



Hypertension Pulmonaire : quand l'imagerie nous parle de génétique

« 6min – 6 diapos »

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**Pôle des voies
respiratoires**

CHU
TOULOUSE

Services pneumologie, chirurgie thoracique, ORL



**UNIVERSITÉ
TOULOUSE III
PAUL SABATIER**



Université
de Toulouse

GROUP 1 Pulmonary arterial hypertension (PAH)

1.1 Idiopathic

1.1.1 Non-responders at vasoreactivity testing

1.1.2 Acute responders at vasoreactivity testing

1.2 Heritable^a

1.3 Associated with drugs and toxins^a

1.4 Associated with:

1.4.1 Connective tissue disease

1.4.2 HIV infection

1.4.3 Portal hypertension

1.4.4 Congenital heart disease

1.4.5 Schistosomiasis

1.5 PAH with features of venous/capillary (PVOD/PCH) involvement

1.6 Persistent PH of the newborn

?

?

Mutations « non syndromiques »

BMPR2+++

SMAD9

CAV1

BMP9

KCNK3

Mutations « syndromiques »

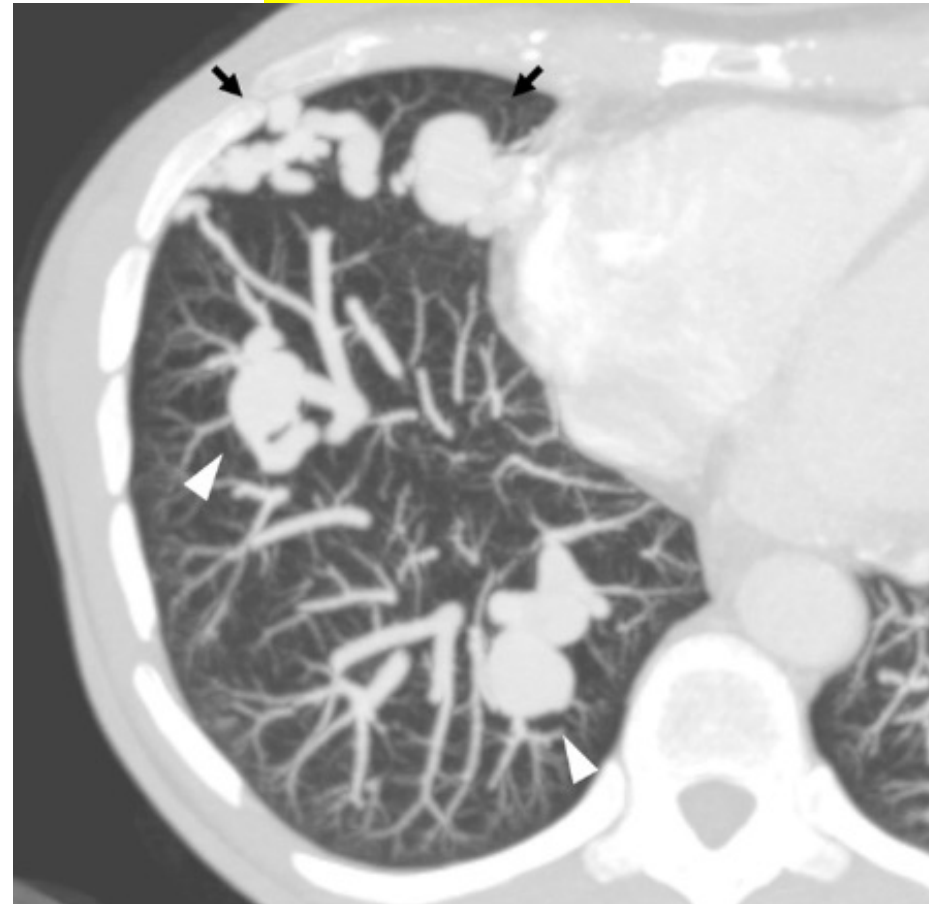
M Eyries 2017

Imagerie d'HTAP

> *Pediatr Radiol.* 2022 Jun 11. doi: 10.1007/s00247-022-05413-8. Online ahead of print.
Chest computed tomography findings of ground-glass nodules with enhancing central vessel/nodule in pediatric patients with BMPR2 mutations and plexogenic arteriopathy

