General History

- Name : 謝X瑜
- Sex : female
- Birth Date : 1979 / 02 /10
- Chart Number : 04767060

Chief Complain

- Epigastric pain with bloody vomitus for 1 day

Present Illness

- 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 regualar F/U at our OPD.Besides,she has IDA under Ferrum Hausman tablet supplement.

- 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 black stool passage in recent one week.Besides, cought with yellowish sputum,fever,dyspnea are noted in recent 3 days.She denied dysuria,headache,chills, nor constipation.

- 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 is admitted.

Family history

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

Personal history

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

Past history

- DM : denied
- HTN :denied
- Renal tumor Angiomyolipoma) :1998/6 ;1998/7
- Neurocutaneous syndrome : 1998/7
- EPS,seizure :1998/12
- Esophageal ulcer :2001/4

Physical examination

- BP : 130/70 mmHg ,TPR : 37.7,104,22
- Conjunitiva :**pale**
- Chest : breathing sound :right side crackle
- Abdomen : Bilateral flank tenderness(+) with **palpable mass**
- Extremities :**Café-au lait spot** on right arm
- Skin :**Sebaceum adenomas** on face,head,back

Laboratory Data

- 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

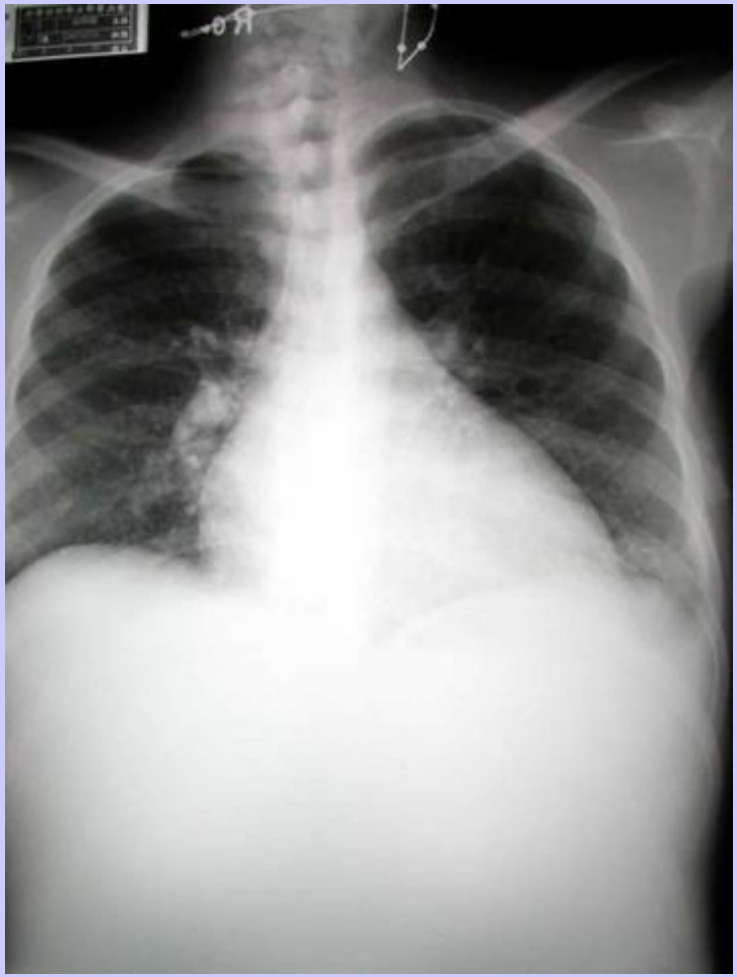
Impression

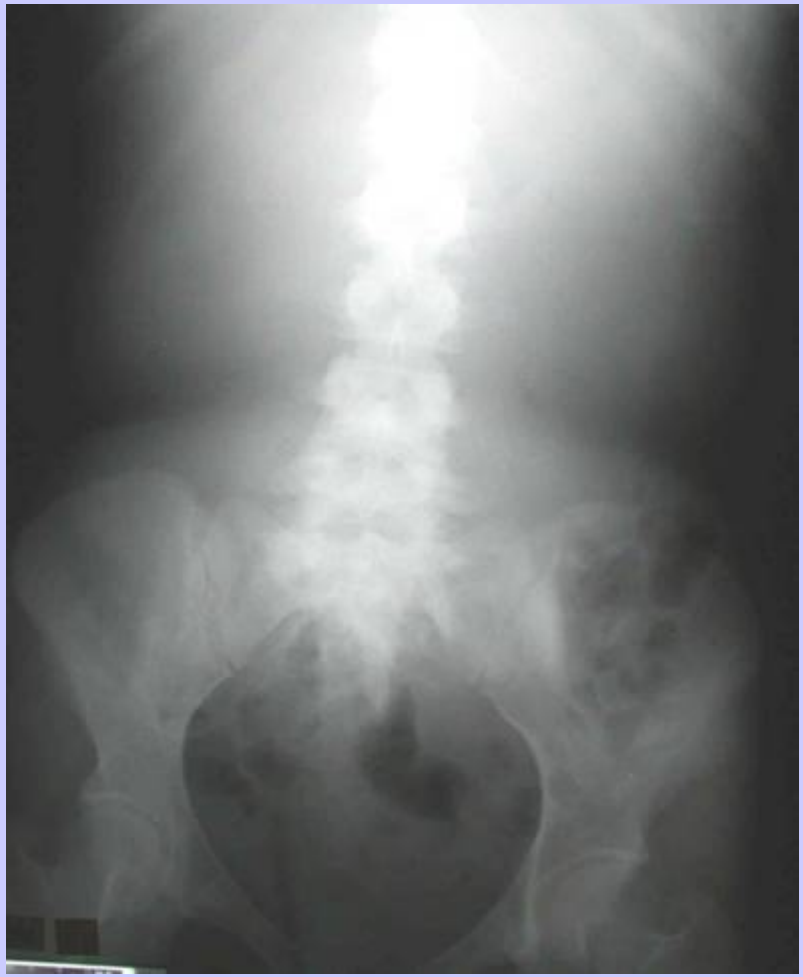
- Anemia ,R/O UGI bleeding
- Pneumonia
- Tuberous sclerosis

