Basic data

- 出生日期：mk 59/8/24
- 年齡：32 y/o
- 婚姻：married

Past history

- 5th proximal phalanx pathologic fracture due to enchondroma , at mk 90
- DM：denied
- HTN：denied
- Asthma：denied
- Denied any other systemic disease
- NKDA

Personal history

- Drinking：denied
- Smoking：denied
- Betel nut：denied

Family history

- 祖母為荷蘭人，有1/4荷蘭血統
- denied any other hereditary disease

Clinical check-up

- Brain tumor , F/U with brain MRI

Present illness

- This 32 year old male was a victim of brain tumor noted for a long time . According to his statement , he was quite healthy before . However , he was found to have brain tumor during health examination in America several years ago . He was suggested not to do surgery and regular F/U .

P.E

- Cons：clear
- HEENT：grossly normal , no JVE , horizontal nystagmus noted
- Chest：symmetric expansion , clear breathing sound , no rales
- Heart：RHB , no murmur
- Abdomen：soft & flat , no mass , no tenderness
- Ext：freely mobile

Lab data (89/4/25)

- CBC/DC

WBC 6.22	RBC 5.15	Plt 317
HGB 14.2	Hct 45.7	MCV 88.8
MCH 27.6	MCHC 31.1	RDW 12.4
neu 39.9	lym 45.1	mono 5.4
eos 6.1	baso 0.9	

Lab data (89/4/25)

