MEDICAL UPDATE
CASE PRESENTATION

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F.C.S (Neurosurgery)

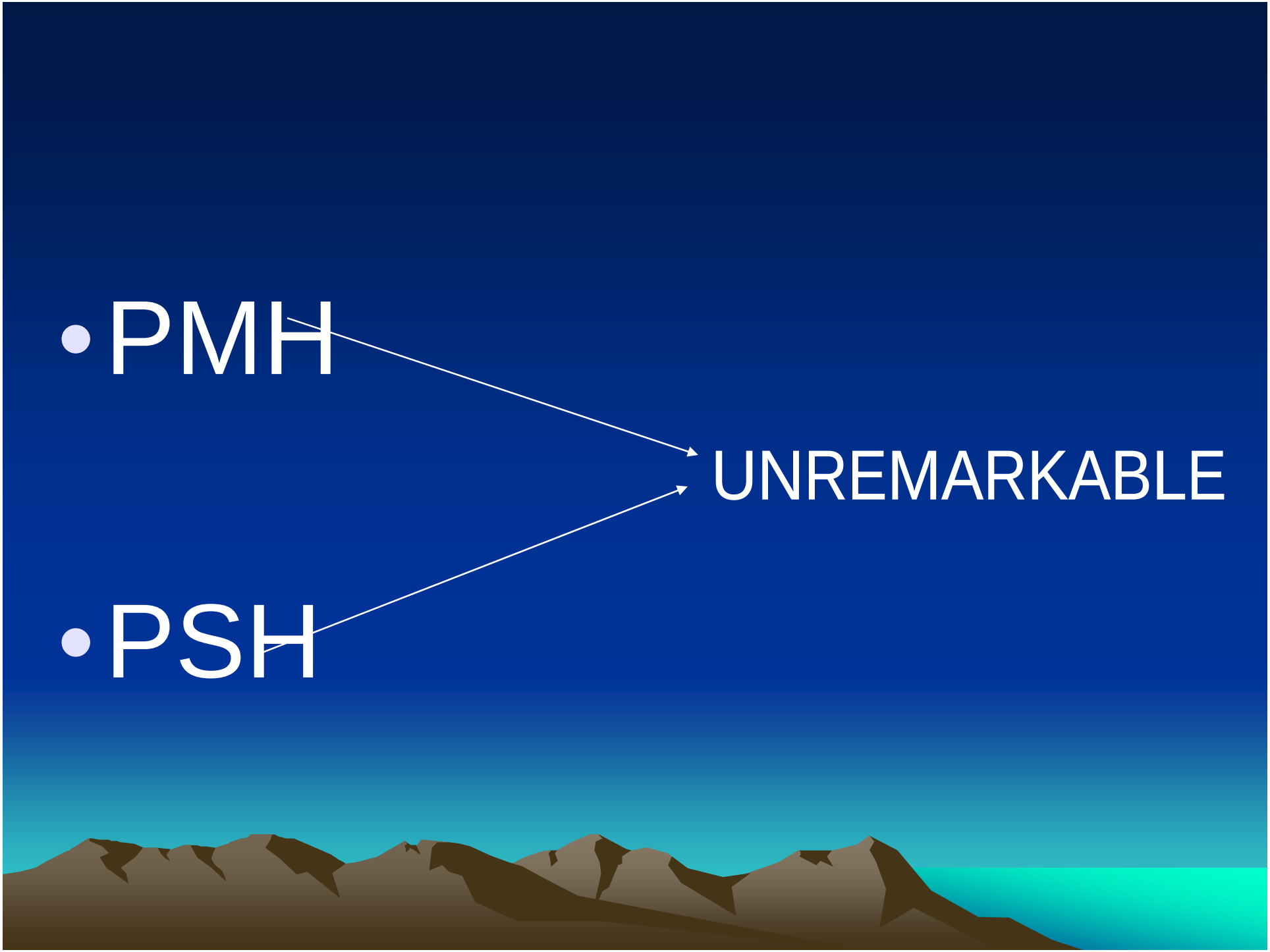
CONSULTANT NEUROSURGEON

DEPT OF NEUROSURGERY
VICTORIA HOSPITAL

- **NAME**: S.M
- **AGE/SEX**: 13 yrs, Male, student
- **D.O.A**: 17/6/11
- **D.O.D**: 29/6/11

CHIEF COMPLAINTS

- Headache on & off since 6/52 over occipital region
- Gets worse on waking up
- Vomitting
- Photophobia & phonophobia
- Blurring of vision & generalised weakness


PMH
PSH
UNREMARKABLE

- **GENERAL PHYSICAL EXAMINATION**:

Normal

- **SYSTEMIC EXAMINATION**:

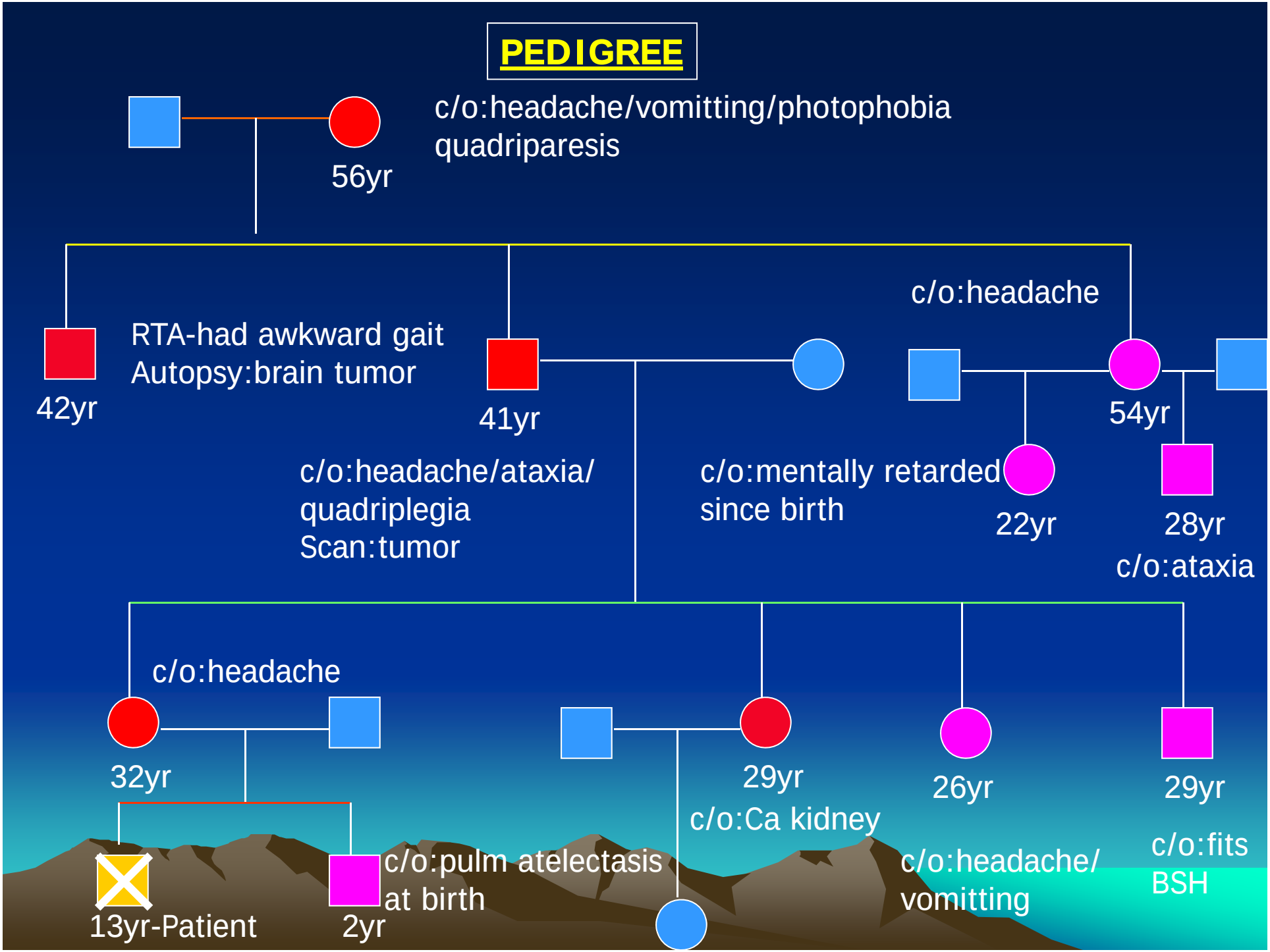
CVS

RS

P/A

Normal

- CNS:
  - GCS: 15/15
  - Pupils: B/L sluggishly reacting to light
  - Higher mental status: Normal
  - Cranial nerves examination: Normal
  - Focal deficits: -Blurring of vision
    -Ataxic gait
  - Power 5/5 in all limbs


PEDIGREE
c/o:headache/vomitting/photophobia
quadriparesis
56yr
RTA-had awkward gait
Autopsy:brain tumor
42yr
41yr
c/o:headache/ataxia/
quadriplegia
Scan:tumor
c/o:headache
c/o:mentally retarded
since birth
22yr
54yr
28yr
c/o:ataxia
c/o:headache
32yr
13yr-Patient
2yr
c/o:pulm atelectasis
at birth
29yr
c/o:Ca kidney
26yr
c/o:headache/
vomitting
29yr
c/o:fits
BSH

INVESTIGATION

- BASELINE BLOOD ANALYSIS: NORMAL
- CT BRAIN WITH CONTRAST:

  Midline 4 x 4 cm post. Cranial fossa lesion s/o pilocystic astrocytoma with mass effect and obstructive hydrocephalus was noted

- MRI

  A mass of ~6 cm in post. Cranial fossa,more on R-side with features s/o brain edema is seen

INVESTIGATION (conti)