Imagerie d'HTAP + Signes associés

HTAP +



Maladie de Rendu-Osler

HTAP +

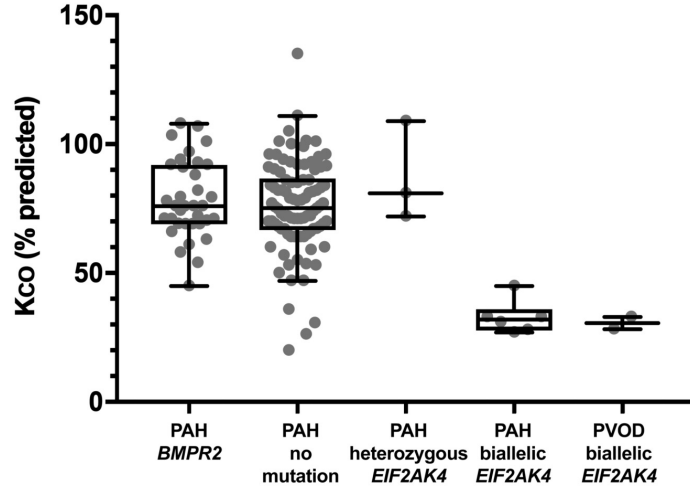
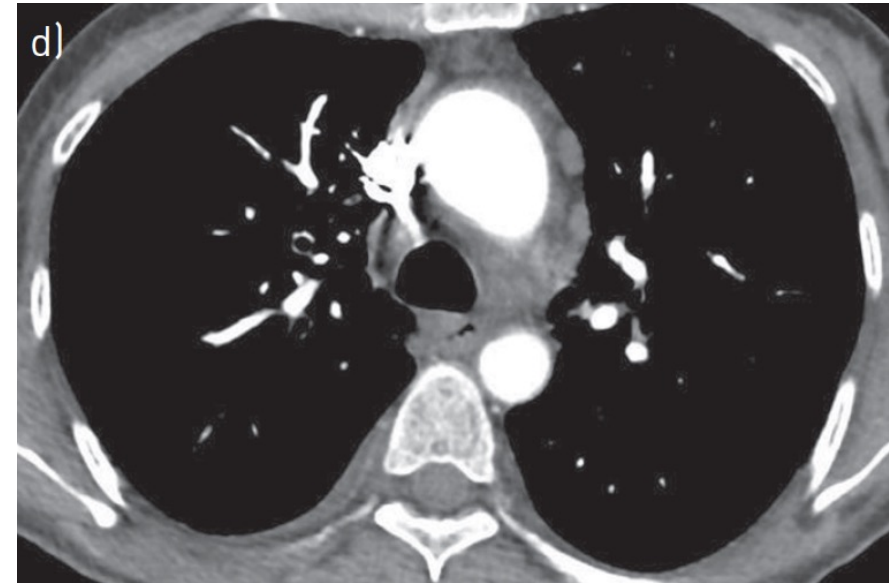
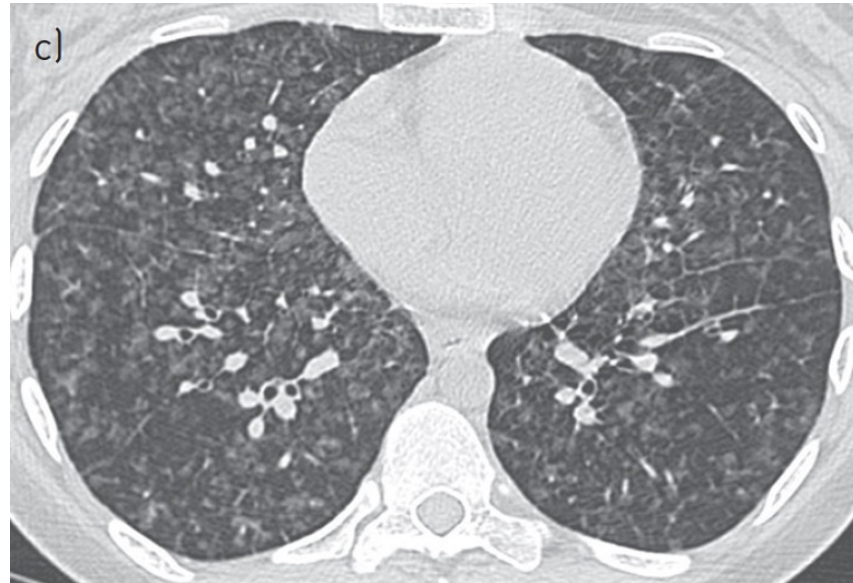


