

# General History

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- Sex : female
- Birth Date : 68 / 02 /10
- Date of Admission : 91 /08 / 04

# Chief Complain

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- Epigastric pain with bloody vomitus for 1 day

# Present Illness

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- This 22 year-old girl is a case of tuberous sclerosis who was diagnosed at Chang-Geng Memory Hospital when she was 4 years old. In recent years, she was on regular F/U at our OPD. Besides, she has IDA under Ferrum Hausman tablet supplement.

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- She came to our ER for help tonight due to epigastric pain with bloody vomitus noted for 1 day at home. Dizziness, palpitation are complained too. Tracing back her recent course, she has tarry stool passage in recent one week. Besides, cough with yellowish sputum, fever, dyspnea are noted in recent 3 days. She denied dysuria, headache, chills, nor constipation.

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- ER :
  - Leucocytosis (WBC 13130 ,Neut 88.0% )
  - CXR : infiltration at RLL and LLL.
  - Hb :4.7, HCT :17.2 , MCH :21.3.
  - So under the impression of anemia ,GI bleeding and pneumonia ,she was admitted.

# Family history

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- Father , grandfather has Neurofibromatosis or lipoma history
- 表姐:brain tumor

# Personal history

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- Smoking : denied
- Alcohol drinking : denied
- Allergies : NKA

# Past history

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- DM : denied
- HTN :denied
- Renal tumor (Angiomyolipoma) :87/6 ;87/7
- Neurocutaneous syndrome : 87/7
- EPS,seizure :87/12
- Esophageal ulcer :90/4



# Physical examination

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■ BP : 130/70 ,TPR : 37.7,104,22

■ Conjunctiva :**pale**

■ Chest : breathing sound :right side crackle

■ Abdomen : Bilateral flank tenderness(+) with **palpable mass**

■ Extremities : **Ash-leaflet hypopigmented spots** on right arm

■ Skin :**Angiofibromata (Sebaceum adenomas)** on face, head, back

# Laboratory Data

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- Iron : (75-198 ug/dl) 18
- RBC (4.2-6.1×10e3/ul) 2.21 ,2.57 ,2.30
- HGB (12-18 g/dl) 4.7, 7.2, 5.3
- HCT(37-52%) 17.2 ,22.9, 18.4
- NEU (40-74%) 88.0

# Impression

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- Anemia ,R/O UGI bleeding
- Pneumonia
- Tuberosus sclerosis

# Plan

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- Check iron profile
- Check reticulocyte count
- NPO with NG decompression
- Correct dehydration note I/O
- Arrange Abd CT
- Blood transfusion
- Blood culture ,sputum culture and smear