- Biochemistry

albumin 4.2 total protein 6.7

BUN 13.4 creatinine 0.8

uric acid 6.9 cholesterol 212

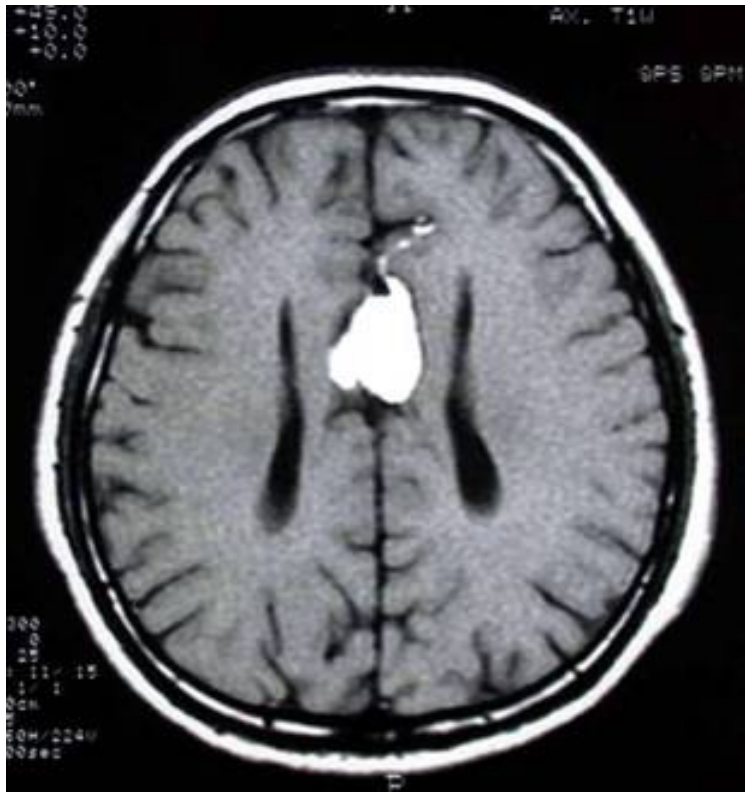
TG 167 GOT 16 GPT 23

ALK-p 72 r-GT 21 Bil-D 0.1

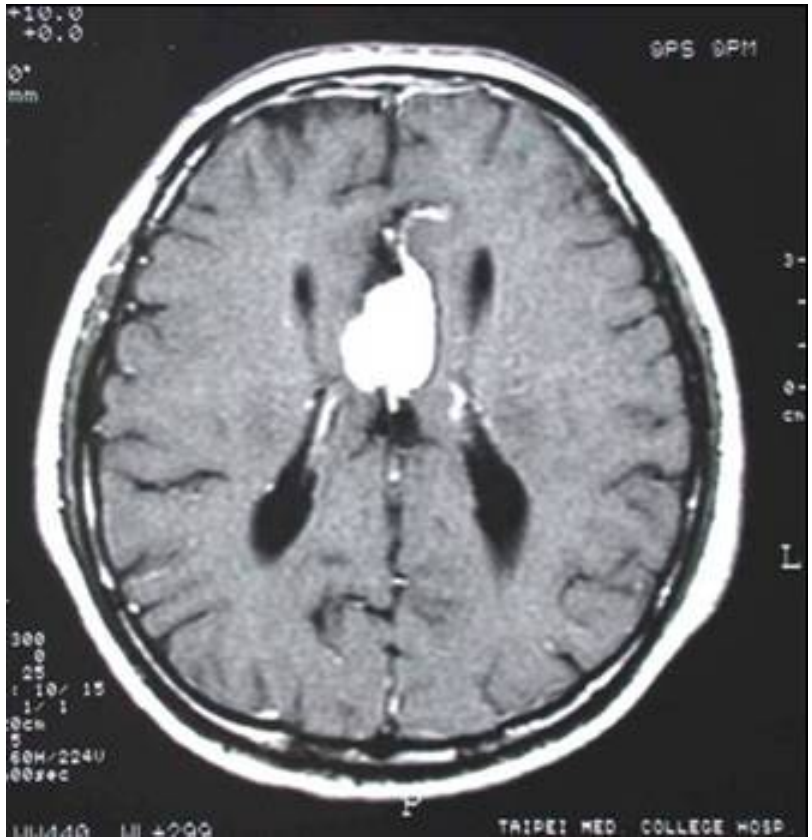
Bil-T 0.2 Na 139 K 4.0

Cl 99 Ca 8.4 glucose AC 92