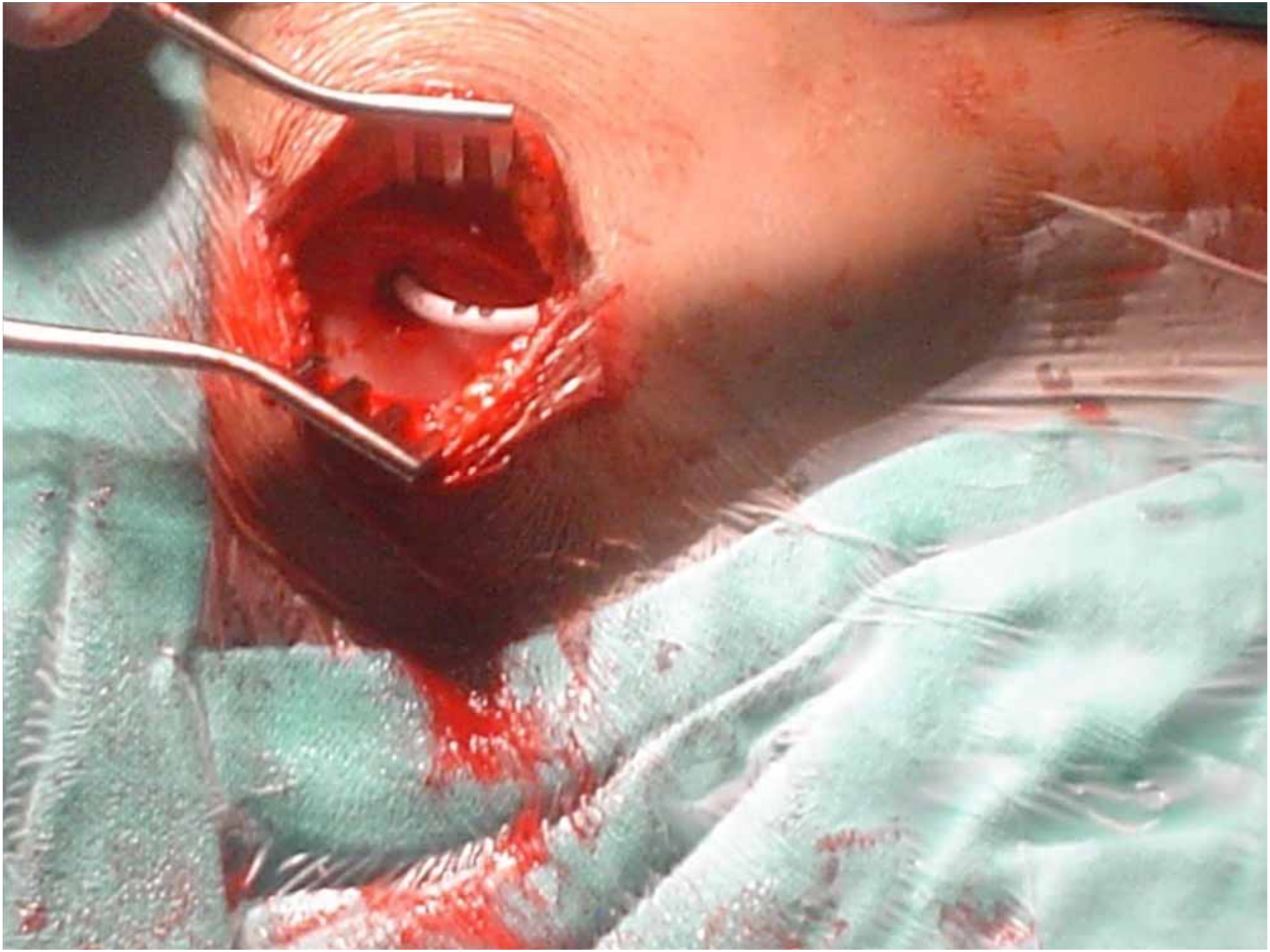
- Ultrasound

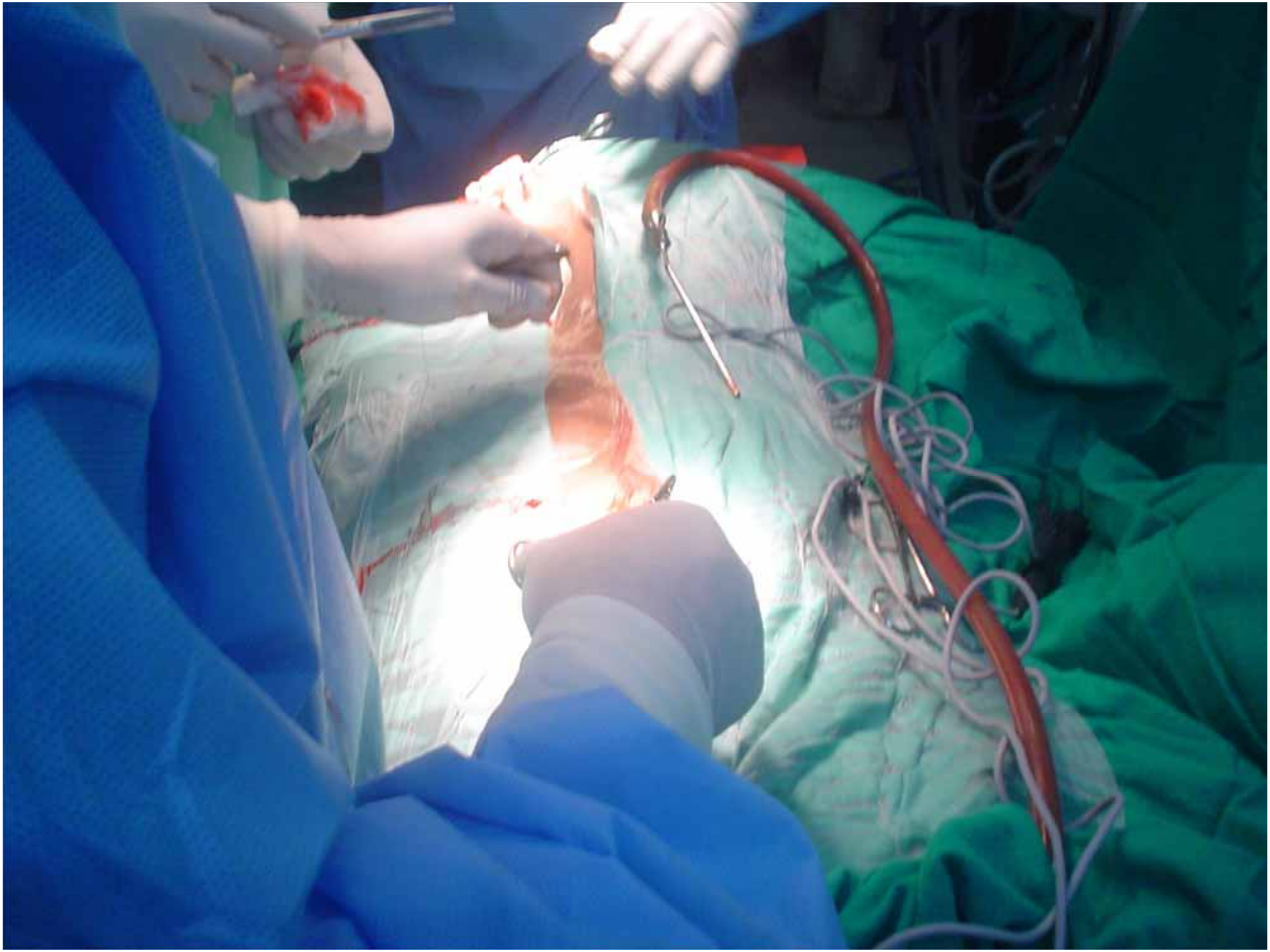
  Abdomen (liver, spleen, pancreas, kidney, suprarenal,pelvis)-Normal