# Chest and KUB

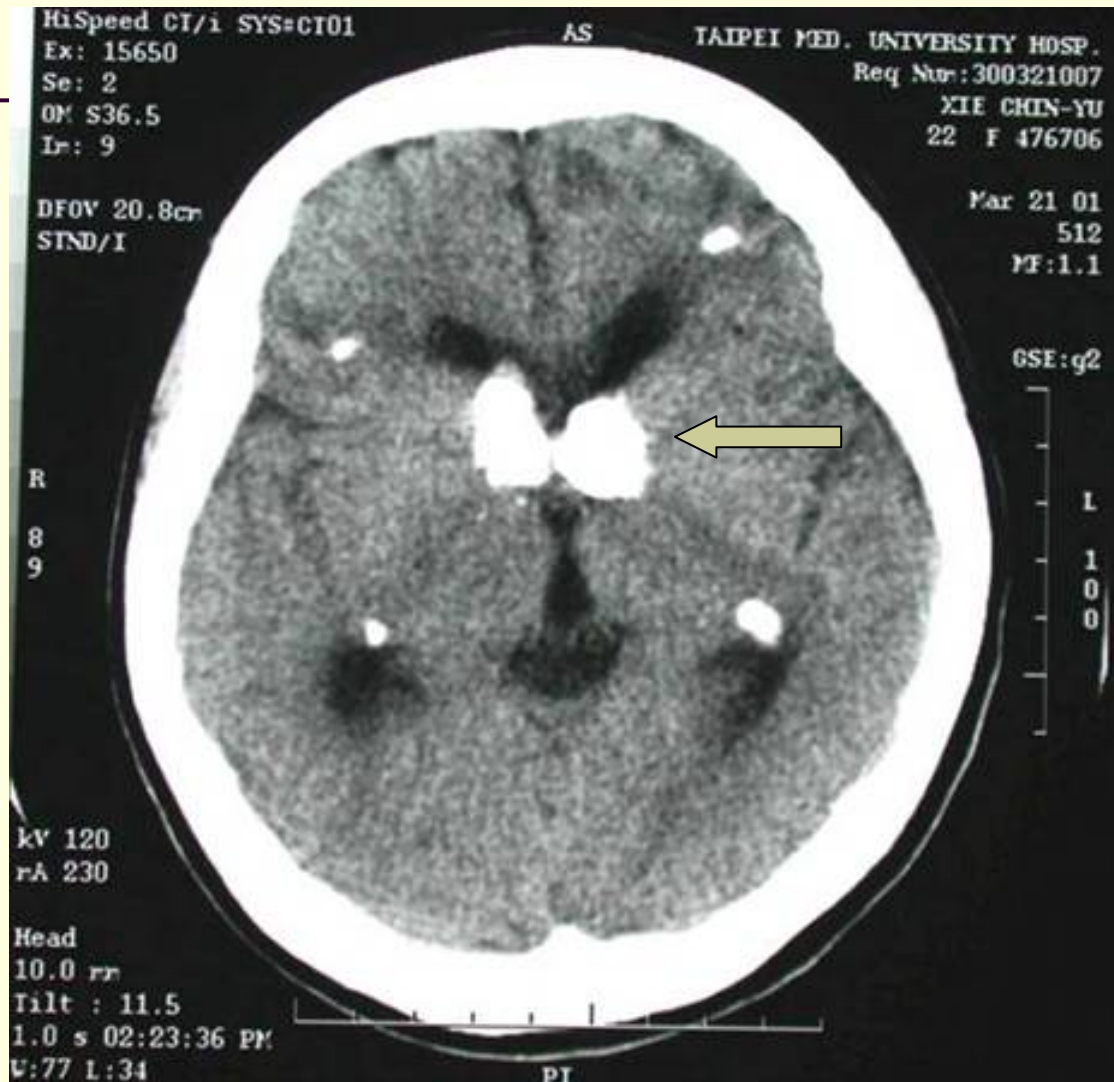


**CXR: Infiltration : pneumonia patch**

**Water bottle shape : pericardial effusion**

**KUB: Increased soft tissue density at bil. renal regions.