Figure 2. The transfer coefficient for carbon monoxide (Kco) is influenced by genotype in pulmonary arterial hypertension. Patients with FEV₁ < 80 % predicted and FVC < 80 % predicted and diagnosed with PAH or PVOD/PCH after 50 years of age excluded from the plot.

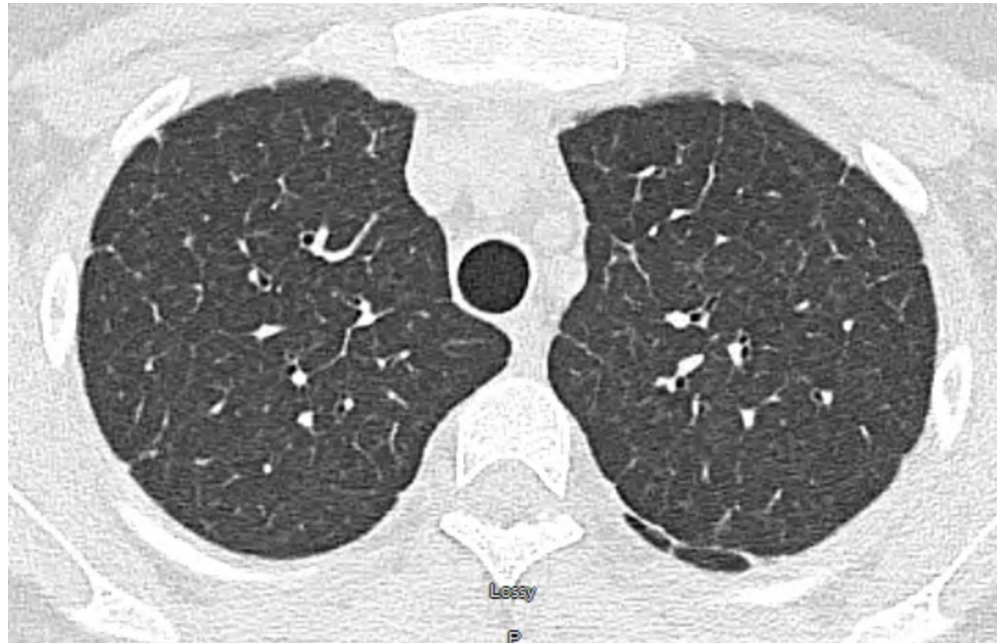
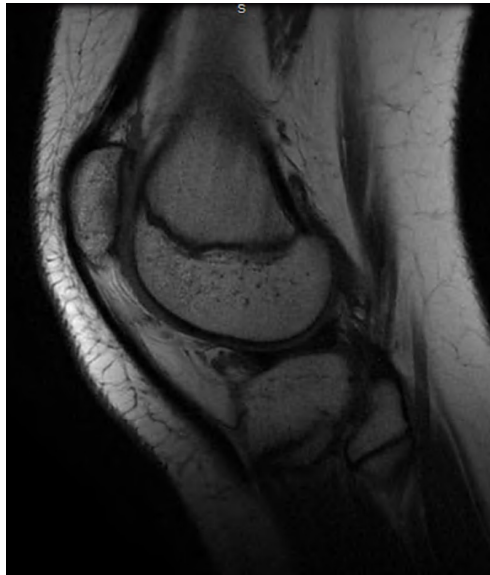