Plan

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

Chest and KUB



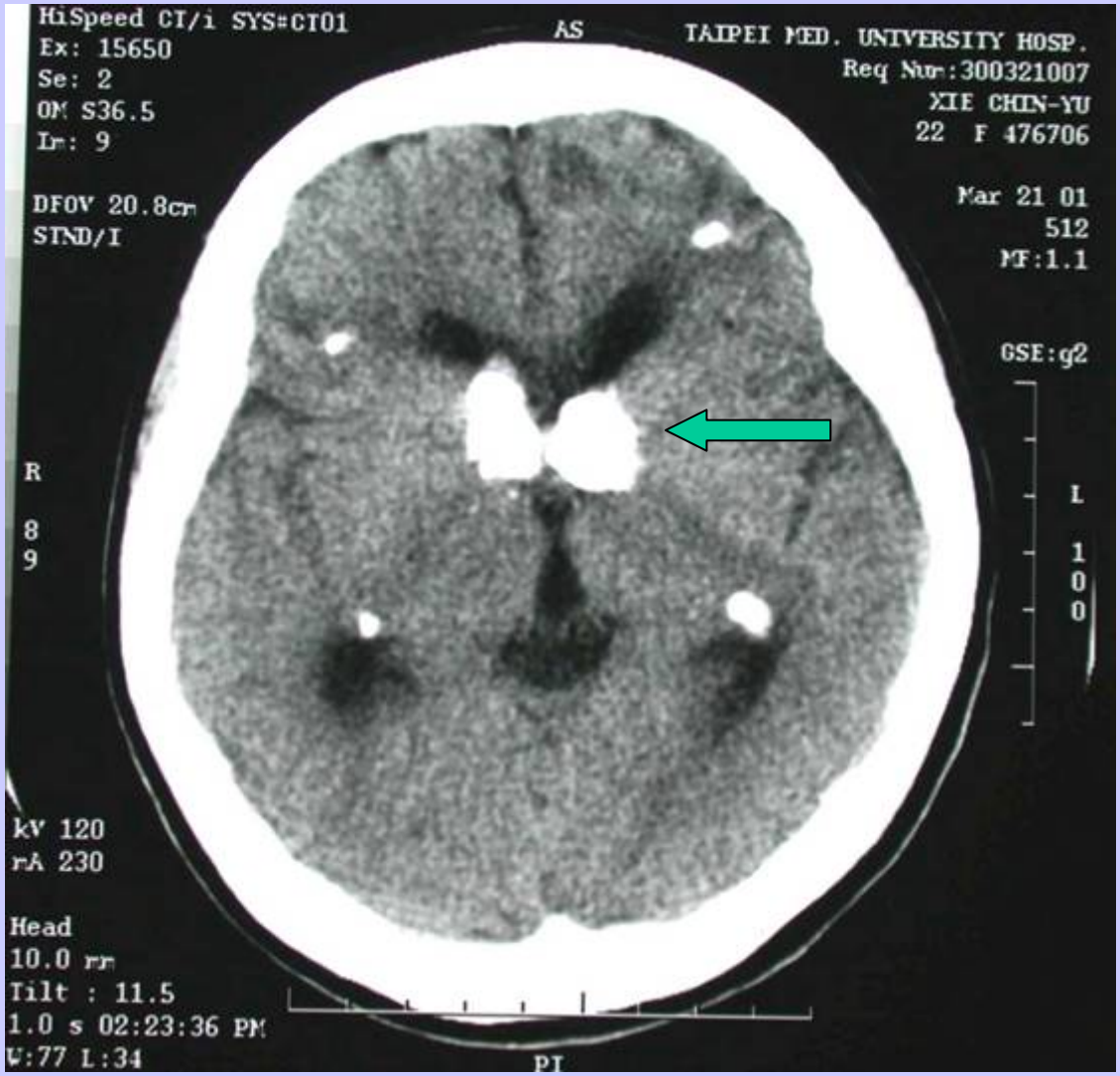


CXR: Infiltration : pneumonia or hemorrhage