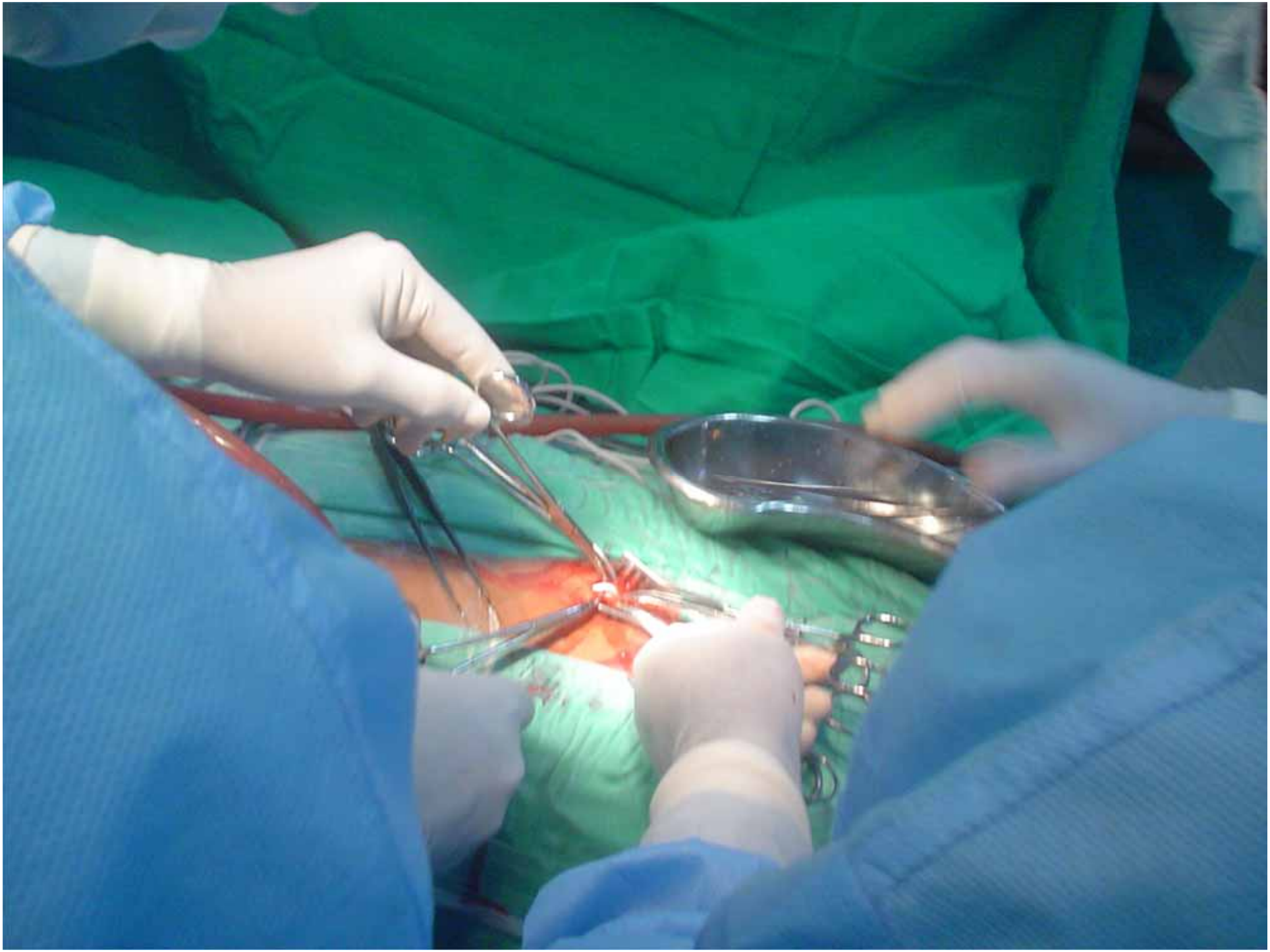
- Opthalmological assessment - Normal

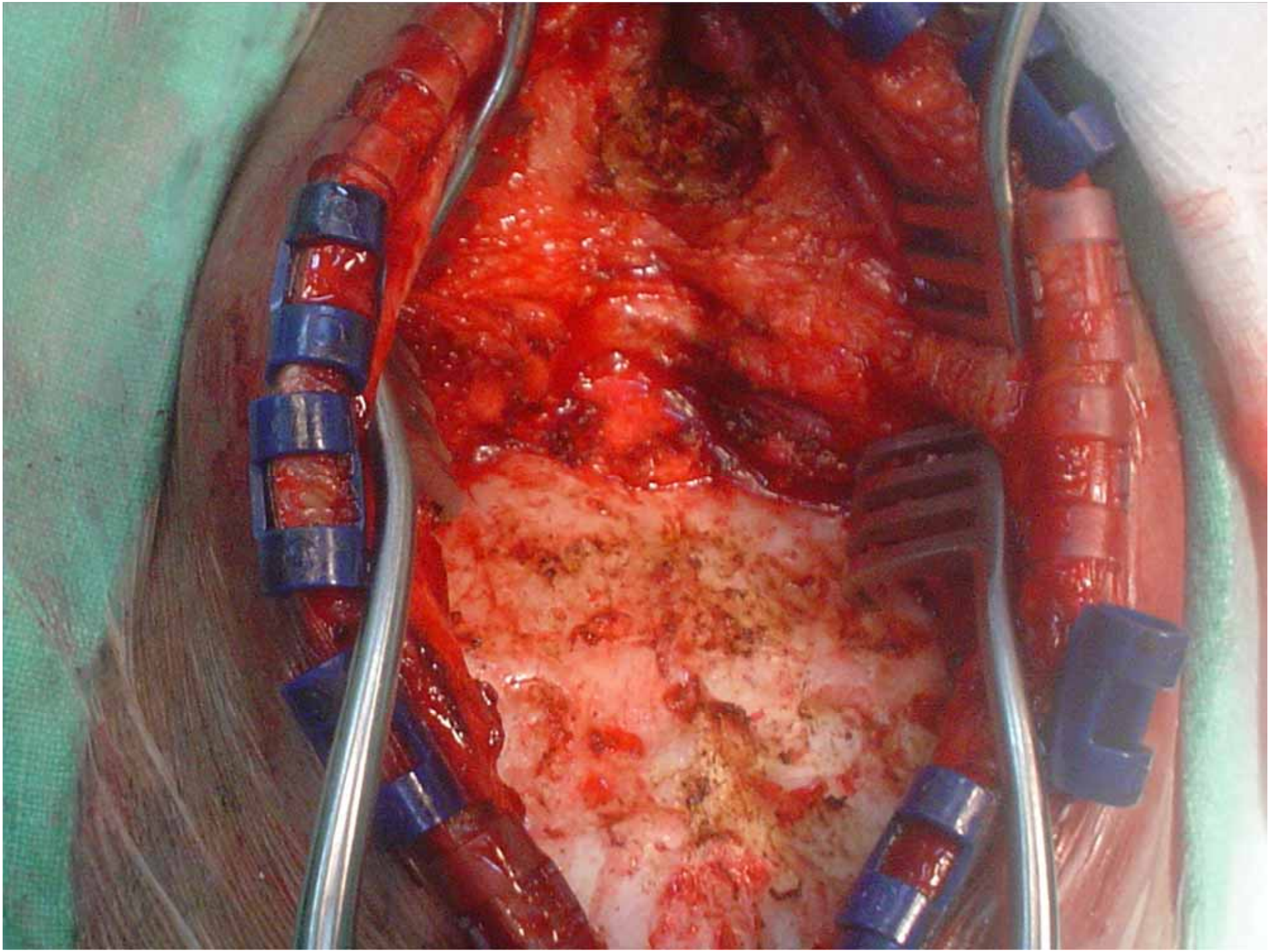
MANAGEMENT

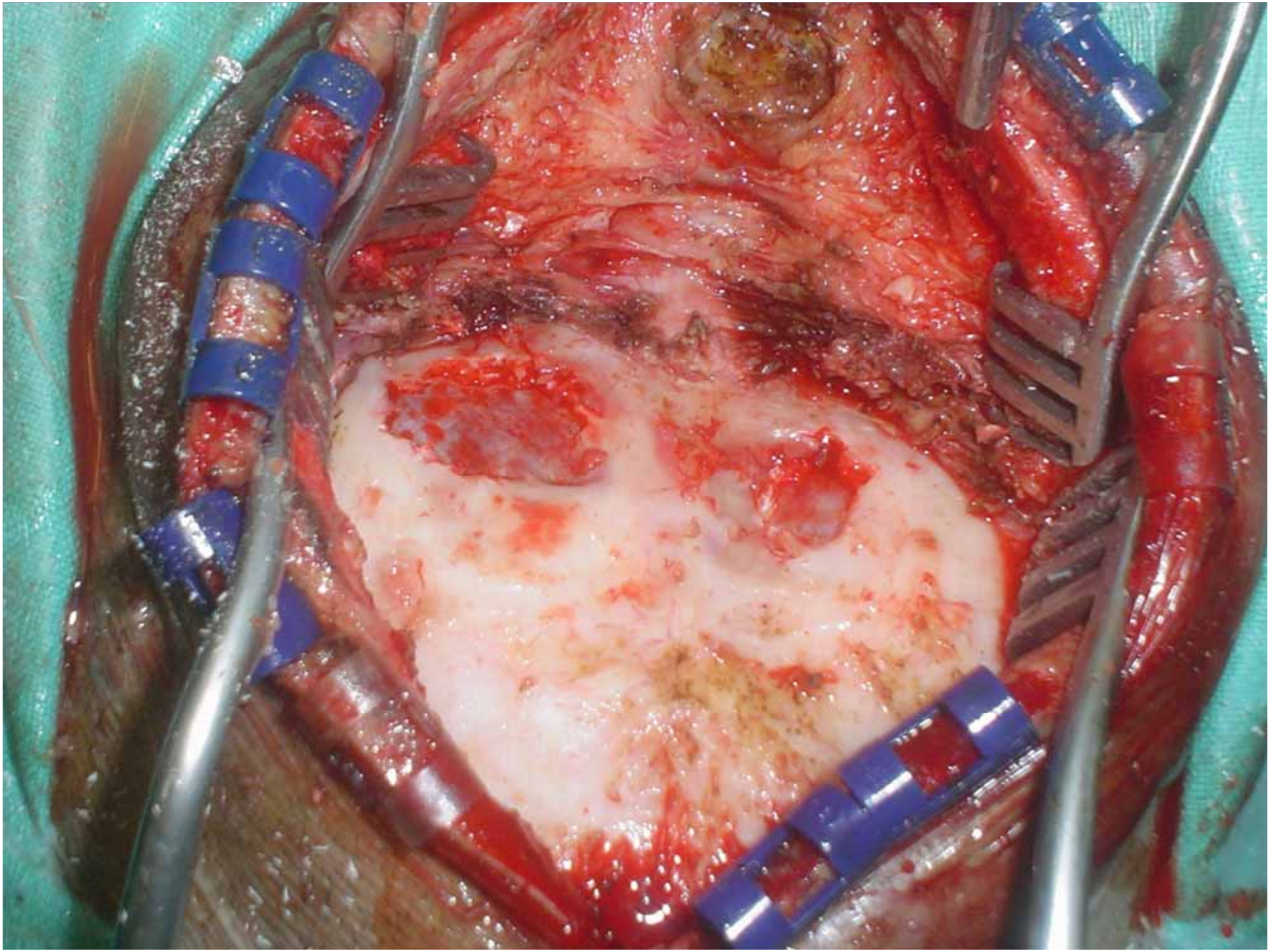
- A ventriculoperitoneal shunt was inserted to relieve increased intracranial pressure on 02/06/11
- Posterior fossa craniectomy for excision of cerebellar cyst and haemangioblastoma was done on 20/06/11