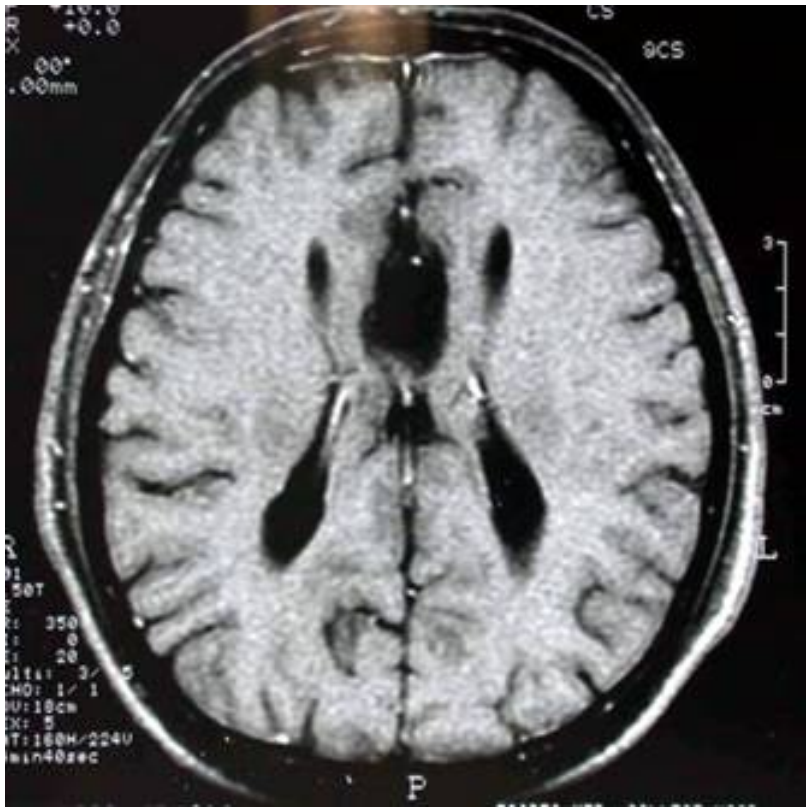
Brain MRI : T1WI , no C



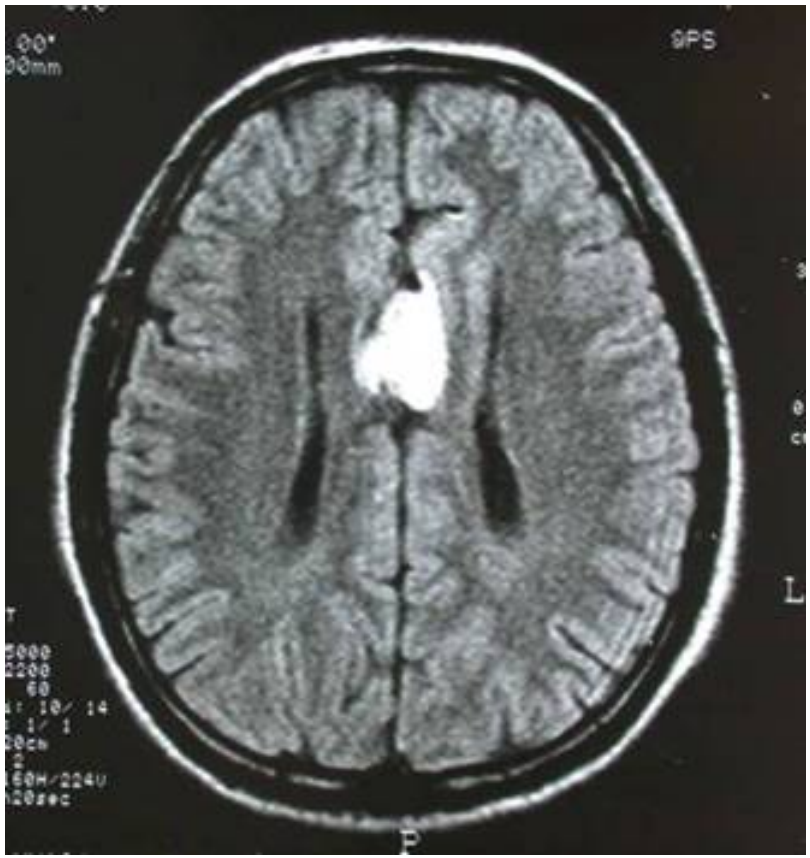
T1WI , with C



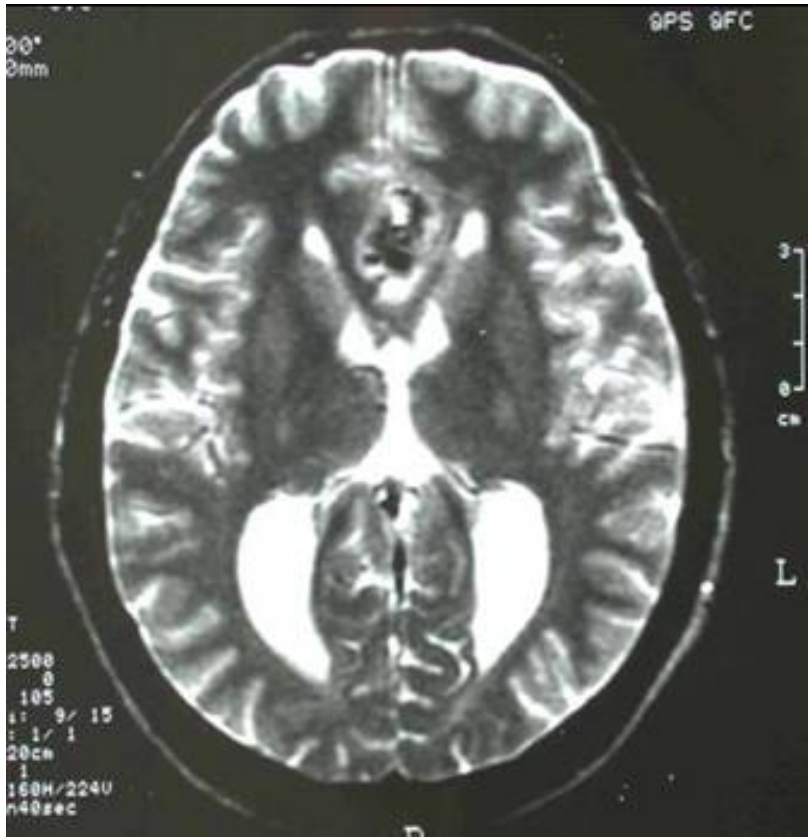
T1WI , fat suppression



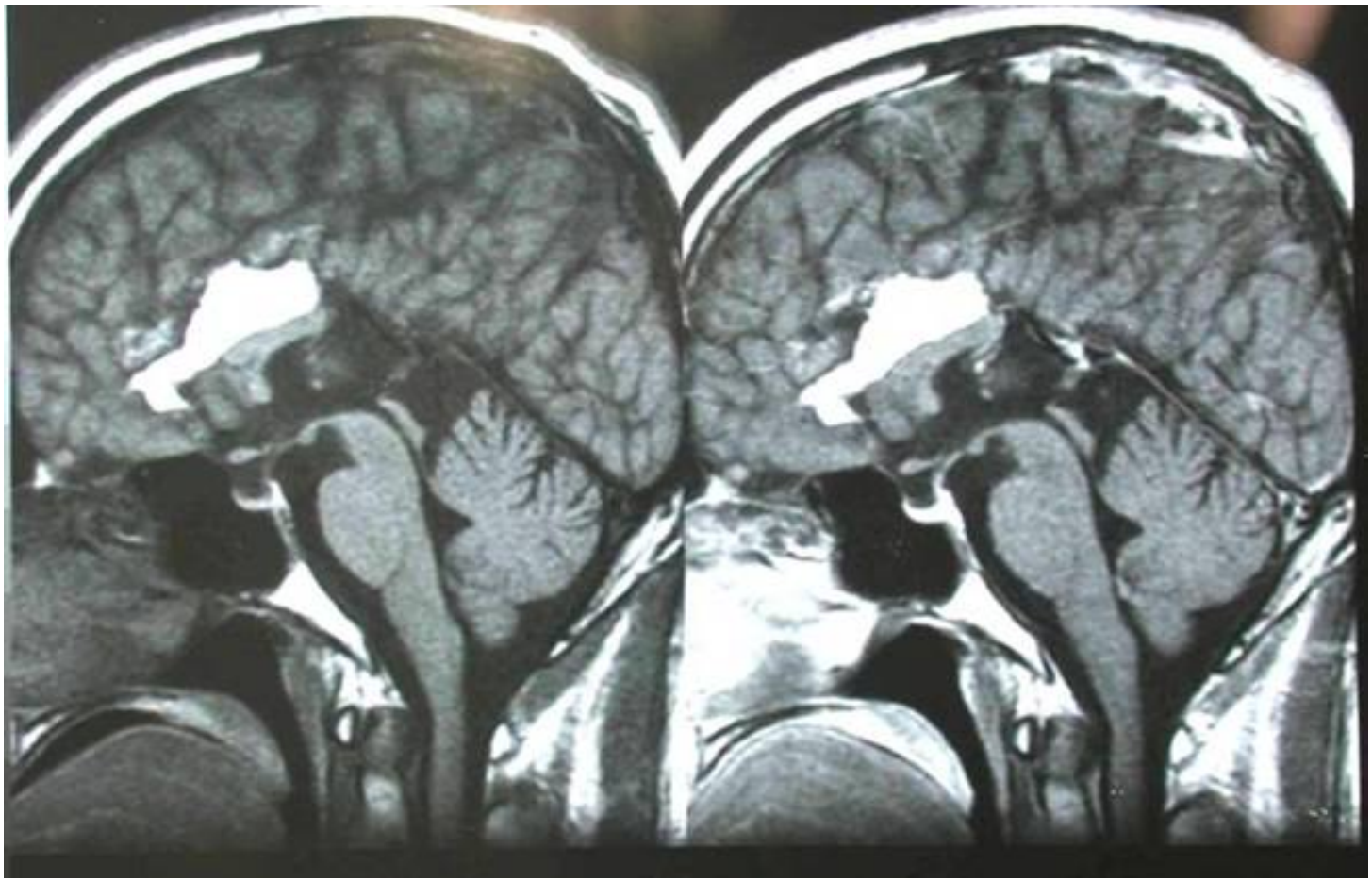
FLAIR



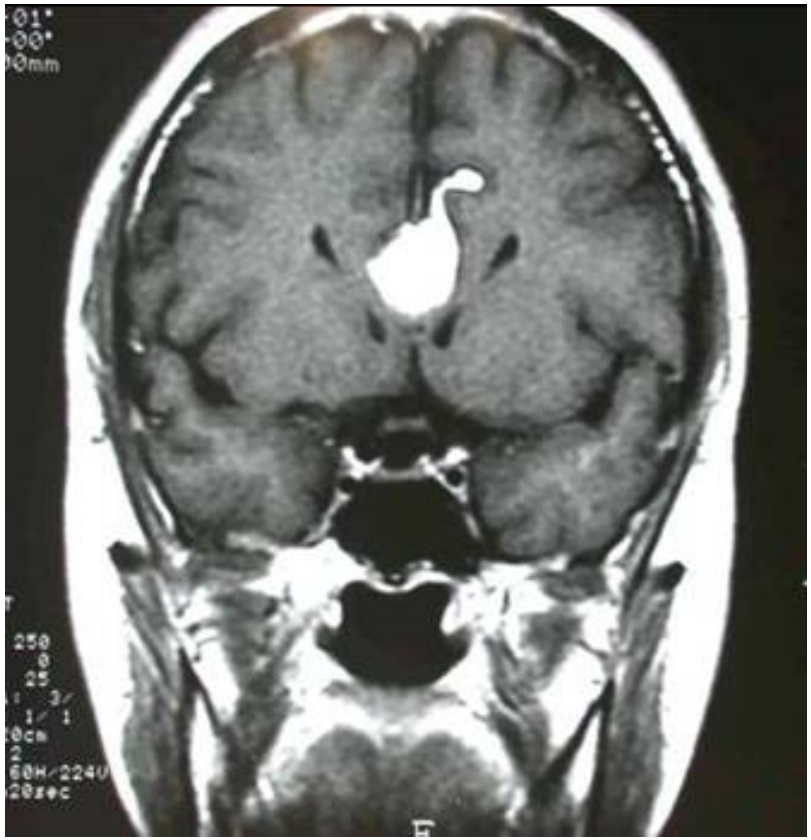
STIR



T1WI , sagittal



T1WI , coronal



Image

- An irregular high SI mass at the genu of corpus callosum on T1wI , FLAIR image and low SI on T1wI(with fat suppression) , STIR .
- Mild hydrocephalus
- Parallel configuration of lat. ventricles
- No midline structures deviation