Water bottle shape : pericardial effusion

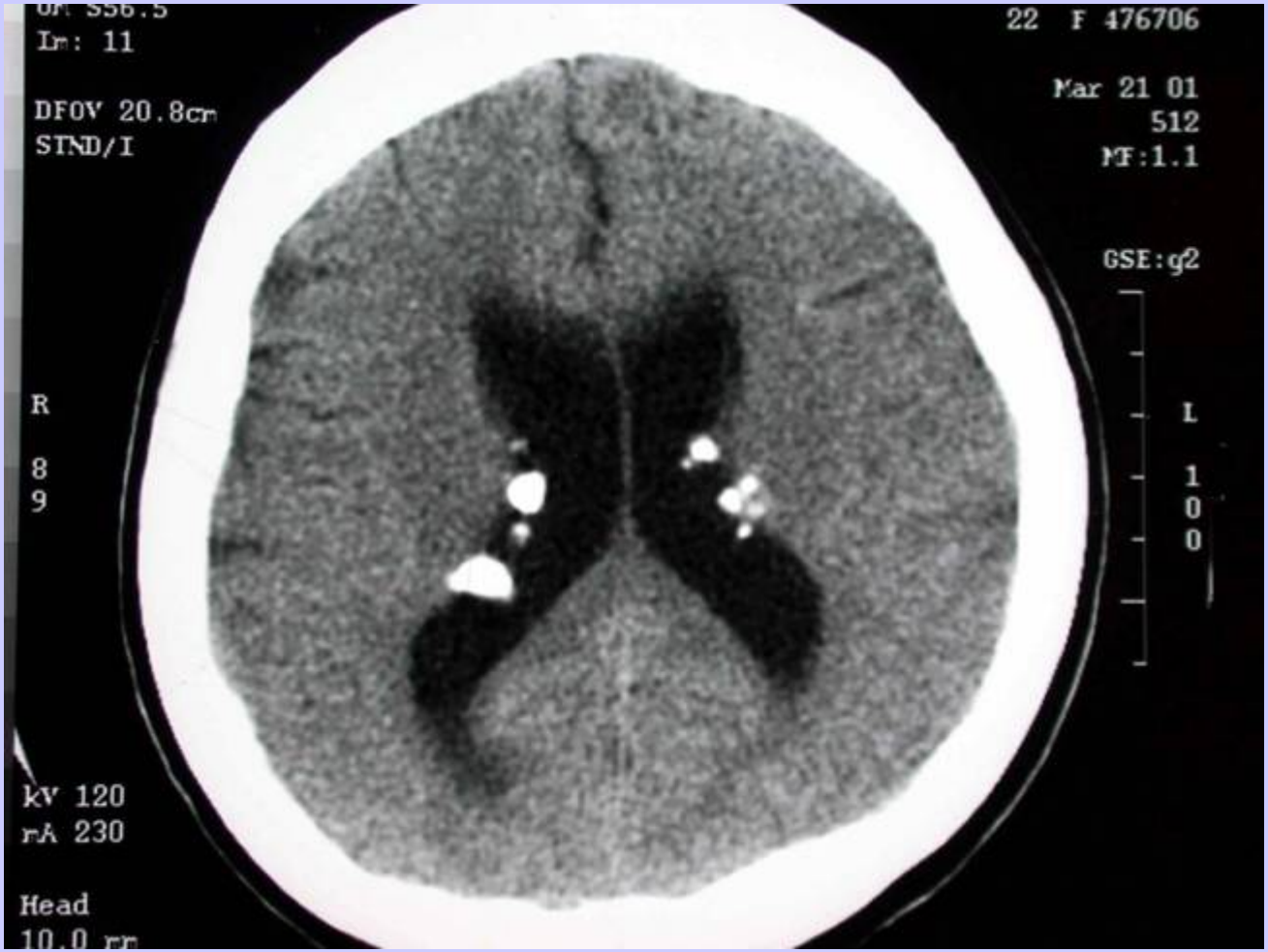
KUB: Increased soft tissue density at bil. renal regions.

