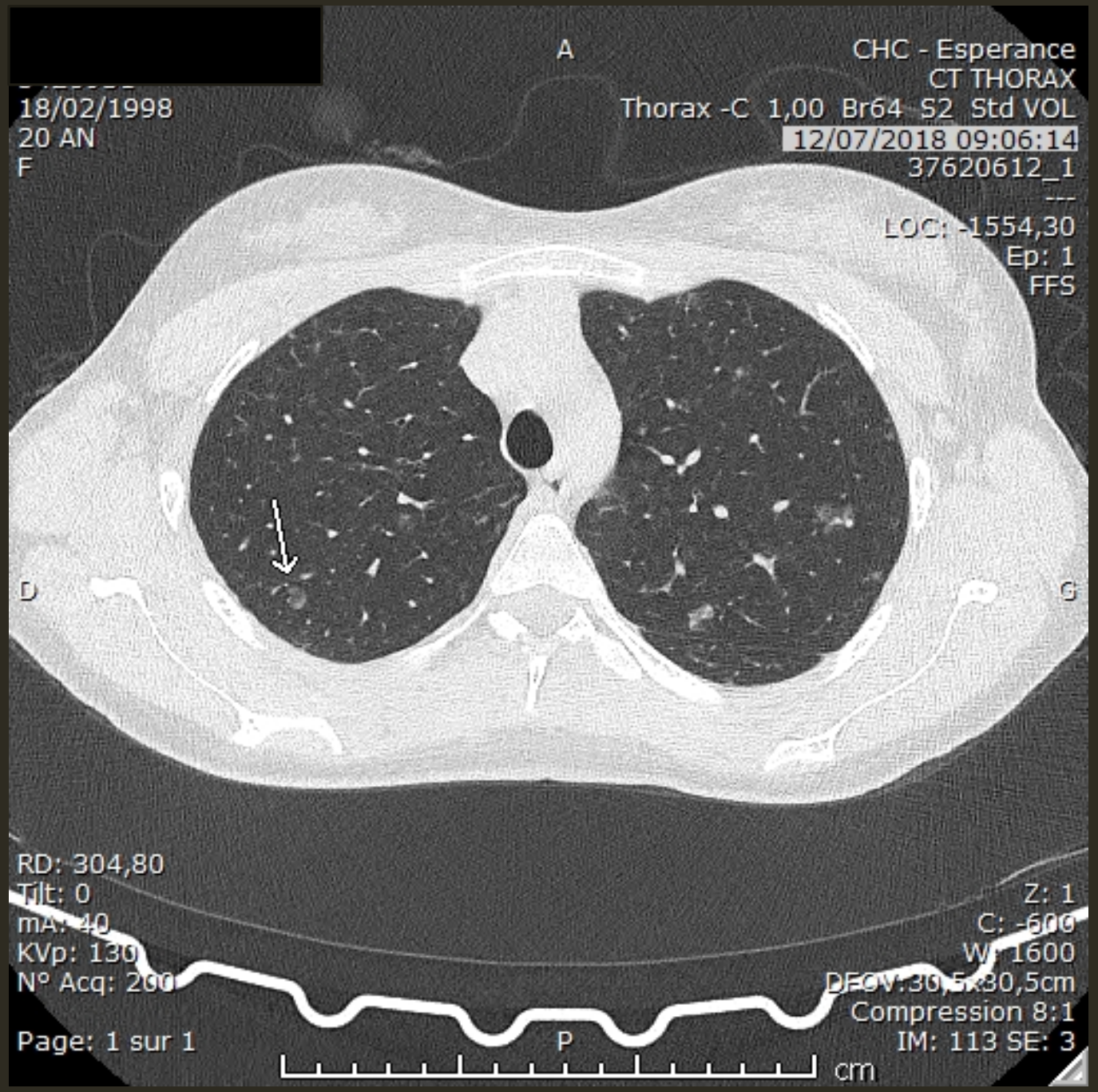