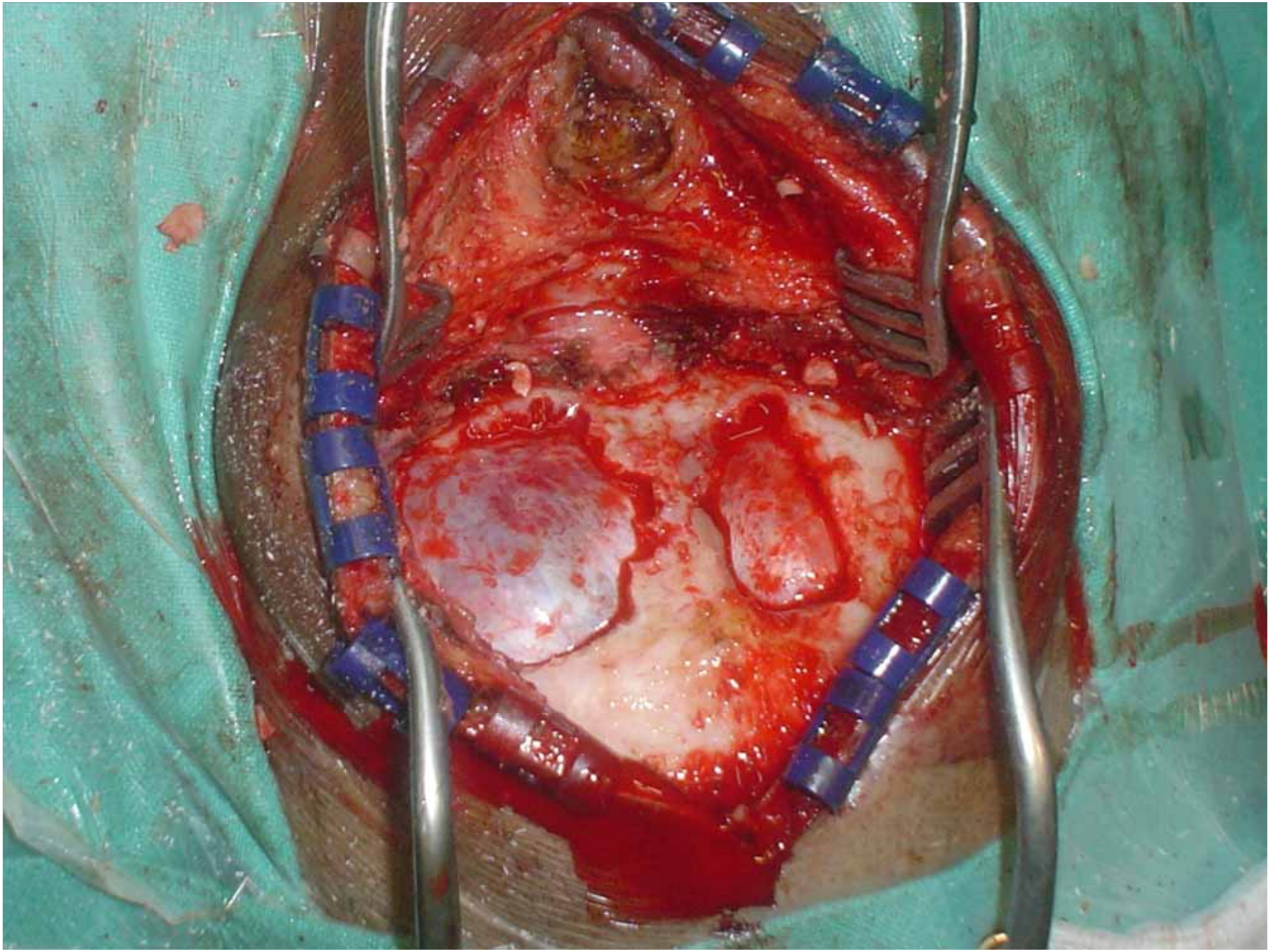


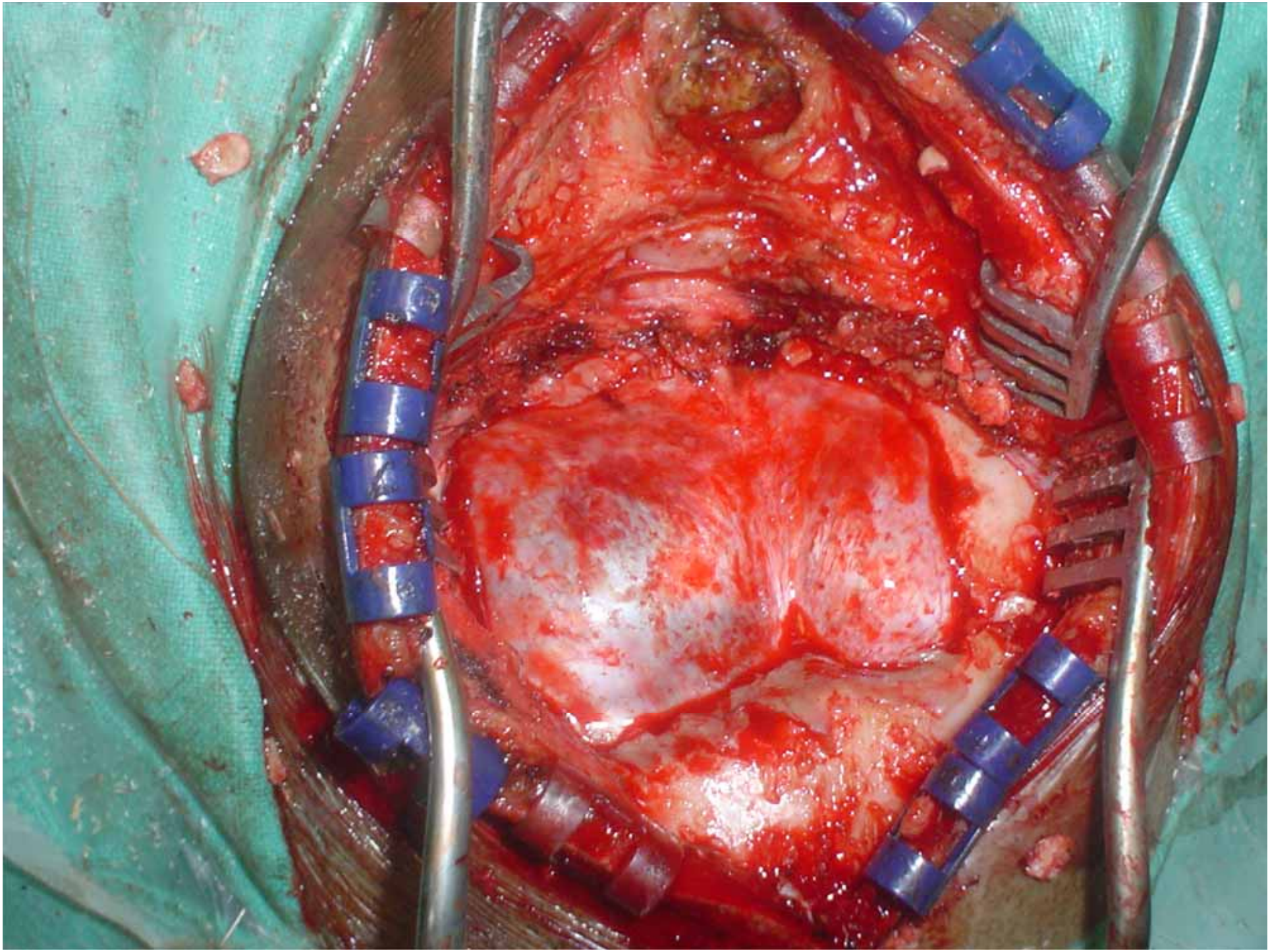


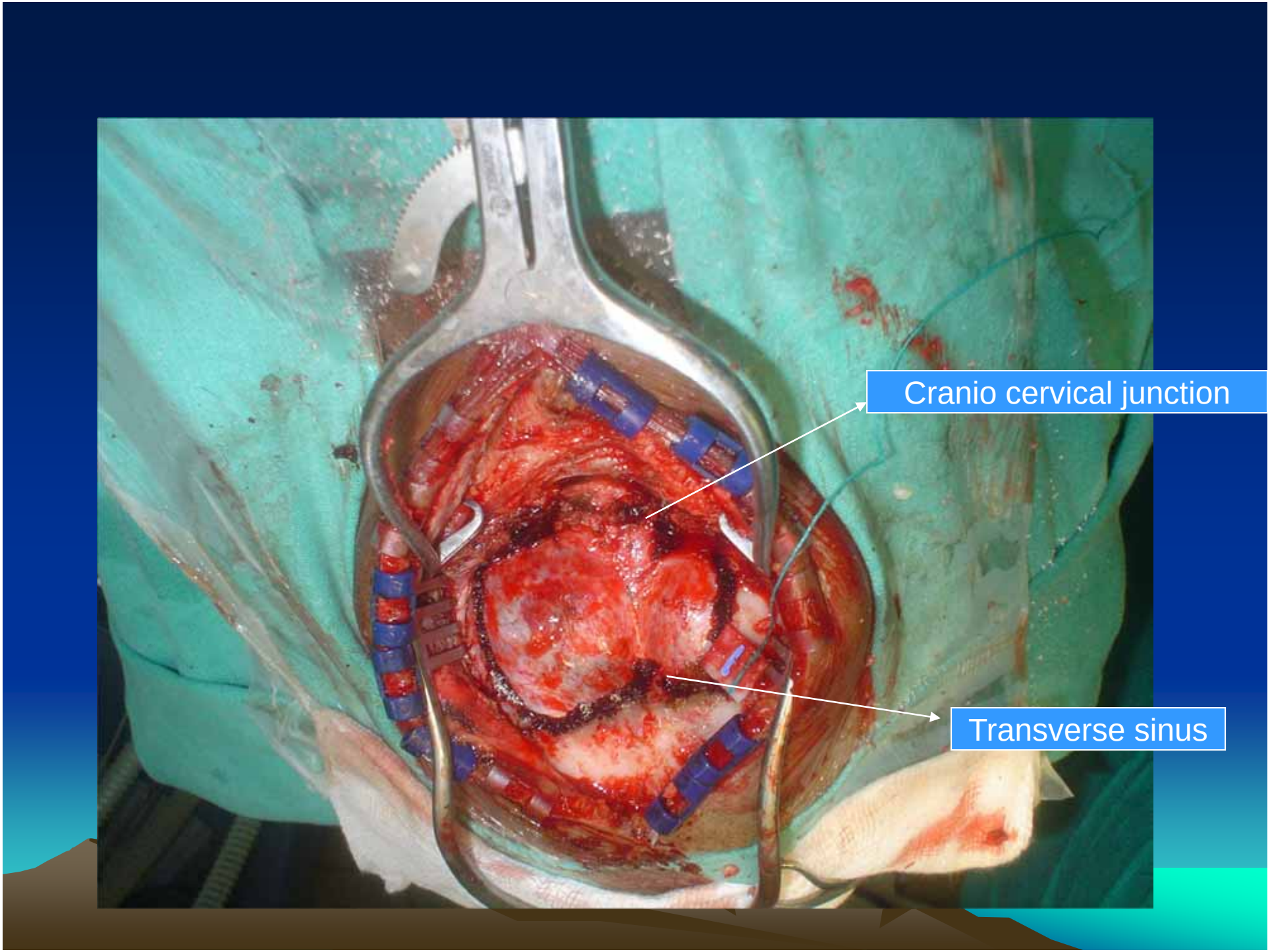


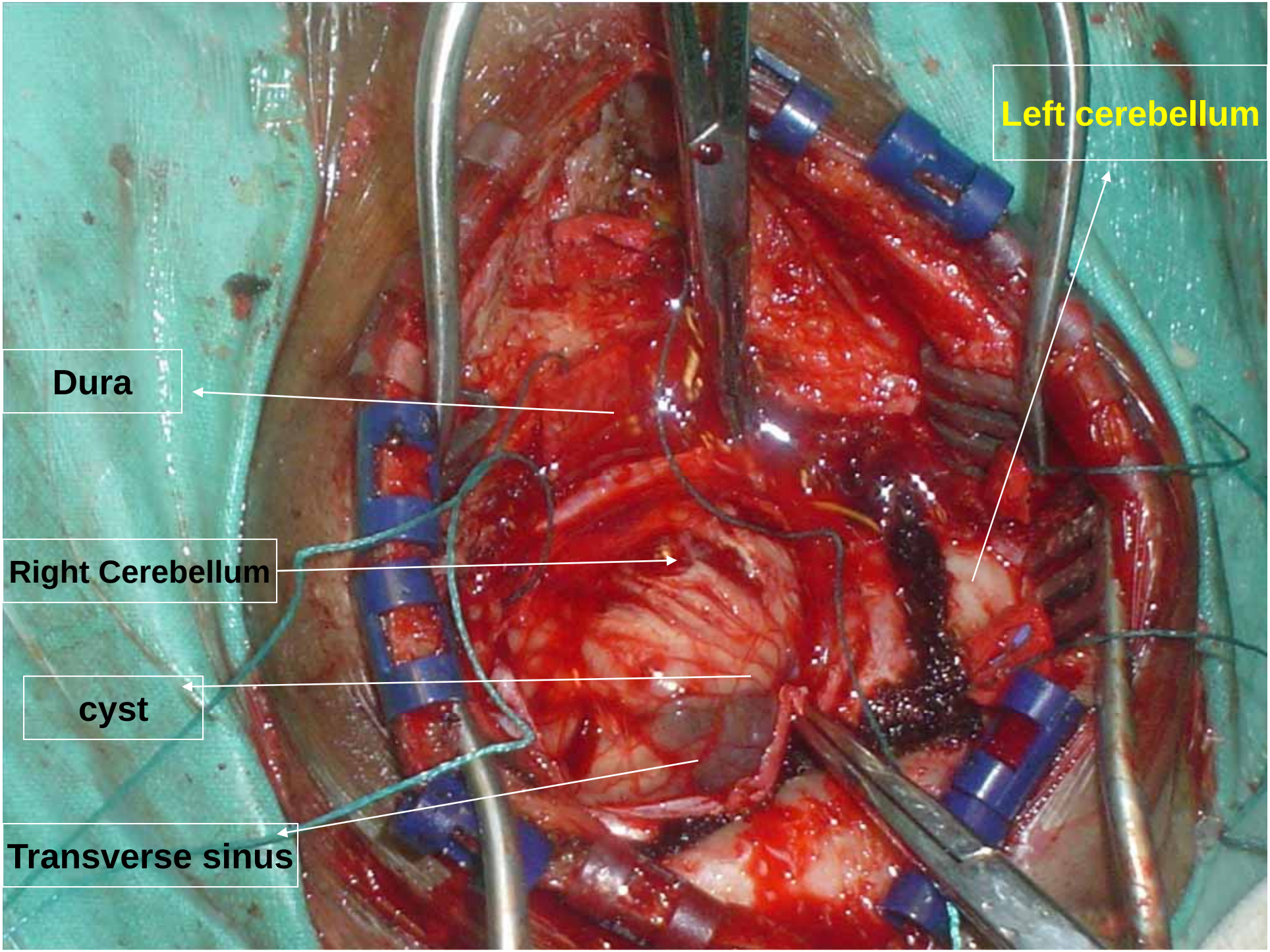


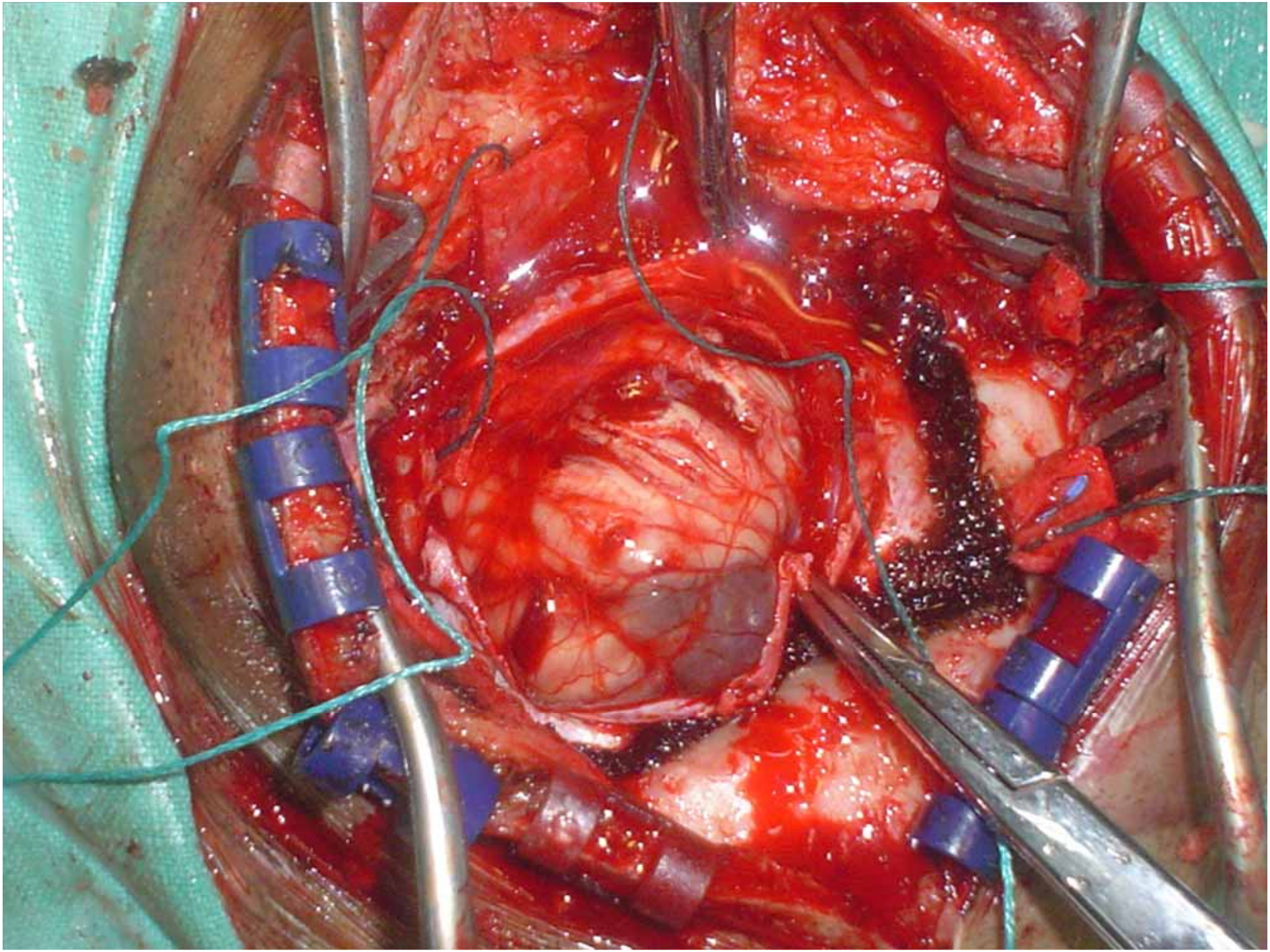