**

# Brain CT

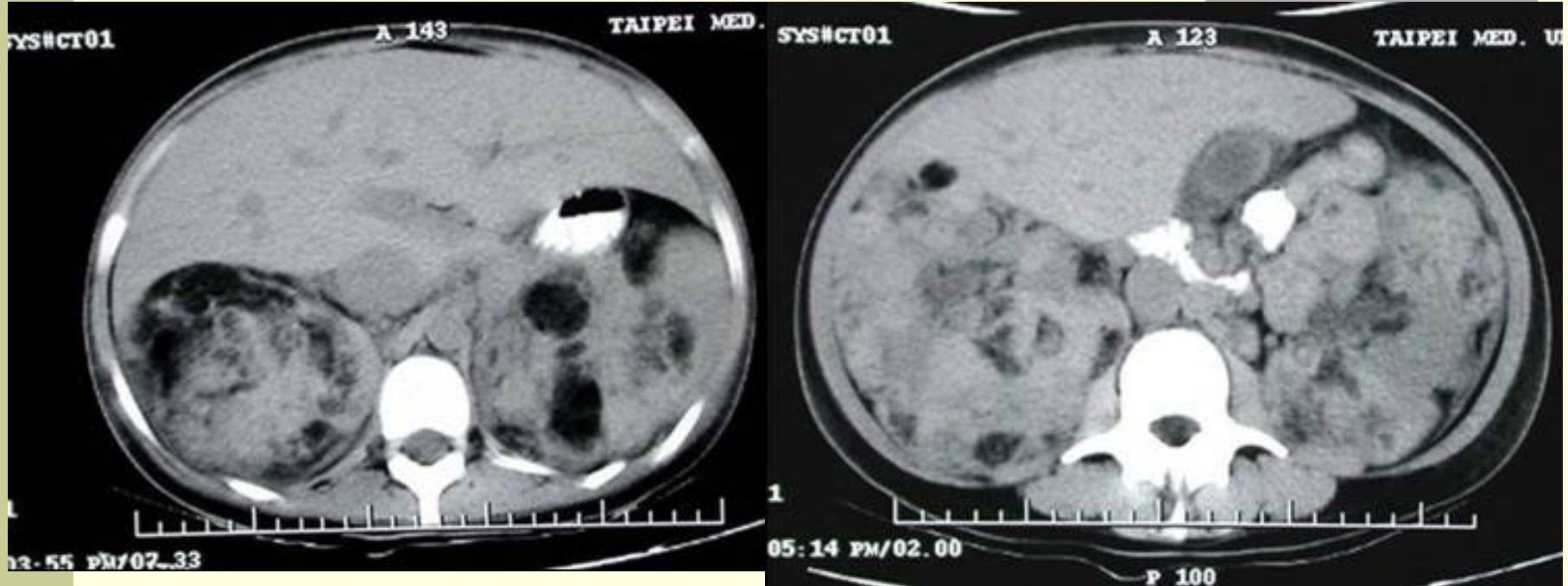


**Calcification of bilateral caudate nuclei**



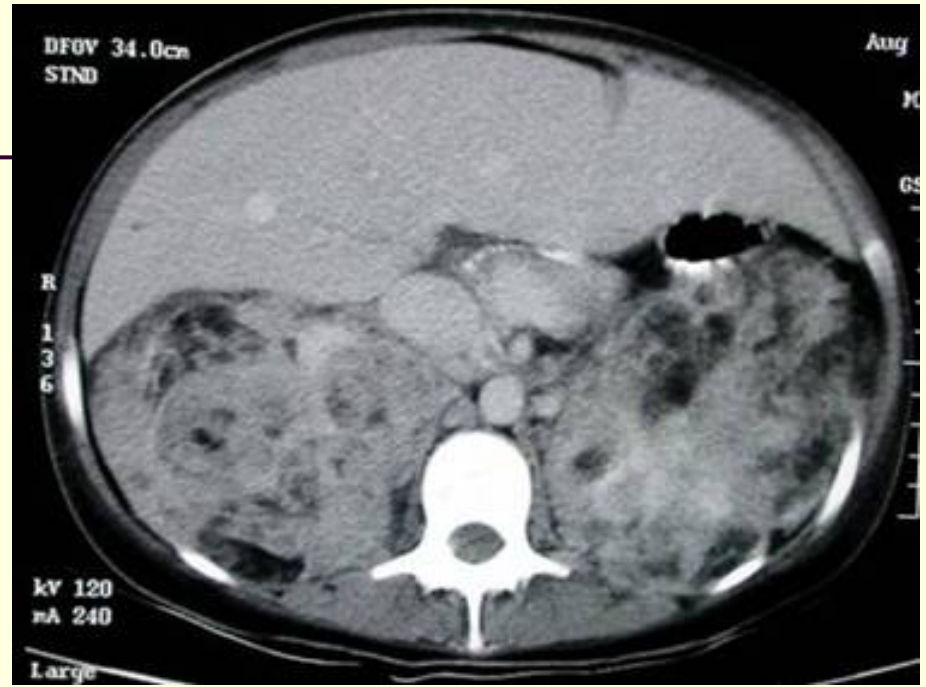
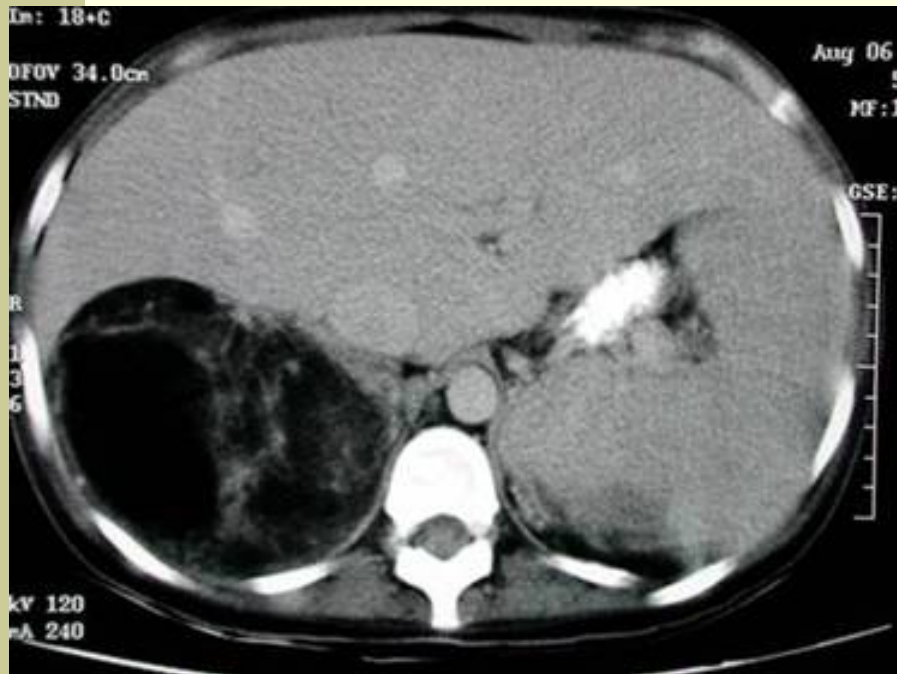
**Multiple Periventricular calcification**