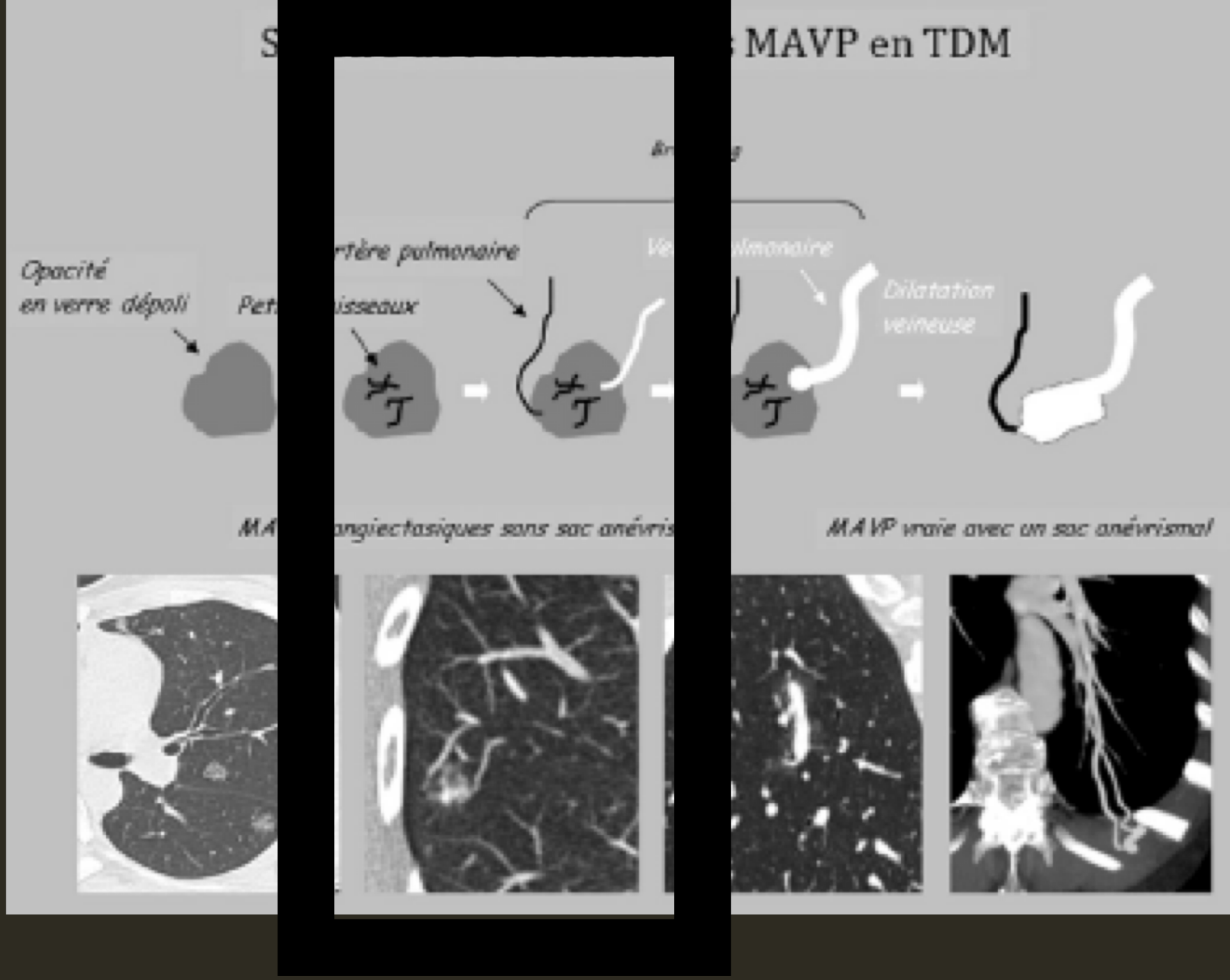