Differential diagnosis

- Agenesis of the corpus callosum and with lipoma

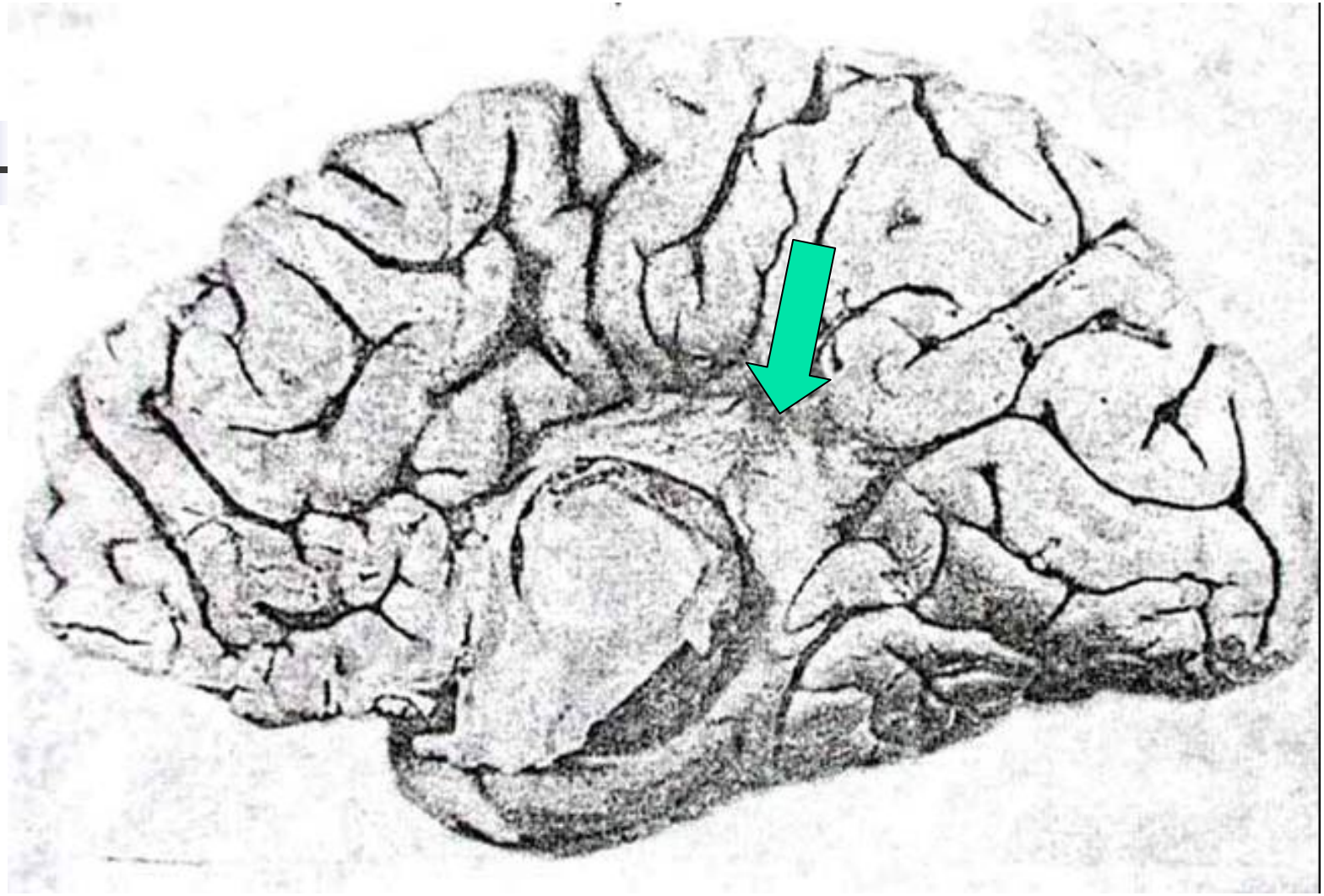
Agenesis of Corpus Callosum

Agenesis of corpus callosum (ACC)

- may form part of a more extensive malformation complex , such as holoprosencephaly , or the callosum may be totally or partially absent or hypoplastic in an otherwise normal brain .