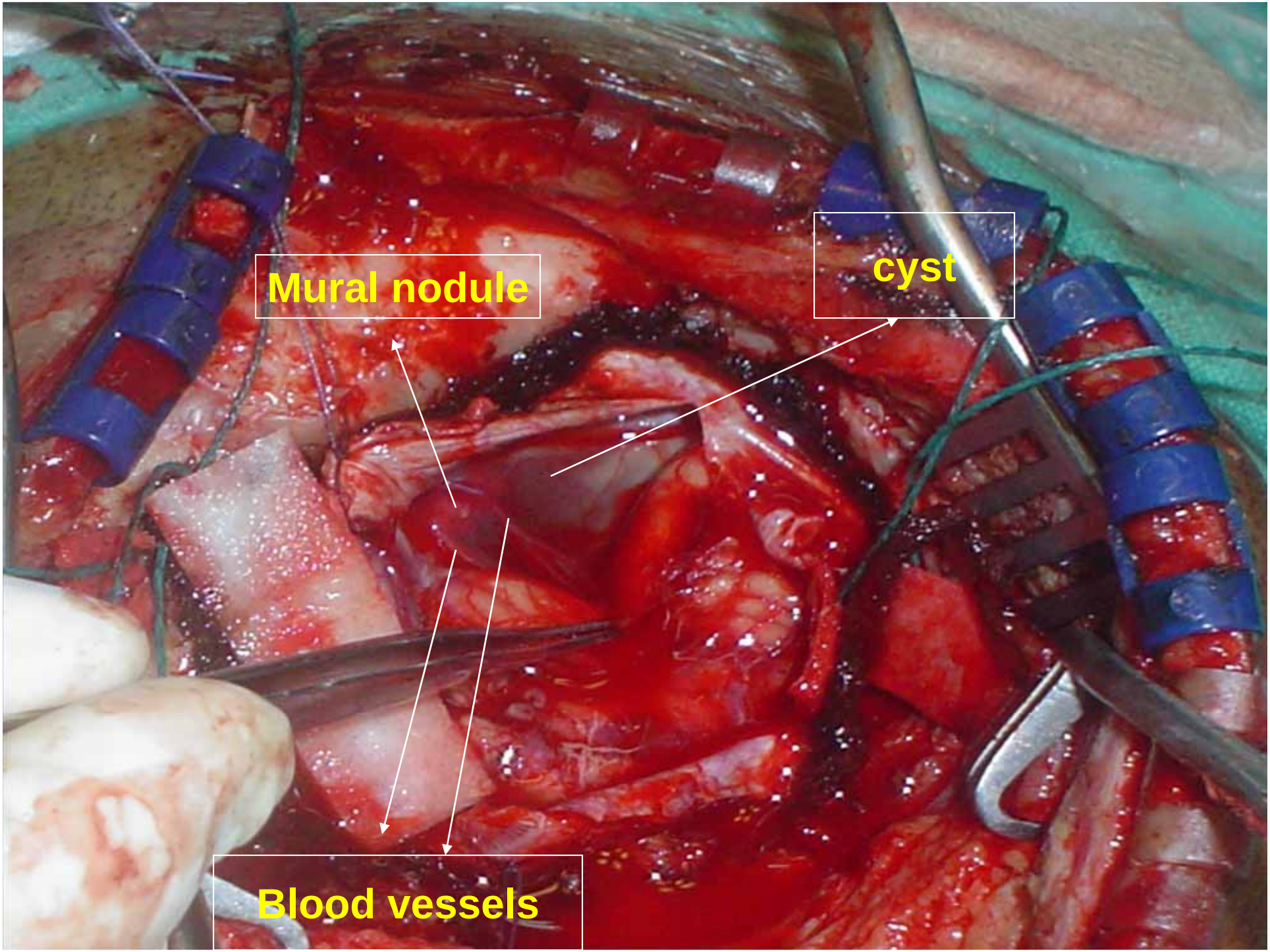


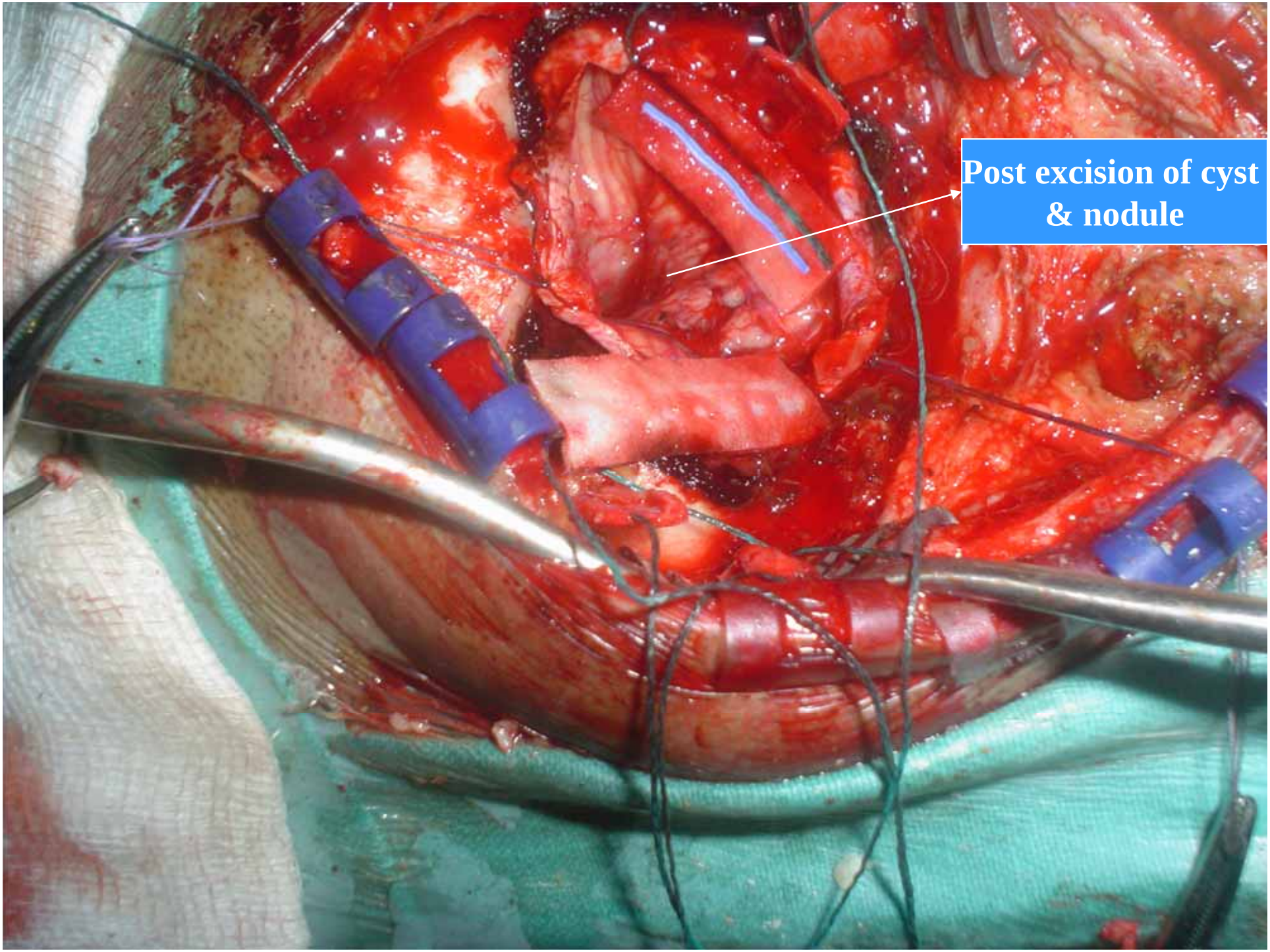

Cranio cervical junction
Transverse sinus

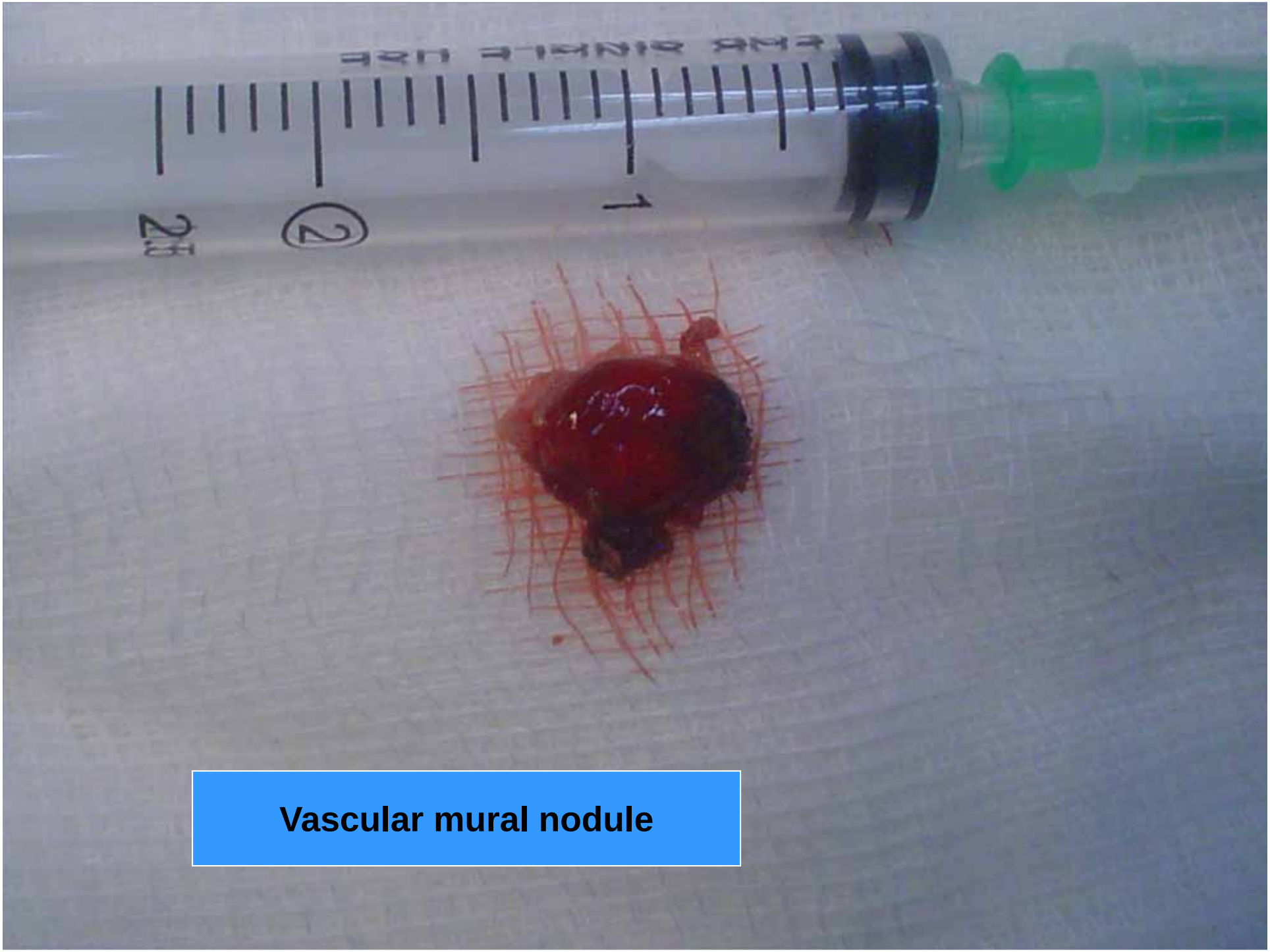

Left cerebellum