Circulation 2017

ERJ 2016

Maladie Veino-Occlusive (génétique)

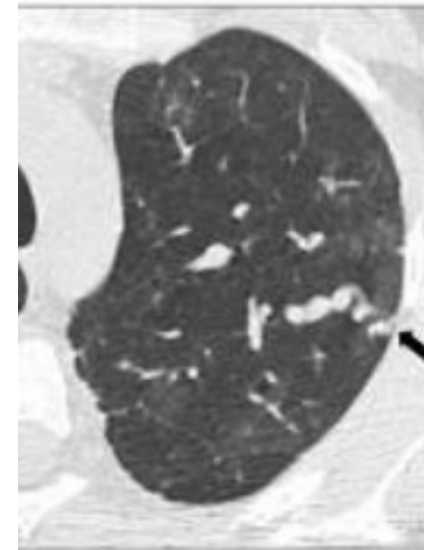
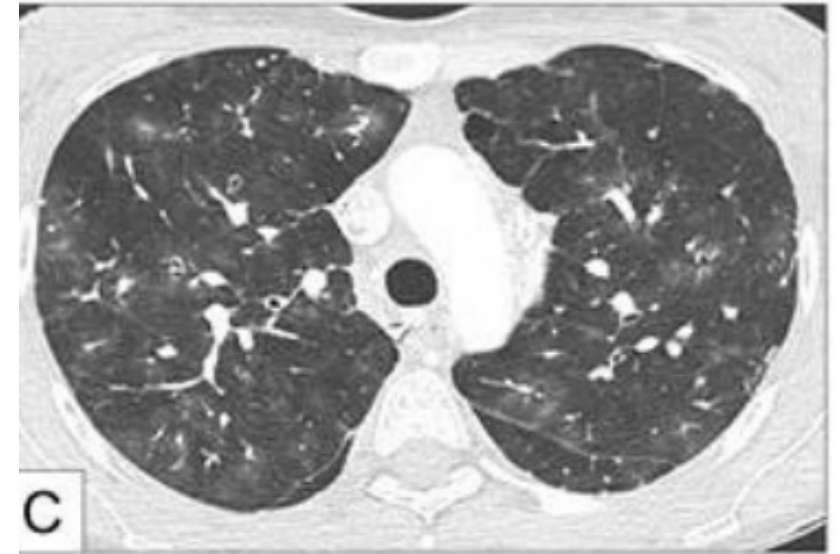
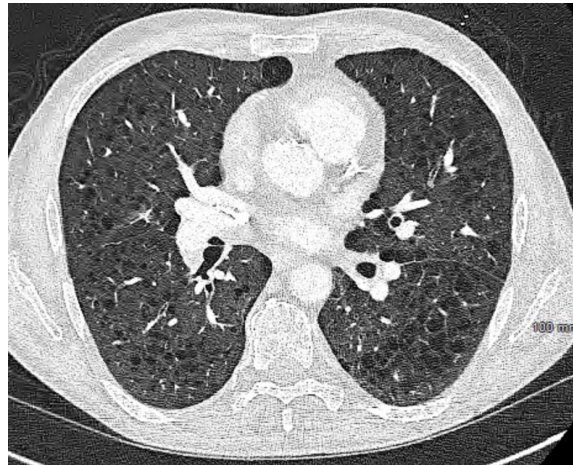
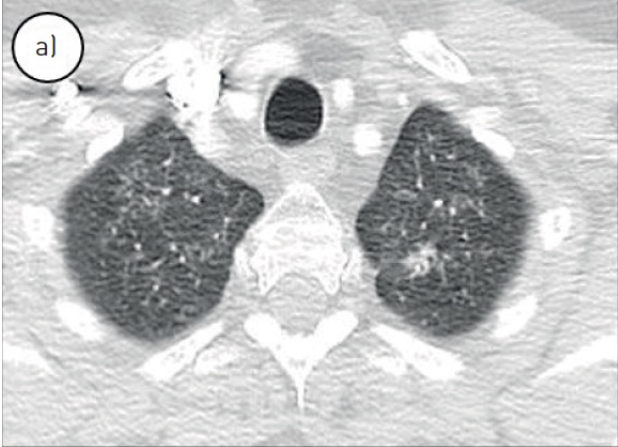
HTAP +