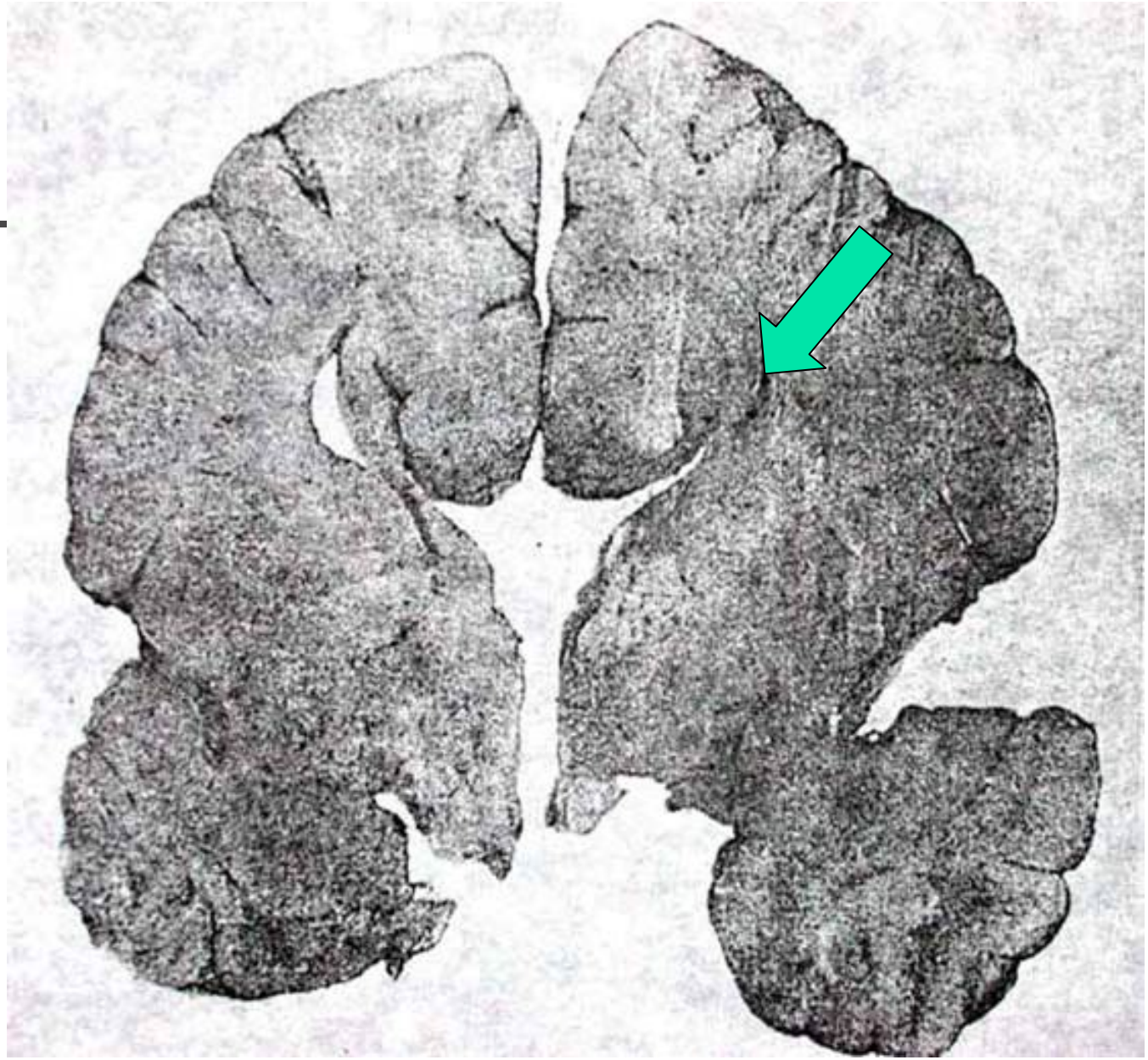
Anatomy of ACC(1)

- In partial agenesis , the posterior portion is usually missing , while rostrum and genu remain
- An abnormal gyral pattern , no cingulate gyrus and the gyri have a radiating pattern extending perpendicularly to the roof of the third ventricle
- On coronal section , no structure separate the lateral ventricles in the midline