# Abdomen CT scan



- **Pre-contrast study**
- **Huge mixed-density masses at both renal regions, with superior displacement of the liver and spleen**



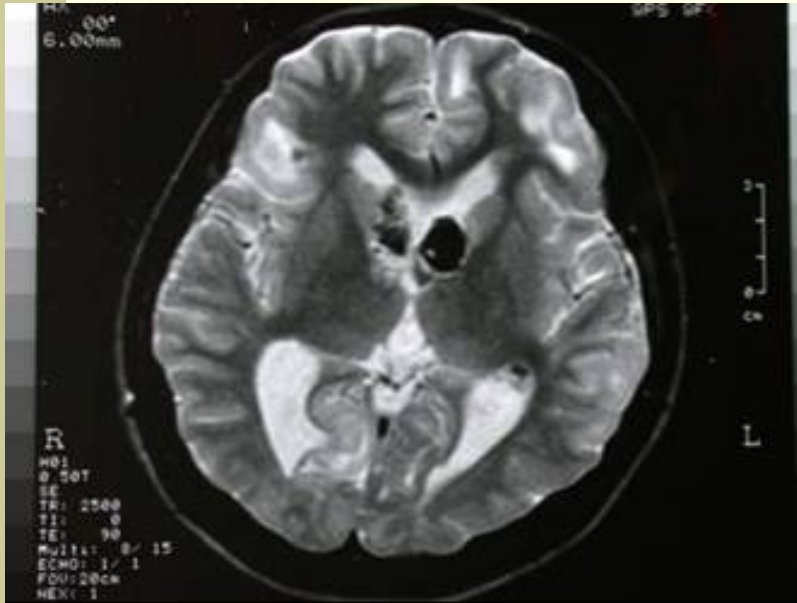


**Post-contrast study.**

**Heterogenous contrast enhancement of renal masses**

**Marked fatty components noted.