Syndrome coxo-podo-patellaire

Autres gènes du développement

HTAP +



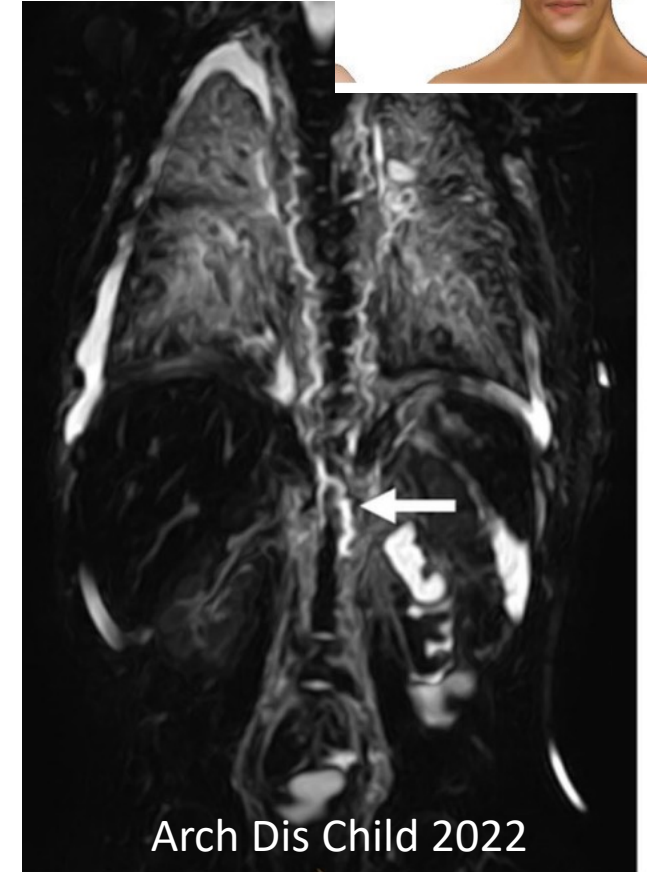
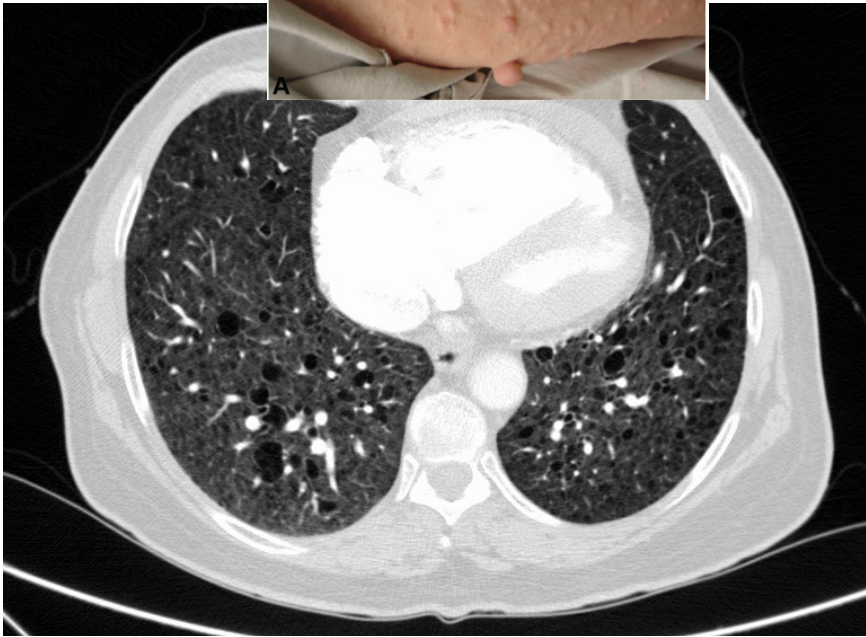
ERJ 2020