Dura
Right Cerebellum
cyst
Transverse sinus

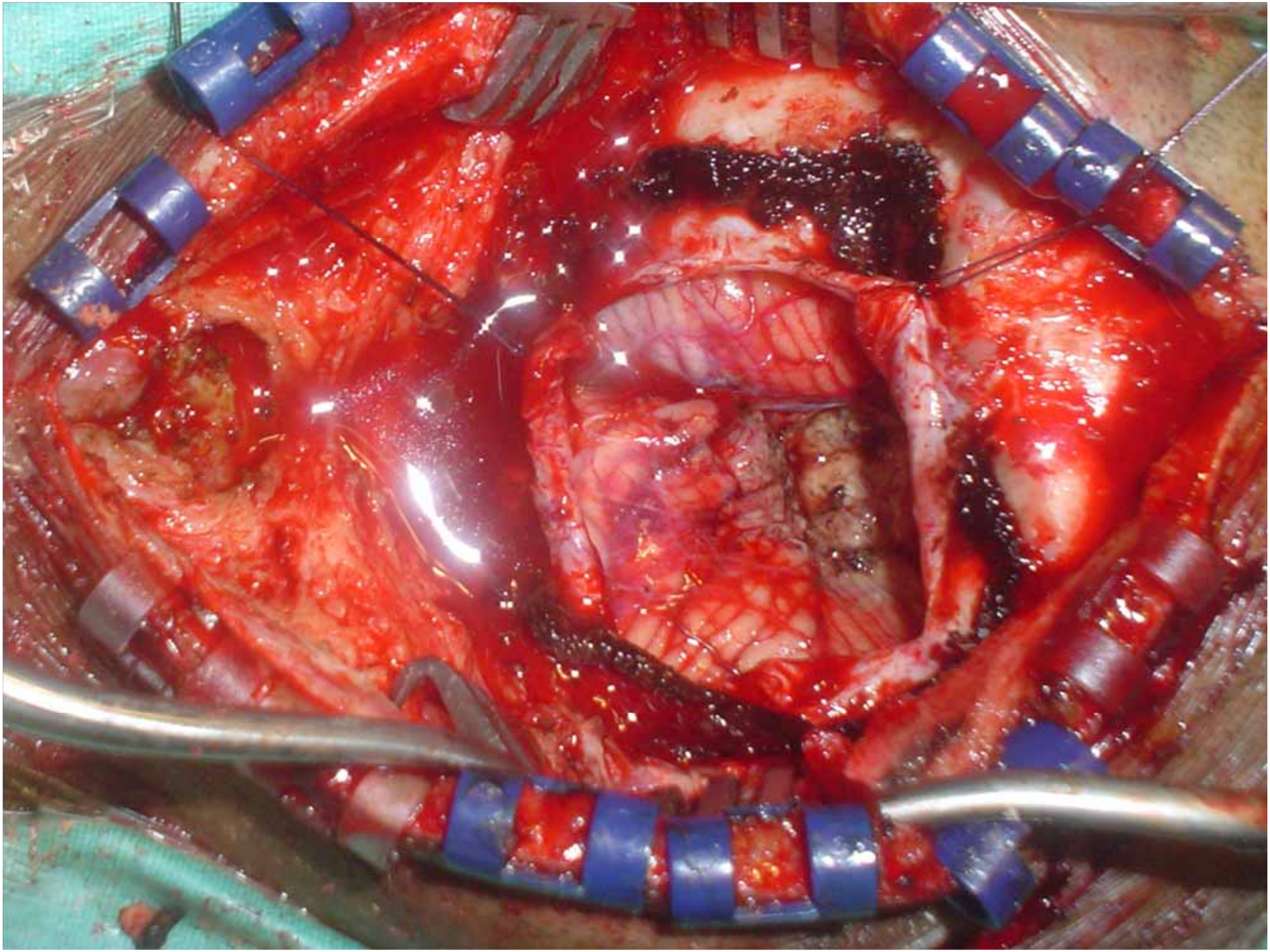


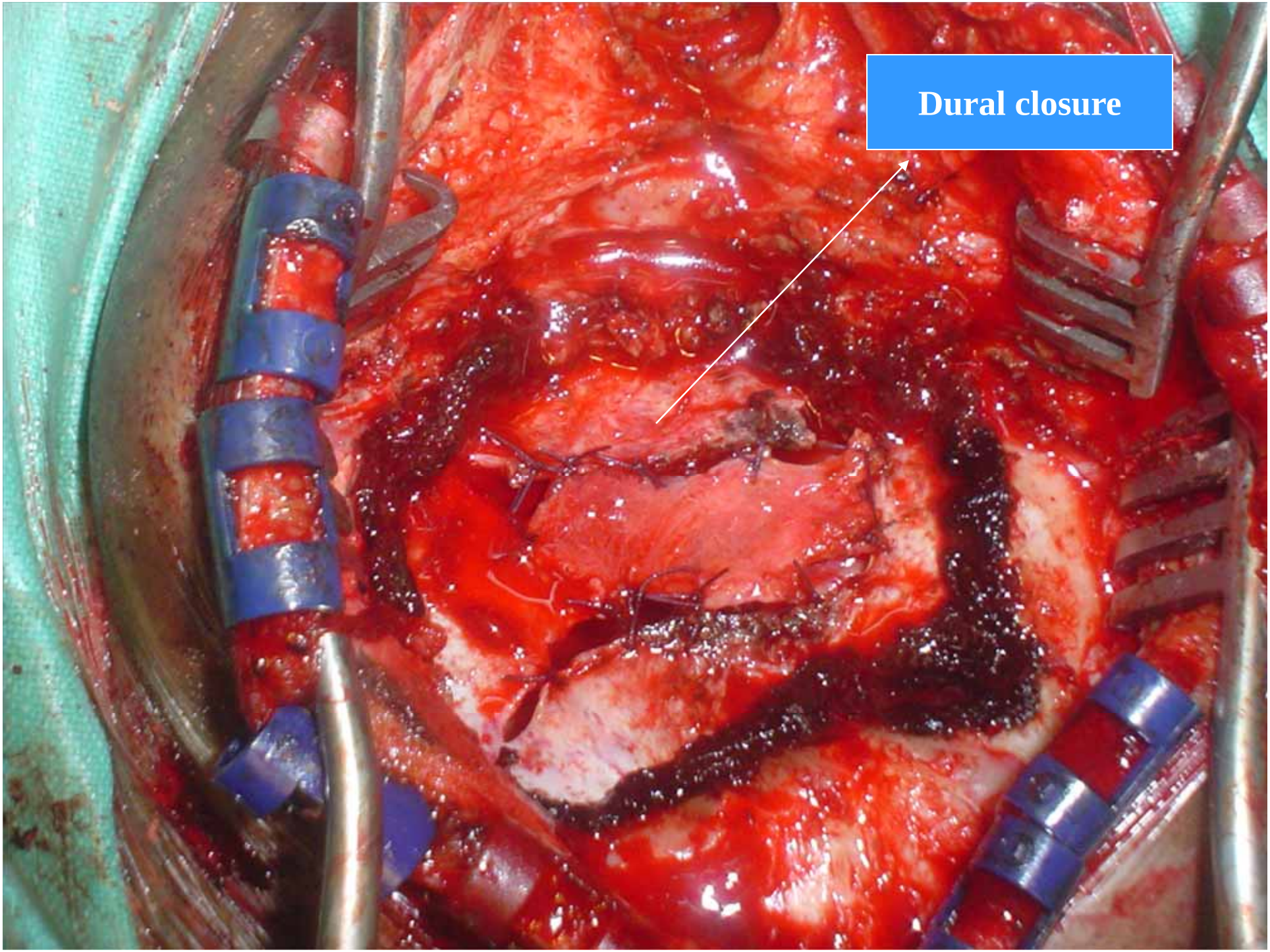
Micro -dissection


Mural nodule
cyst
Blood vessels


Post excision of cyst