**

# MRI of brain



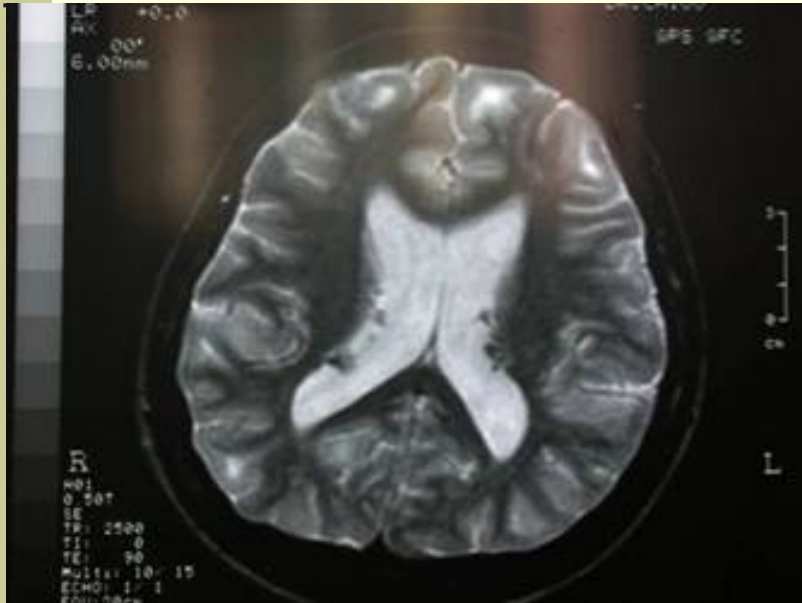
**T2WI**



**T1WI**

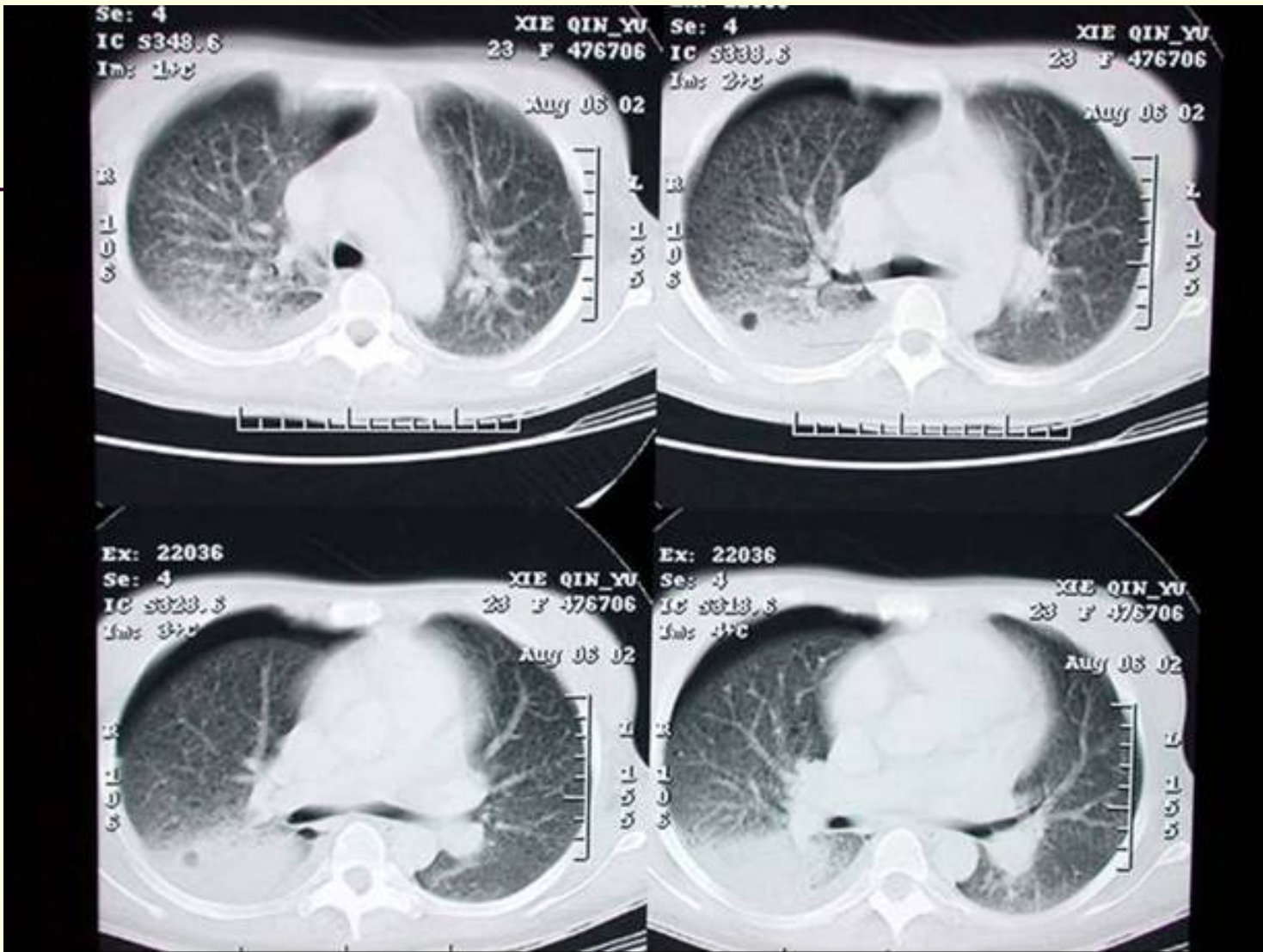


**T1WI with contrast**



Multiple subependymal nodules with candle dripples appearance at lining of lateral ventricles