Brain CT (Pre-contrast)



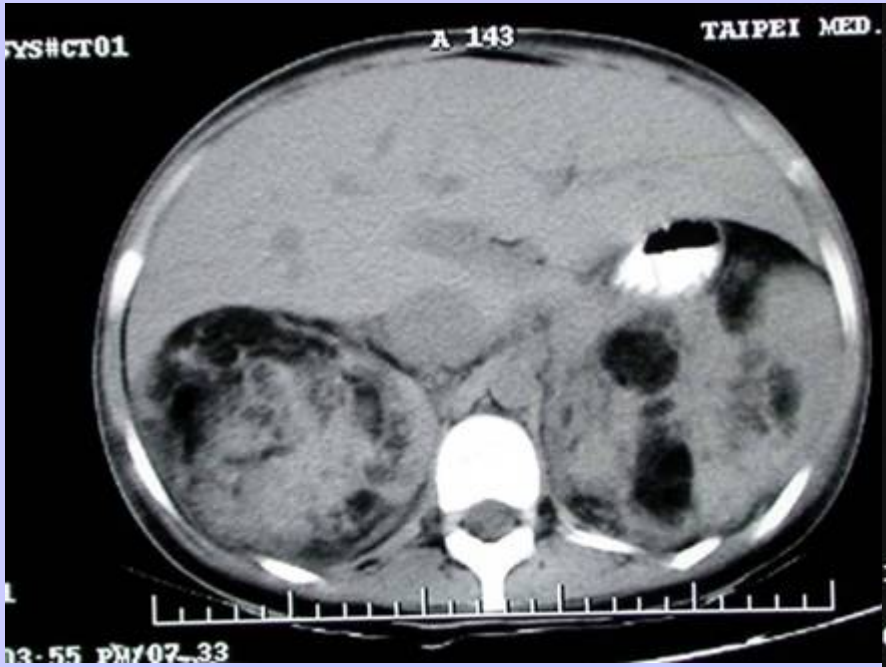
Calcification of bilateral caudate nuclei

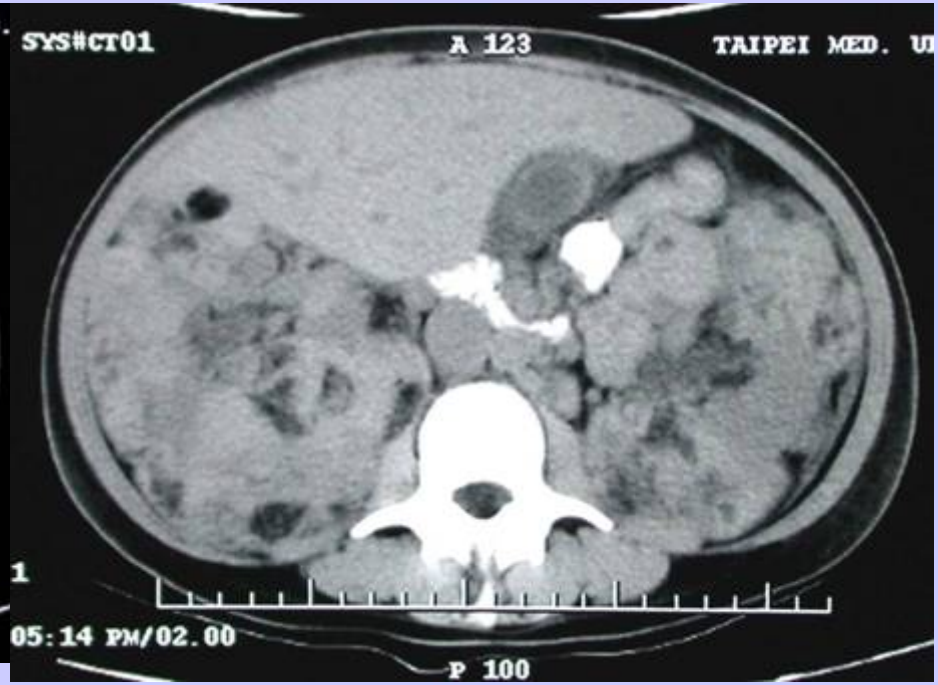
Brain CT (Pre-contrast)