& nodule


Vascular mural nodule




Dural closure

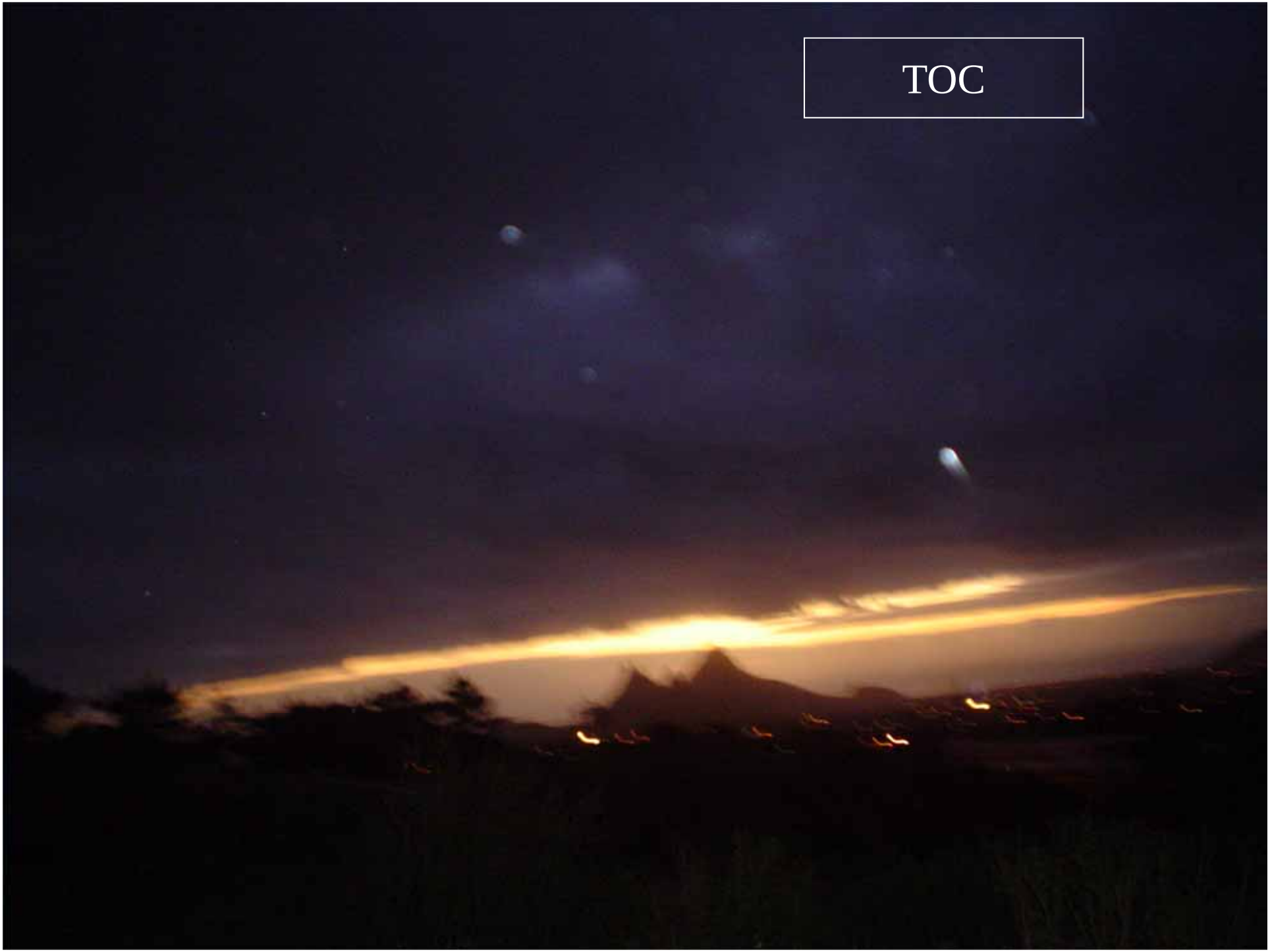
FOLLOW UP

- ✓ after one week for stitch removal
- ✓no headache
- ✓Vision improved
- ✓normal gait