STEP 2

Small vessels visible in the ground glass nodule represent vascular connection and communication between the precapillary pulmonary artery and the postcapillary venules within the capillary sector



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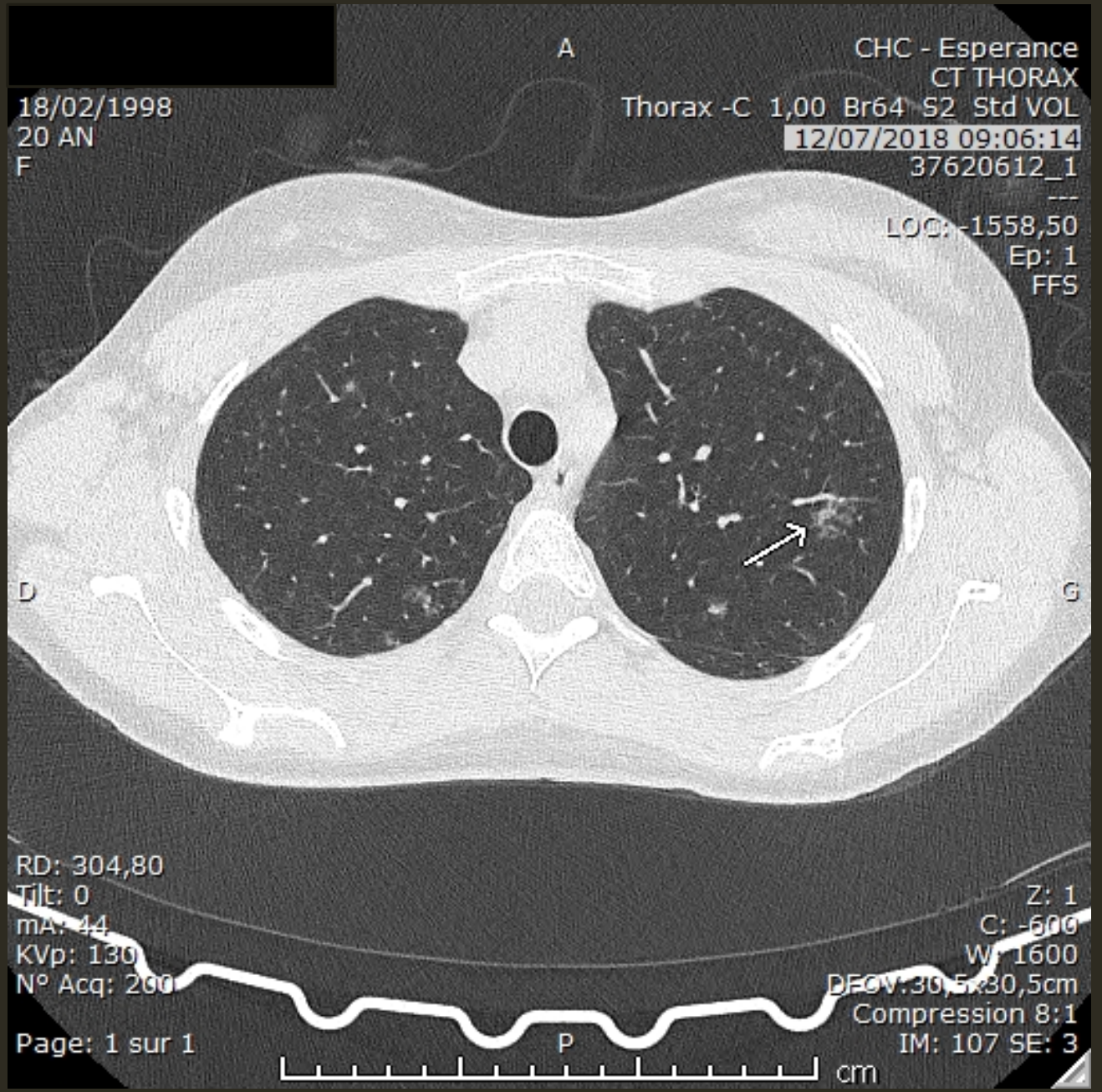
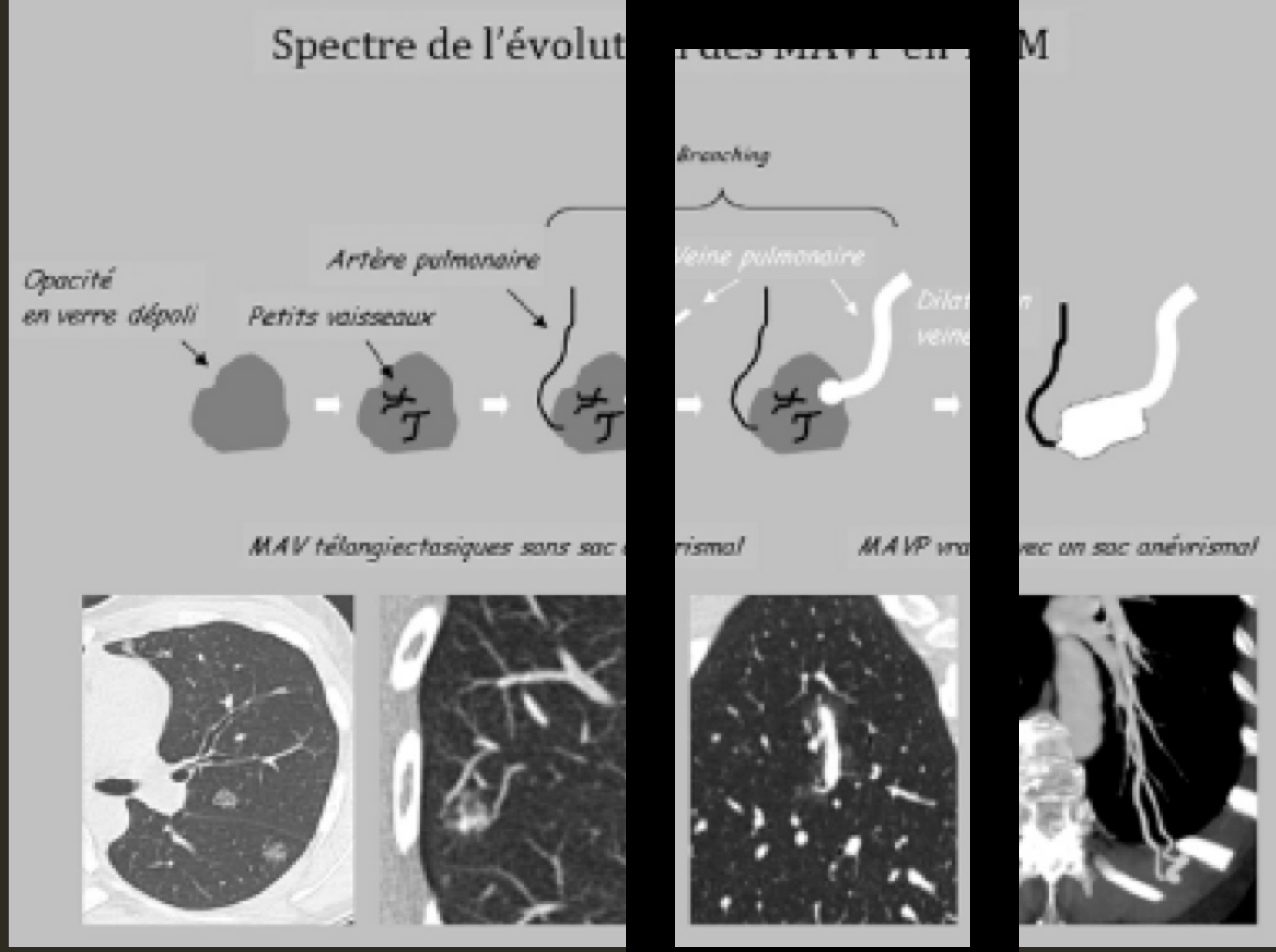