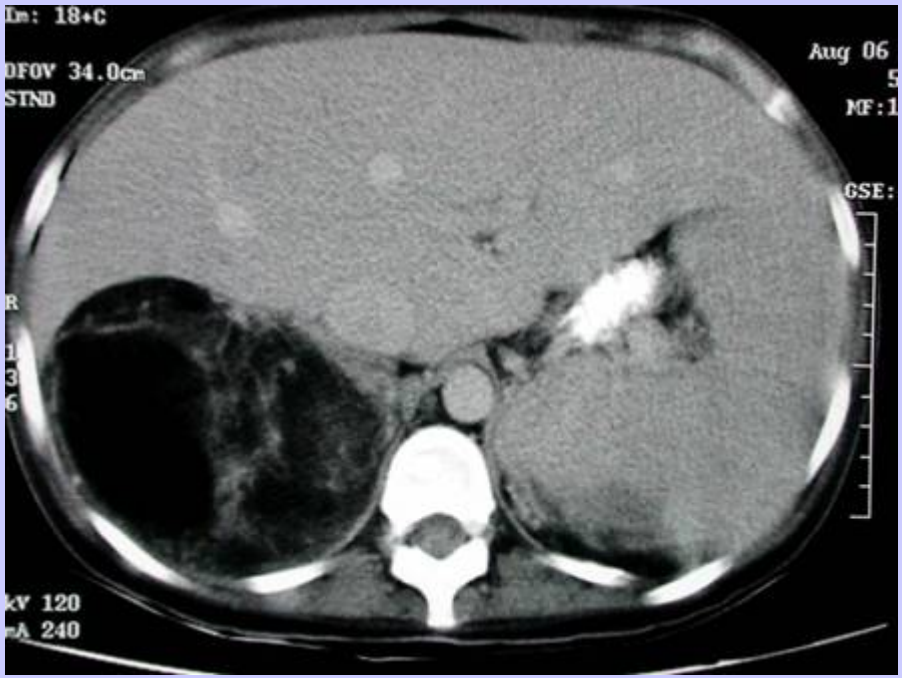
Multiple Periventricular calcification

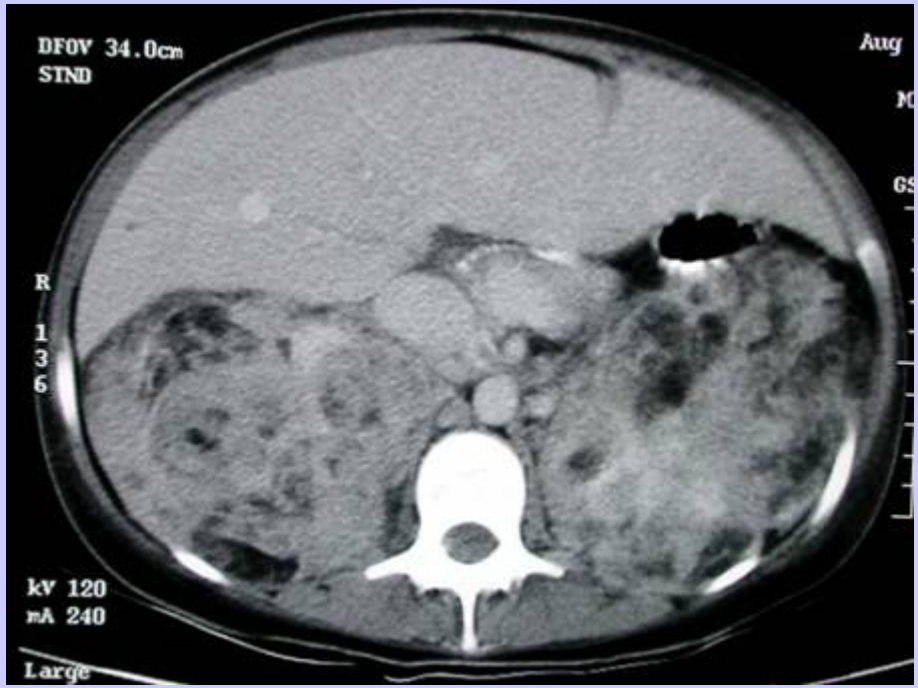
Abdomen CT scan





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



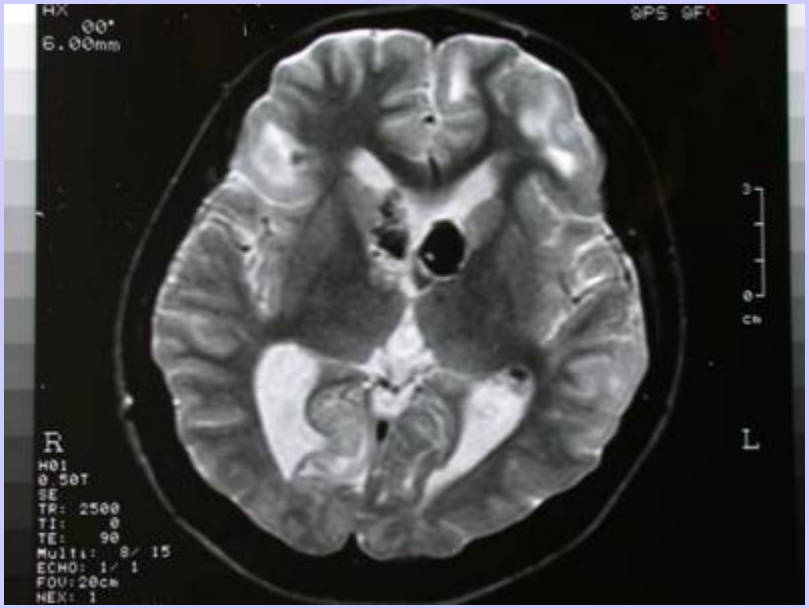


Post-contrast study.