**Pneumothorax.**

**Consolidation on right lower lobe**

# Imaging finding-1

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- Brain CT – calcification of bilateral caudate nuclei and multiple periventricular calcifications.
- MRI of brain – Low signal to iso-signal masses at periventricular region on T1WI and T2WI, with contrast enhancement, consistent with subependymal hamartomas.

# Imaging finding-2

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- Abdomen CT – Bilateral fatty renal masses, angiomyolipomas are most likely.
- Lung – Pneumothorax

# Differentiation of periventricular calcifications.

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- 1. Tuberosus sclerosis.
- 2. Congenital infection:
- 3. CMV
- 4. Toxoplasmosis

# Differential diagnosis of bilateral renal masses-1

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- 1. Malignant tumor
  - - Malignant lymphoma/Hodgkin disease
  - - Metastases
  - - Renal cell carcinoma
  - - Wilms tumor



# Differential diagnosis of bilateral renal masses-2

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- 2. Benign tumor
  - - Angiomyolipoma
  - - Nephroblastomosis
- 3. Cysts
  - - Polycystic kidney disease
  - (adult or acquired)

# Diagnosis:

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- TUBEROUS SCLEROSIS

# Tuberous sclerosis

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- Neuroectodermal disorder: genetic alternation of ectodermal and mesodermal cells with hyperplasia, with a disturbance in cellular differentiation
- Clinical triad:
  - 1. Skin manifestations (96%)
  - 2. Seizures (86%)
  - 3. Mental retardation (49%)

# Etiology

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- 1/3 of cases are inherited as an autosomal dominant trait, others sporadic mutations
- TSC1 on chromosome 9q34 – hamartin
- TSC2 on chromosome 16q13.3 – tuberin
- Majority relate to TSC2

# Clinical manifestation-CNS

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- 1. Subependymal hamartomas
- 2. Giant cell astrocytoma
- 3. Tubers (cortical/subcortical hamartomas)
- 4. Heterotopic gray matter islands in white matter

# Clinical manifestation-Skin

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- **Hypomelanotic macules:**

usually more than 3, 3 to 4 cm  
lance ovate or ash-leaflet spots

- **Confetti macules:**

multiple, discrete, small (1~2 mm)  
hypopigmented macules. These lesions are pathognomonic.

# Clinical manifestation-Skin

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- **Angiofibroma:**

occur in the center of face. They are confirm and disseminated but may coalescence.

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- Ocular – Phakoma (whitish disk-shaped retinal hamartoma =astrocyte proliferation in/near optic disc)

Small calcification in region of optic nerve head

Optic nerve glioma



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- **Renal – Angiomyolipoma** (38%) multiple + bilateral risk of spontaneous hemorrhage

Multiple cysts of varying size in cortex + medulla mimicking adult polycystic kidney disease

Renal cell carcinoma(3%)bilateral

40%

## ■ Lung

- – interstitial fibrosis in lower lung fields and miliary nodules pattern (lymphangiomyomatosis)
- - Cystic change of lung parenchyma
- - Spontaneous pneumothorax (50 %)
- - chylothorax
- - cor pulmonale

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## ■ Heart

- - Congenital cardiomyopathy
- - Rhabdomyoma (5%)
- - Aortic aneurysm

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- **Other visceral involvement:**

- 1. Adenomas + lipomyomas of liver
- 2. Adenomas of pancreas
- 3. Tumors of spleen.

# Treatment

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- Symptomatic – anticonvulsion therapy for control of seizures

# Reference

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- Diagnostic radiology, Grainger and Allison's ,4th edition
- Adams and Victor's Neurology, 7th edition
- Color atlas and synopsis of clinical dermatology, 4th edition