STEP 3

An increase in the size of the drainage vein



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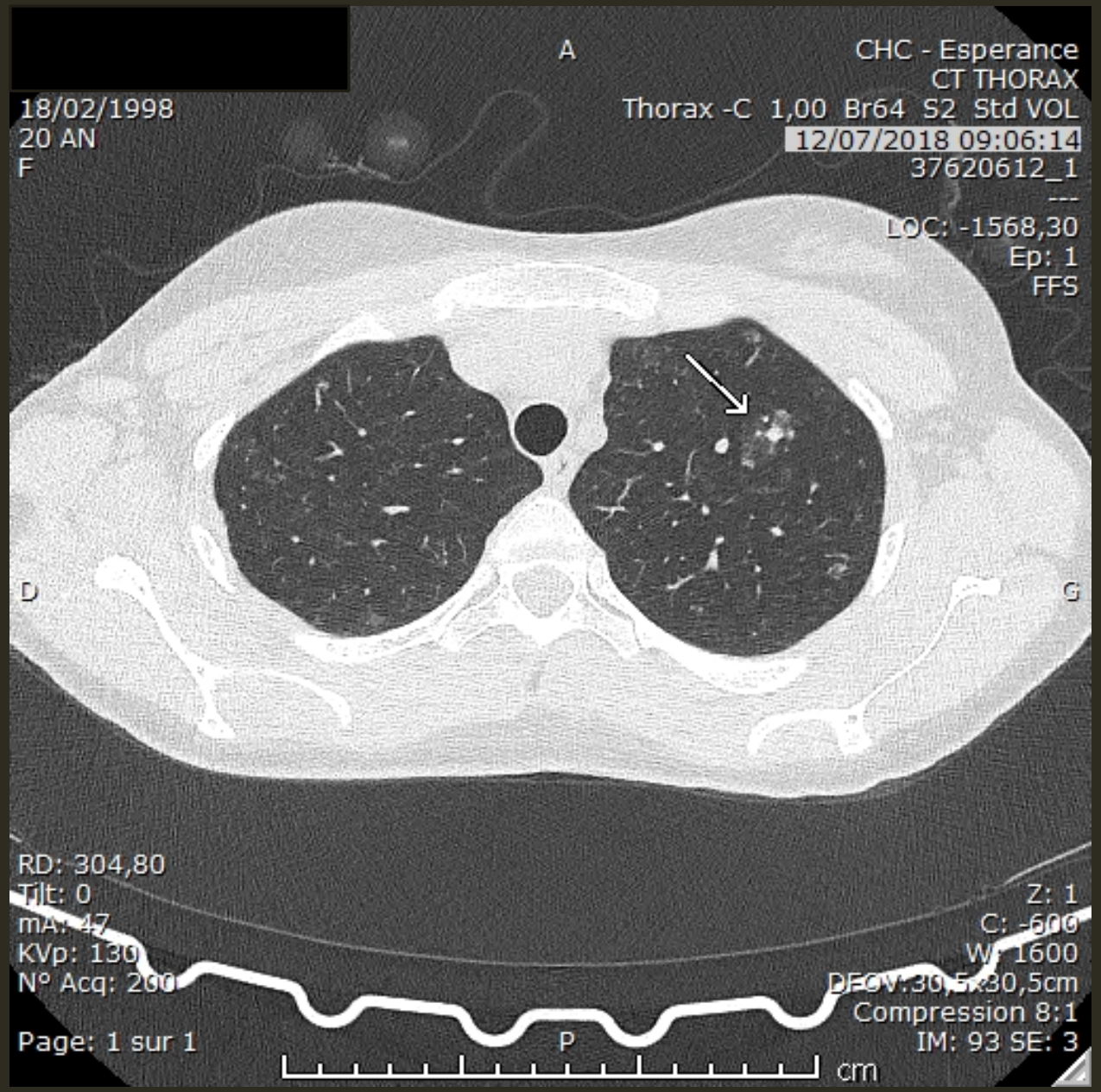
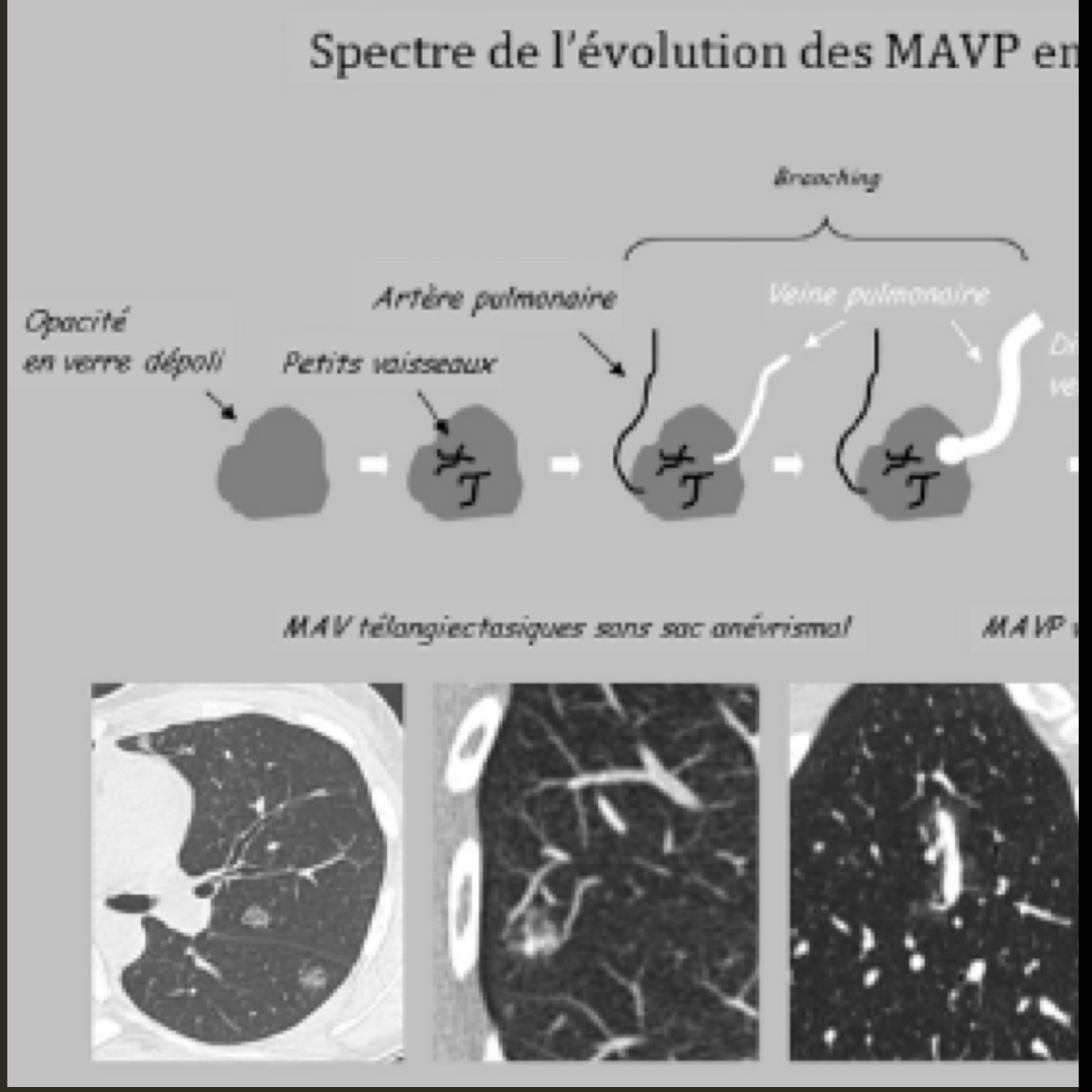
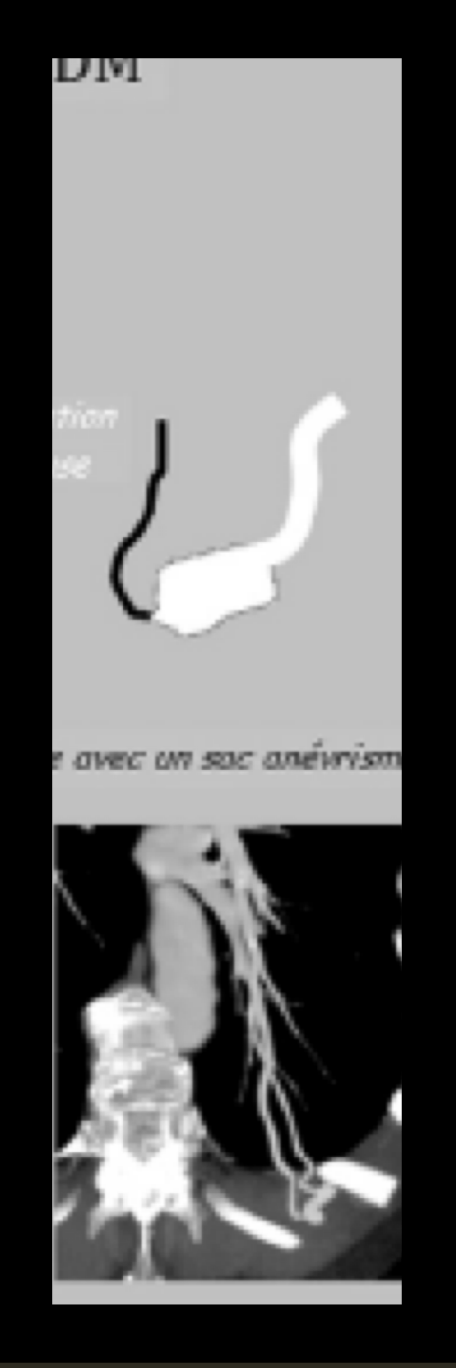