KDR

SOX17

ERJ 2022

HTAP +

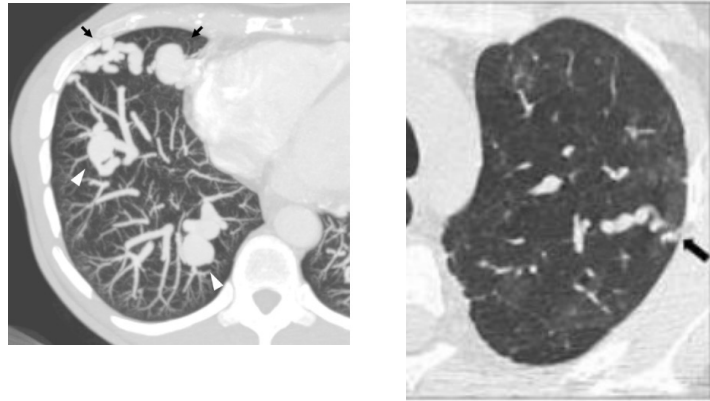


Neurofibromatose 1

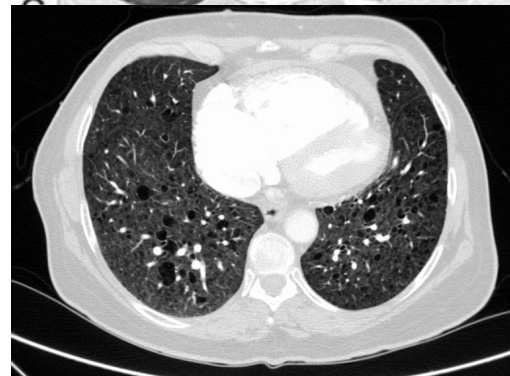
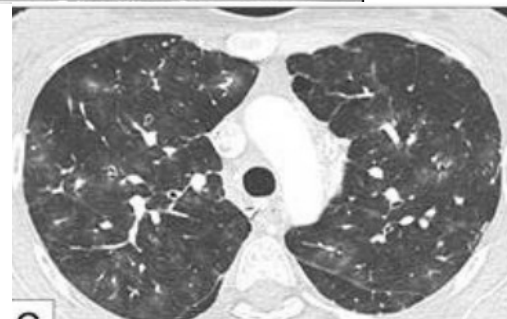
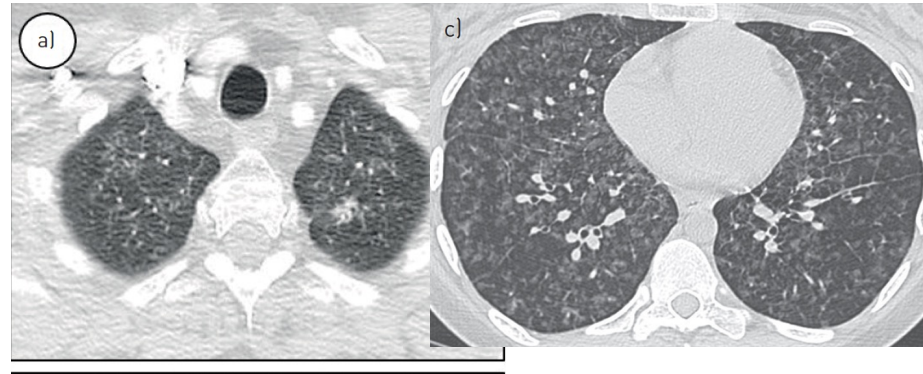
S. d'Alagille

S. de Noonan

Les vaisseaux



Le parenchyme



Autres atypies