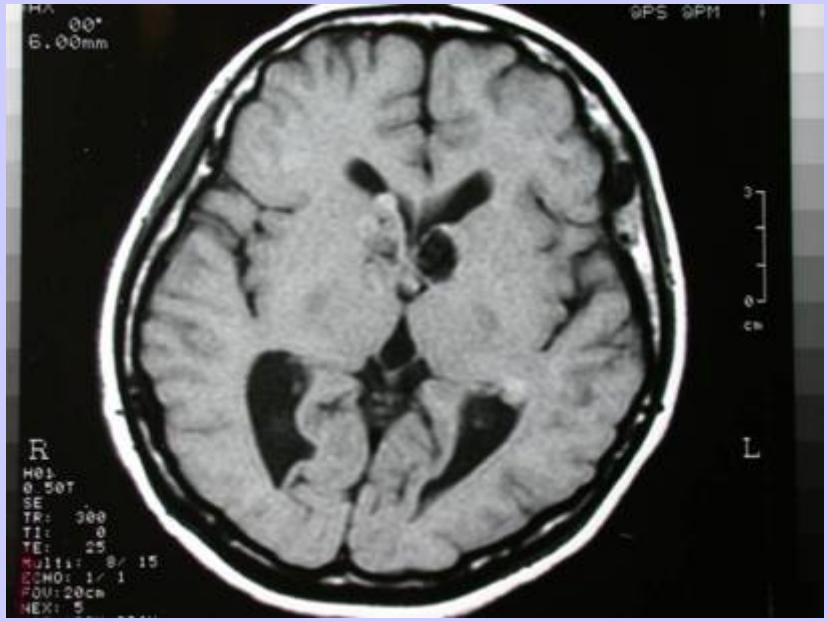
Heterogenous contrast enhancement of renal masses

Marked fatty components noted.