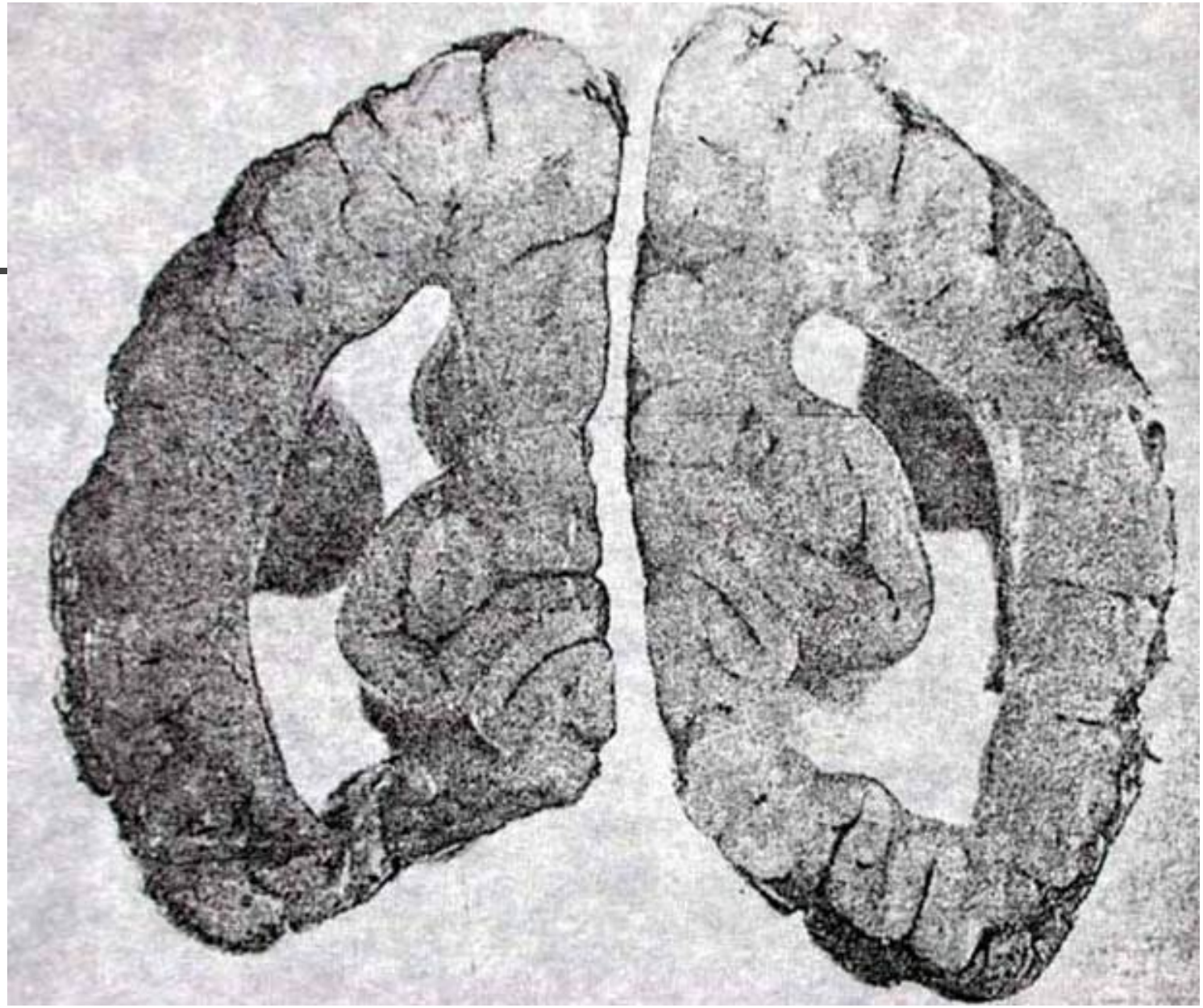
Anatomy of ACC(2)