STEP 4

The definitive PAVM corresponds to a feeder pulmonary artery, an aneurysm sac and a drainage vein increased in diameter, with concomitant disappearance the ground glass appearance





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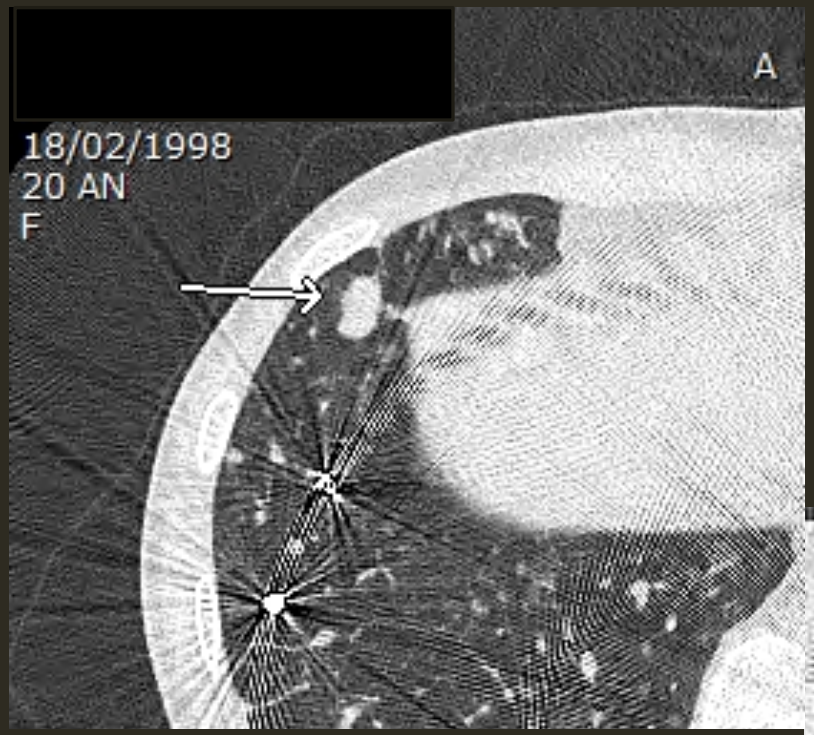


PAVM
DIAGNOSIS IN THE HHT POPULATION

Patients with HHT are diagnosed based on the Curaçao Criteria

CT pulmonary angiography is considered the gold standard for diagnosis of PAVMs

Magnetic Resonance Imaging (MRI) has seen greater usage in detecting, pre-embolization planning and evaluating treated PAVMs

PAVM
SCREENING IN THE HHT POPULATION

- *Transthoracic contrast echocardiography (in high-risk patients)*

- *Positive screening can be confirmed with multidetector thoracic CT C-*

In children : physical exam (for cyanosis, dyspnea, clubbing), pulse oximetry, chest radiography and/or TTCE (100% sensitivity and negative predictive value)

PAVM
CLINICAL MANIFESTATIONS AND COMPLICATIONS(1)

PAVMs are usually asymptomatic

PAVMs have a tendency to grow and increase in size over time

Various factors affect growth : puberty, pregnancy and pulmonary arterial hypertension