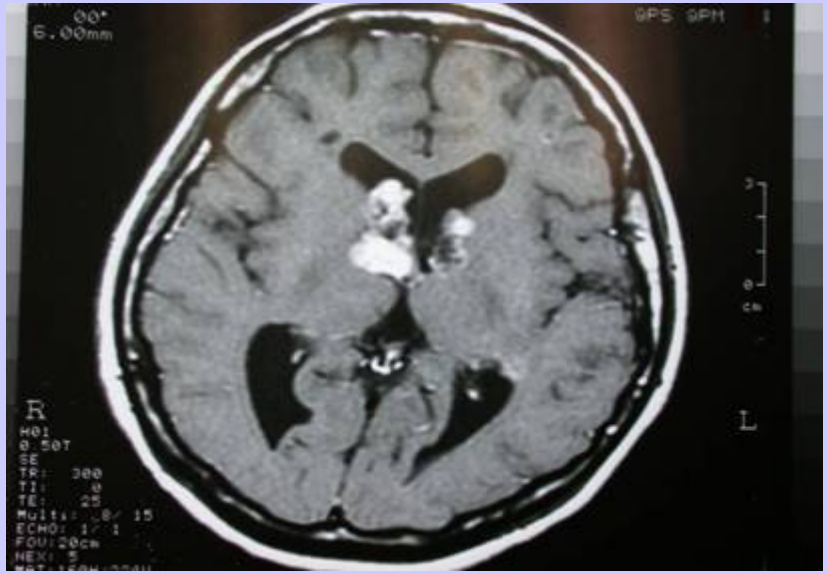
MRI of brain



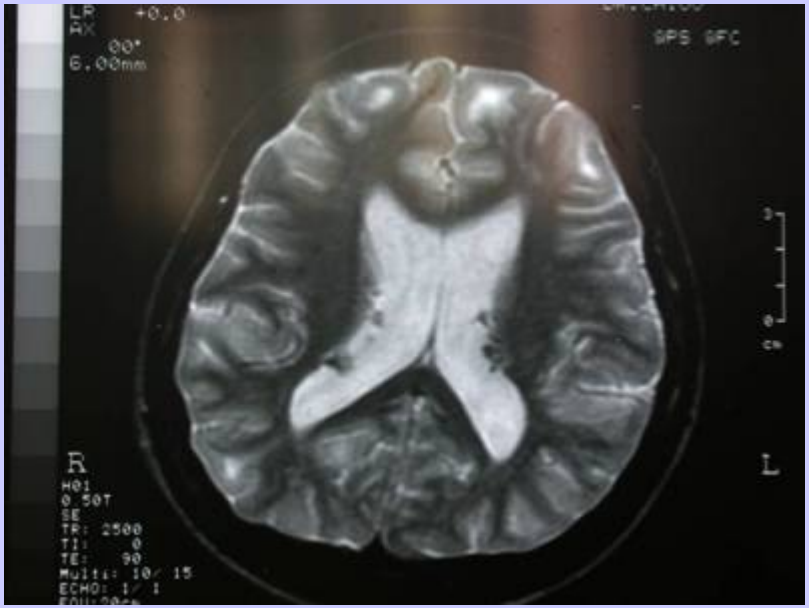
T2WI

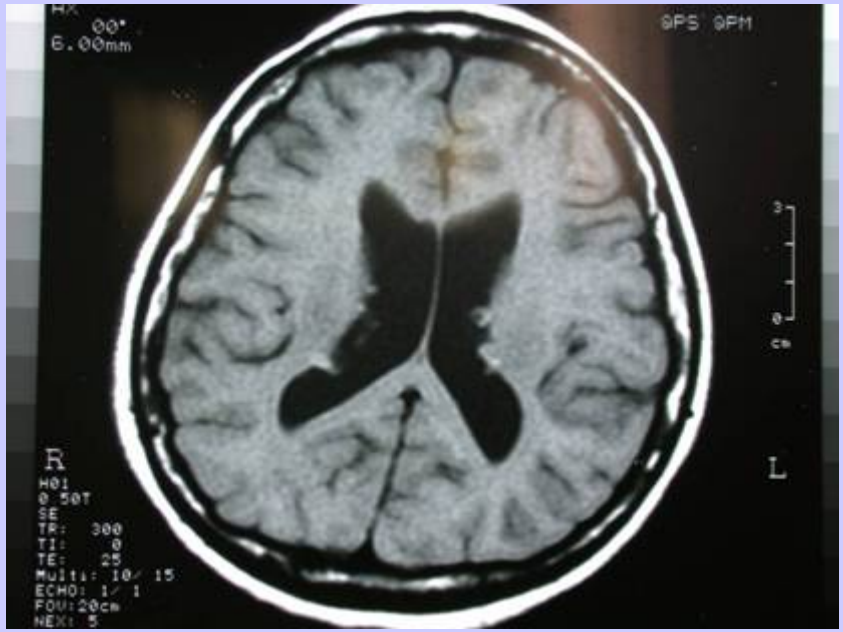


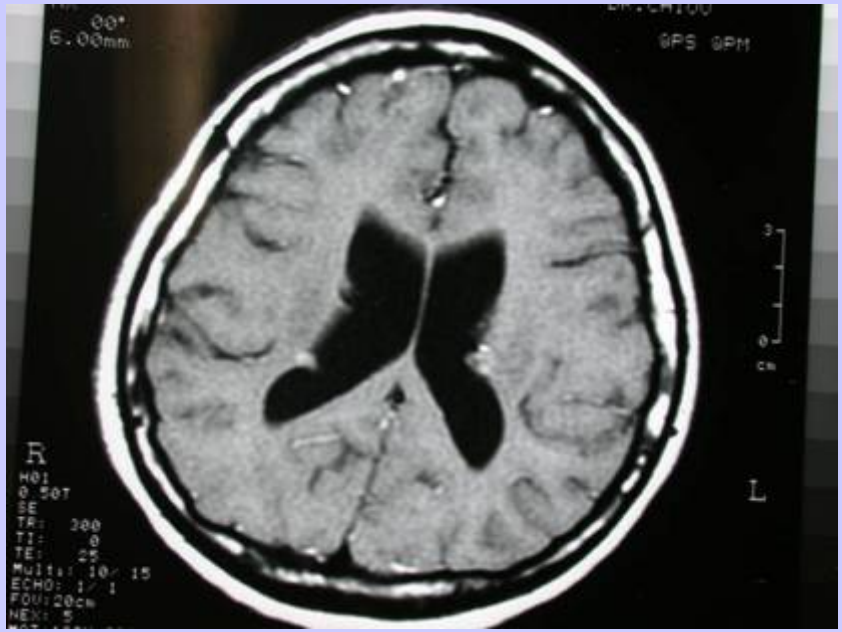
T1WI



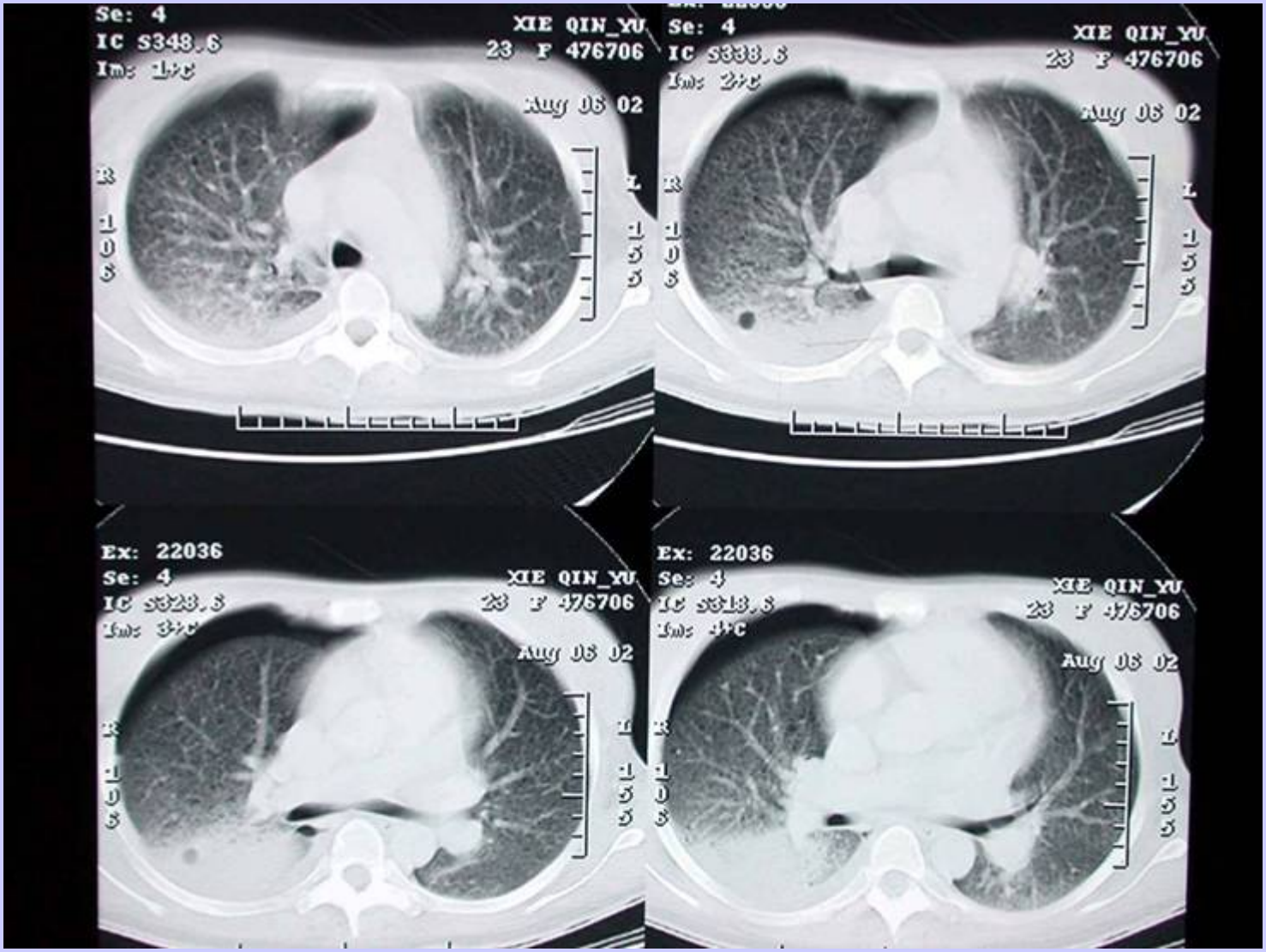
T1WI with contrast







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



Pneumothorax.

Consolidation on right lower lobe

Imaging finding

- Brain CT – calcification of bilateral caudate nuclei and multiple periventricular calcifications.
- MRI of brain – Low signal to isointense masses at periventricular region on T1WI and T2WI, with contrast enhancement, consistent with subenpendymal hamartomas.
- Abdomen CT – Bilateral fatty renal masses, angiomyolipomas are most likely.
- Lung – Pneumothorax.

Differentiation of periventricular calcifications.

- 1. Tuberous sclerosis.
- 2. Congenital infection:
- CMV
- Toxoplasmosis

Differential diagnosis of bilateral renal masses

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

Differential diagnosis of bilateral renal masses

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

Diagnosis:

- TUBEROUS SCLEROSIS

Tuberous sclerosis

- Neuroectodermal disorder
- Clinical triad:
- 1. Adenoma sebaceum (30%)
- 2. Seizures (80%)
- 3. Mental retardation (70%)

Etiology

- 1/3 of cases are inherited as an autosomal dominent trait ， others sporatic mutations
- TSC1 on chromosome9q34 – hamartin
- TSC2 on chromosome16q13.3 – tuberin
- Majority relate to TSC2

Clinical manifestation

- CNS – subependymal hamartomas
  Giant cell astrocytoma
  Tubers(cortical/subcortical hamartomas)
  Heterotopic gray matter islands in white matter

- Skin – **Adenoma sebaceum(wartlike nodules** of brownish red color average 4 mm in size with bimalar distribution

  Shagreen rough skin patches(80%)

  Ash leaf patches

  Ungual fibromas

  Café-au lait spots

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

  Small calcification in region of optic nerve head

  Optic nerve glioma

- 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**
- **Heart**
- **- Congenital cardiomyopathy**
- **- Rhabdomyloma (5%)**
- **- Aortic aneurysm**

- **Other visceral involvement**:
- 1. Adenomas + lipomyomas of liver
- 2. Adenomas of pancreas
- 3. Tumors of spleen.

Treatment

- Symptomatic – anticonvulsion therapy for control of seizures