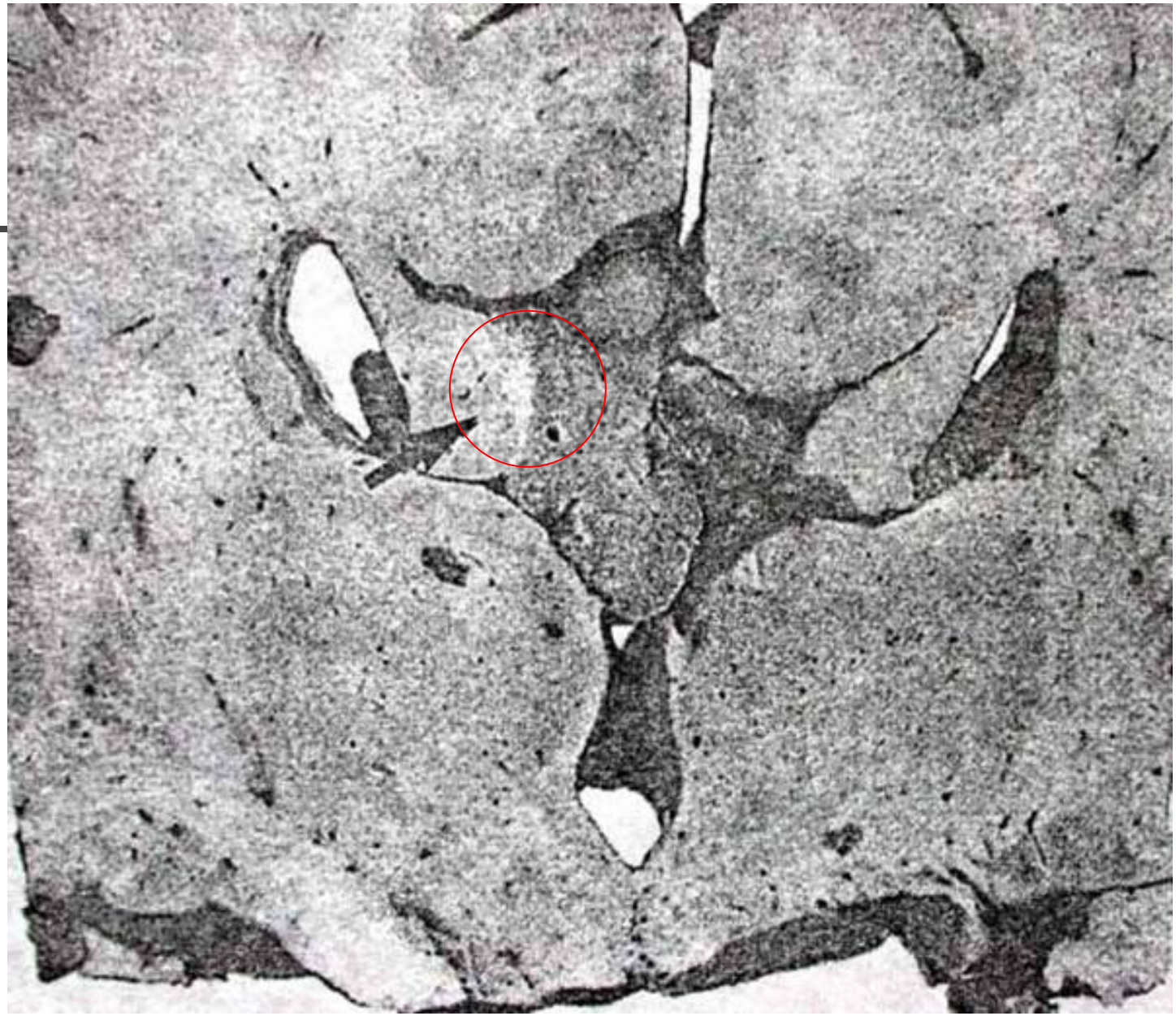
- Lateral vedntricles have a membranous roof and upturned pointed corners , like Bat's wing.
- The membranous roof of third ventricle is often distended and bulges into the interhemispheric fissure , displacing the fornices laterally .
- A prominent bundle of fibers , Probst bundle , is situated usually in the lateral part near the apex roof of lat. ventricle.



Anatomy of ACC(3)

- The septum pellucidum may be absent
- The occipital horn or lat. ventricle often markedly dilated .
- Posterior commisure is always present , but the anterior commissure is variable .
- Local callosal defect is associated with a midline mass → rarely meningioma , cyst , hamartoma ; more often lipoma





Pathology

- Interhemispheric lipoma and callosal defects may be closely contiguous , a dorsal lipoma overlying a hypoplastic callosum , wrapping round it or associating with partial agenesis , and both are regularly associated with intraventricular choroid plexus lipoma. It has been suggested that rests of residual meningeal tissue differentiate into adipose tissue and cause mechanical obstruction to the growth of the corpus callosum .

Syndrome associated with callosal agenesis

- ACC may be sporadic or familial
- Besides the anomalies described above , there are now several well-defined syndromes that callosal agenesis is an important feature .

Syndrome associated with callosal agenesis

- Aicardi syndrome：X-linked dominant , only affects female , combined with infantile spasms , choriorentinal lacuna , mental retardation , and vertebral anomalies .
- Menkes et al → AR syndrome , which seizures were a prominent early feature.

Syndrome associated with callosal agenesis

- Acrocallosal syndrome：includes polydactyly , macrocephaly and mental retardation.
- Pineda et al → two siblings with callosal agenesis , hypothermia and apnoeic spells .

Conclusions

(1)Clinical symptomatology varies , and may largely depend on associated malformations .

(2)ACC can be entirely asymptomatic and only come to light with sophisticated psychological testing .

(3)The diagnosis is readily confirmed radiologically .