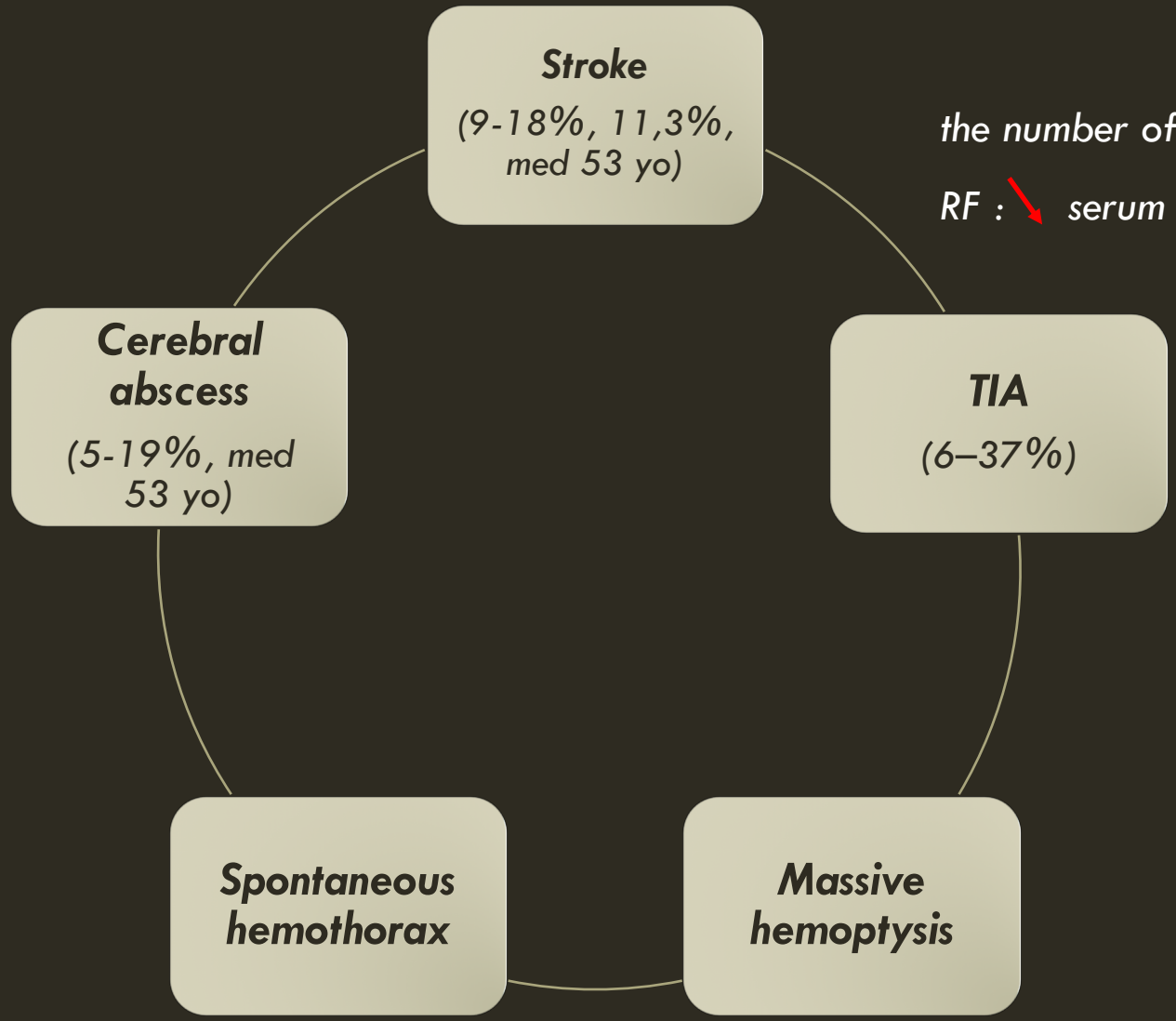
PAVM
CLINICAL MANIFESTATIONS AND COMPLICATIONS(2)

The presence of symptoms best correlates with the size of the PAVMs rather than the number of the lesion

Symptoms : dyspnea, intrapulmonic hemorrhage, neurological symptoms/deficits, palpitations, cough, and chest pain

The major consequences of PAVMs are impairment of gas exchange (hypoxemia) and paradoxical emboli (feeding artery >3 mm)

PAVM
CLINICAL MANIFESTATIONS AND COMPLICATIONS(3)



the number of PAVMs +++

RF : ↘ serum iron, ↗ serum fibrinogen

PAVM
MANAGEMENT(1)

The treatment options include :

- Percutaneous image guided embolization (success: 75-83% , no size threshold for treatment, major complications: 1%)
- Surgical removal
- Hormonal therapy

PAVM
MANAGEMENT(2)

The success of embolization is appreciated by the retraction complete MAVP embolized on a control scanner

Follow up (Ct scan) :

-1x/year : diffuse PAVM

-1x/5 years : other PAVM

PAVM management requires life-long follow-up and continued vigilance observing for PAVM growth or recanalization

THANKS...

BIBLIOGRAPHIE

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