HISTOPATHOLOGY REPORT

CASE ONE

Posterior fossa benign haemangioblastoma

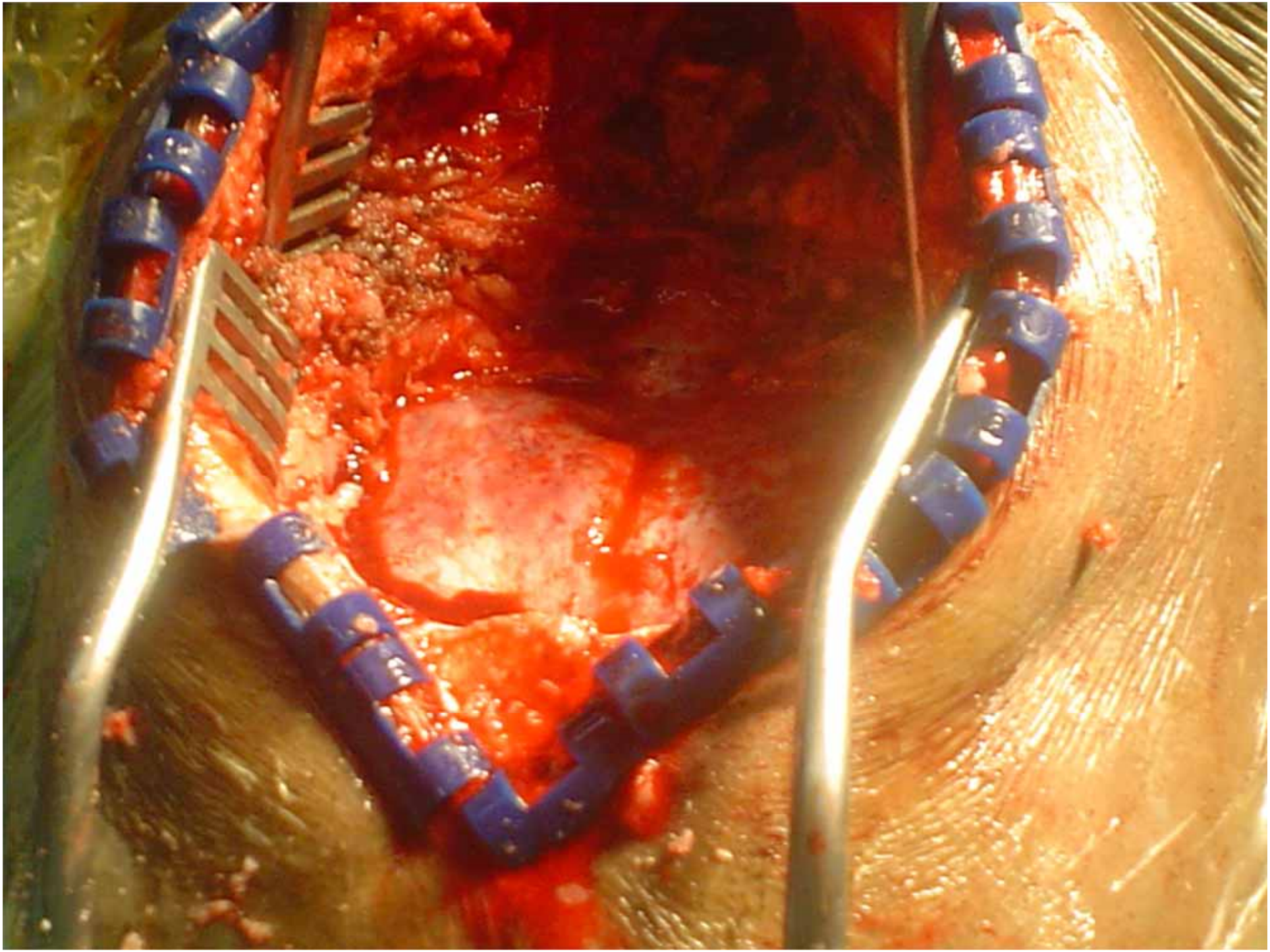

TOC

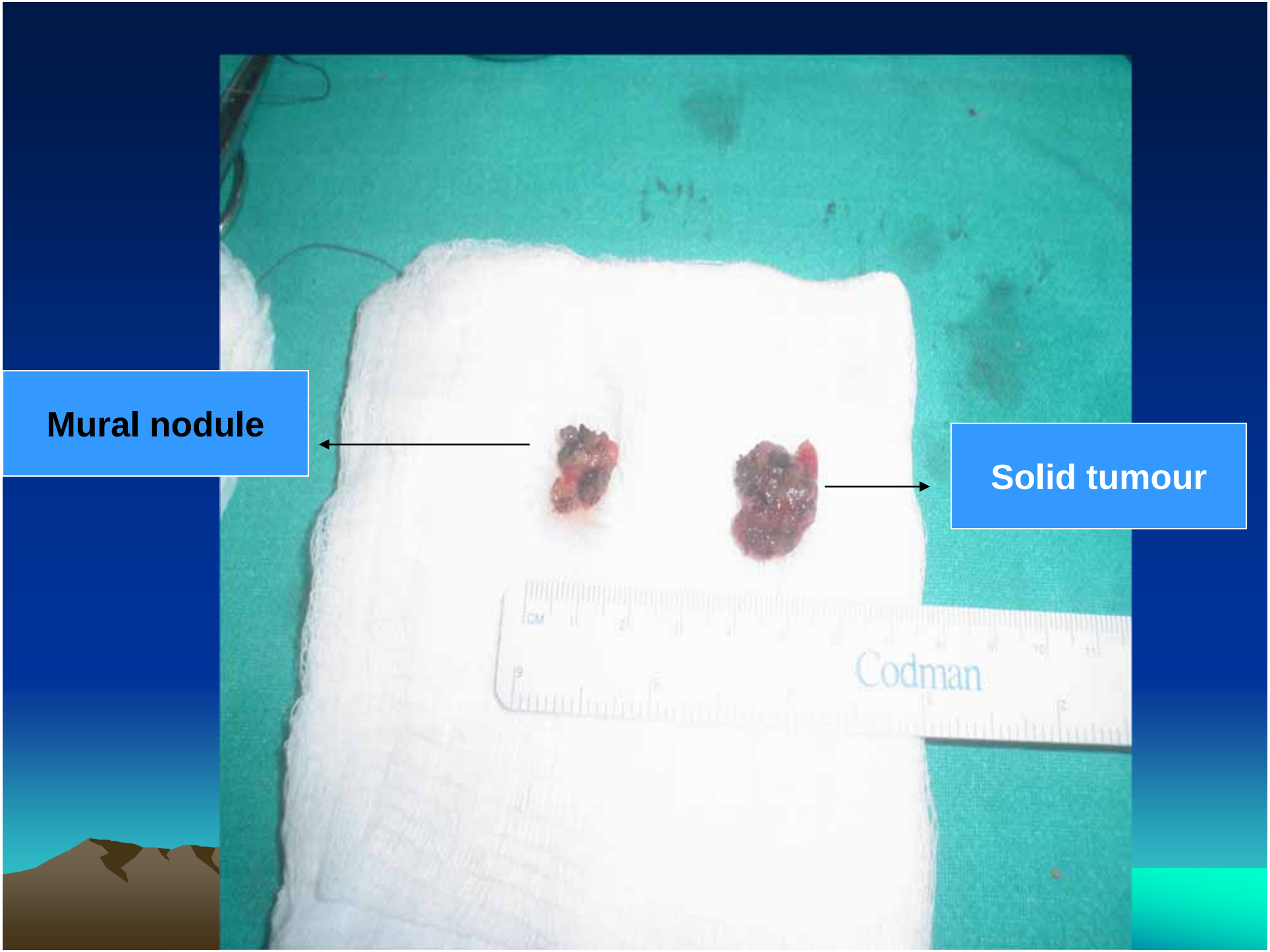
CASE 2

- **D.G -45 YRS**
  MALE
- **MAY 2012**
- **COMPLAINTS**
  - **Unsteadiness of gait**
  - **Headache**

- G.C.S 15/15
- Cranial nerves normal
- Dysmetria
- Truncal ataxia
- Baseline investigation- normal
- Chest X-Ray -normal

- MRI BRAIN
- ULTRASOUND ABDOMEN + NECK
- CT ABDOMEN




Mural nodule
Solid tumour
Codman

Autum dry leaves
Oil on canvas.Size-
1994
73 x 107 cm

HISTOPATHOLOGY REPORT

CASE 2

- 1 Cerebellar metastasis of a Renal Cell carcinoma (clear cell)
- 2 Haemangioblastoma

VON HIPPEL LINDAU

- 1927 Arvid Lindau
- Connection between Retinal angiomas and hemangiomes of the cerebellum
- Rare autosomal dominant genetic condition in which haemangioblastomas are found in the cerebellum, spinal cord, kidney and retina
- Other associated pathologies include Retinal angioma, Renal Cell carcinoma and phaechromo cytoma
- Mutation in the Von Hippel tumour suppressor gene on chromosome 3p25

- Cerebellar haemangioblastoma affect 48 % of the patient with VHL

GENETICS

- Inherited in an autosomal dominant Mendelian pattern with a frequency of approximately of 1 case per 36,000 newborn

VON HIPPEL LINDAU (VHL)

- 1927 Arvid Lindau
- Connection between Retinal angiomas and hemangiomes of the cerebellum
- Cerebellar haemangioblastoma affect 48 % of the patient with VHL

DIAGNOSIS

- Family history + a single cerebellar hemangioblastomas

OR

- one hemangioblastoma + 1 visceral tumour

RELATED CONDITIONS

- Renal cell carcinoma (25 %)
- Pheochromocytoma (10 %)
- Polycythemia (9-20 %)

HEMANGIOBLASTOMAS IN OTHER LOCATION

- Supratentorial hemangioblastomas
- Optic nerves
- Spinal cord
- Peripheral nerves
- Retina

PATHOLOGY

- Posterior cranial fossa around IV ventricle, cerebellar hemisphere, vermis, Medulla or pons
- Retinal hemangioblastoma 6 %
- No distant metastasis
- Well circumscribed but do not have a true capsule
- Solid part is a mural nodule

DIAGNOSTIC STUDIES

- MRI
- CT
- Angiography
- Ultrasonography

IMAGING

- Contrast enhanced MRI best methods for VHL to identify small nodules, cyst and solid components
- Use of gadolinium constrast agent mandatory, careful evaluation of other small enhancing nodules
- Preoperative angiography- feeding vessels and embolisation

FOUR TYPES

- Simple cyst form
- Macrocystic form
- Solid form
- Microcystic form

MICROSCOPIC FEATURES

Three groups of cell

1 endothelial cells
2 pericytes
3 stromal cells

- 40 % will develop Renal cell carcinoma (primary cause of death)
- Second most common cause of mortality is CNS haemangioblastoma
- Phaechromocytoma
- Pancreatic cysts

PROGNOSIS

- Solitary cystic - 2 % mortality
- Solid-usually involve brain stem mortality – 15-20 %
- Deep mid line attachment to medulla –lethal
- Cerebellar hemangioblastomas + visceral tumours +Renal cell = poor prognosis
- After total excision – 3- 10 % recurrence rate

GENETIC TESTING FOR VHL DISEASE

- Complete sequencing of the coding regions
- Southern blot analysis
- Fluorescent in situ hybridisation (FISH)-70 % sensitivity

FOLLOW UP

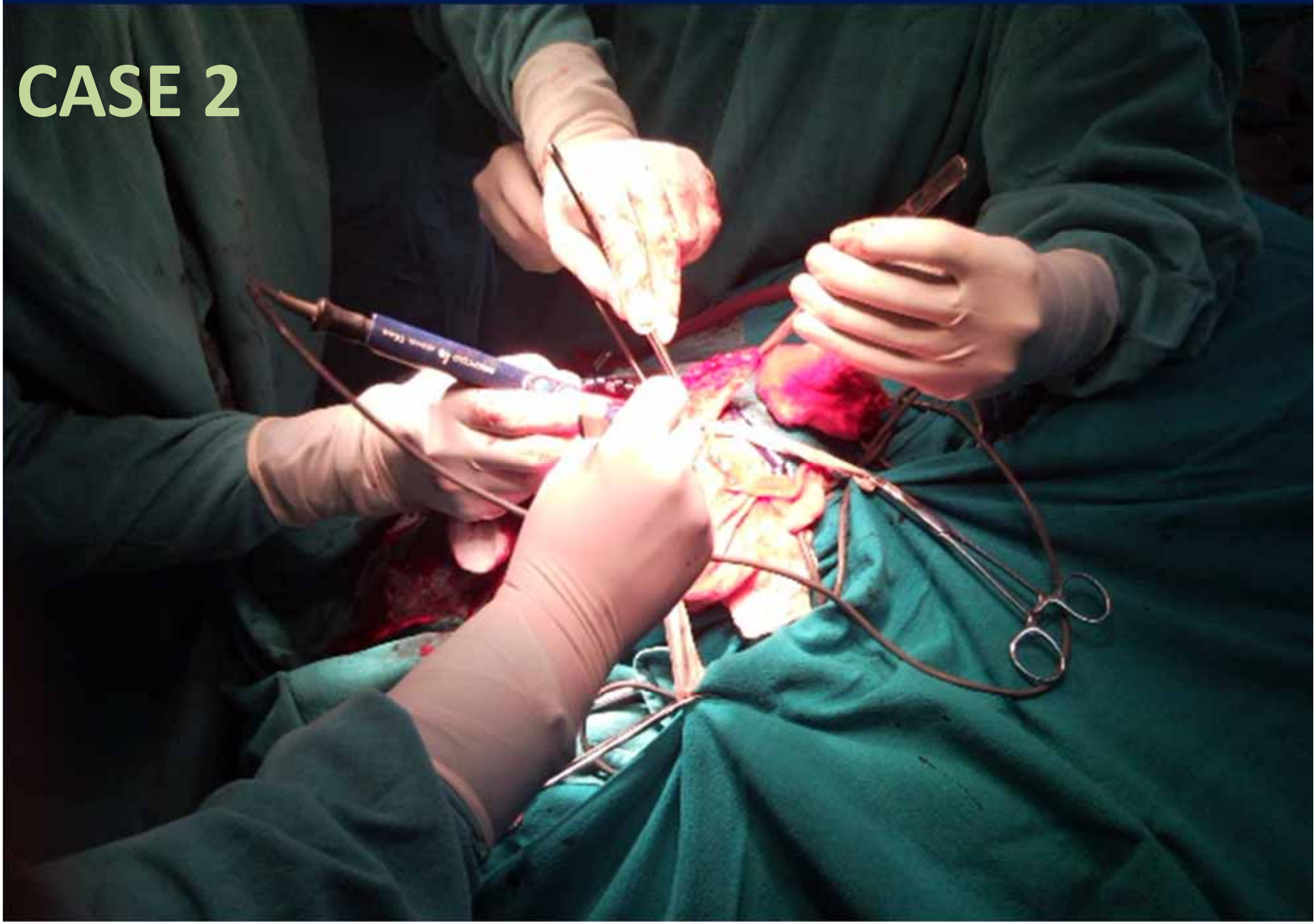
- VHL = lifetime disease
- Constantly check for tumours and cyst
- Future= molecular targeting antiagiogenic drug
- Genetic counsellors (improve psychological condition)

CAMBRIDGE PROTOCOL FOR SCREENING PATIENT FOR VHL

- Annual physical examination and urine test
- Annual direct or indirect ophthalmoscopy
- Annual angiography
- Annual renal ultrasound exam
- MRI or CT brain every 3 years to age 50 and 5 yrs thereafter
- Abdominal CT scanning every 3 years
- Annual 24 hr urine collection for Vanillylmandelic (VMA) levels

Sunset
oil colour
1999
30 x 50 cm

CASE 2



CASE PRESENTATION

NAME OF PATIENT : R. P

AGE : 41

SEX : Male

PAST HISTORY

- **Past medical History** : HBP since 5 years
- **Past surgical History** : Nil
- **Allergic History** :Nil
- **Social History** : Nil

CHIEF COMPLAINTS

- Headache since 5 months

  no other complaints.

ON EXAMINATION

- **General physical examination**
  - Unremarkable
- **Systemic examination**
  - Normal
- **Neurological examination**
  - Unremarkable
- **Local examination of the head**
  - Mild tenderness over left temporal region

INVESTIGATIONS

- **BASELINE investigations**
  - within normal limit

- **<u>SPECIFIC investigations</u>**:

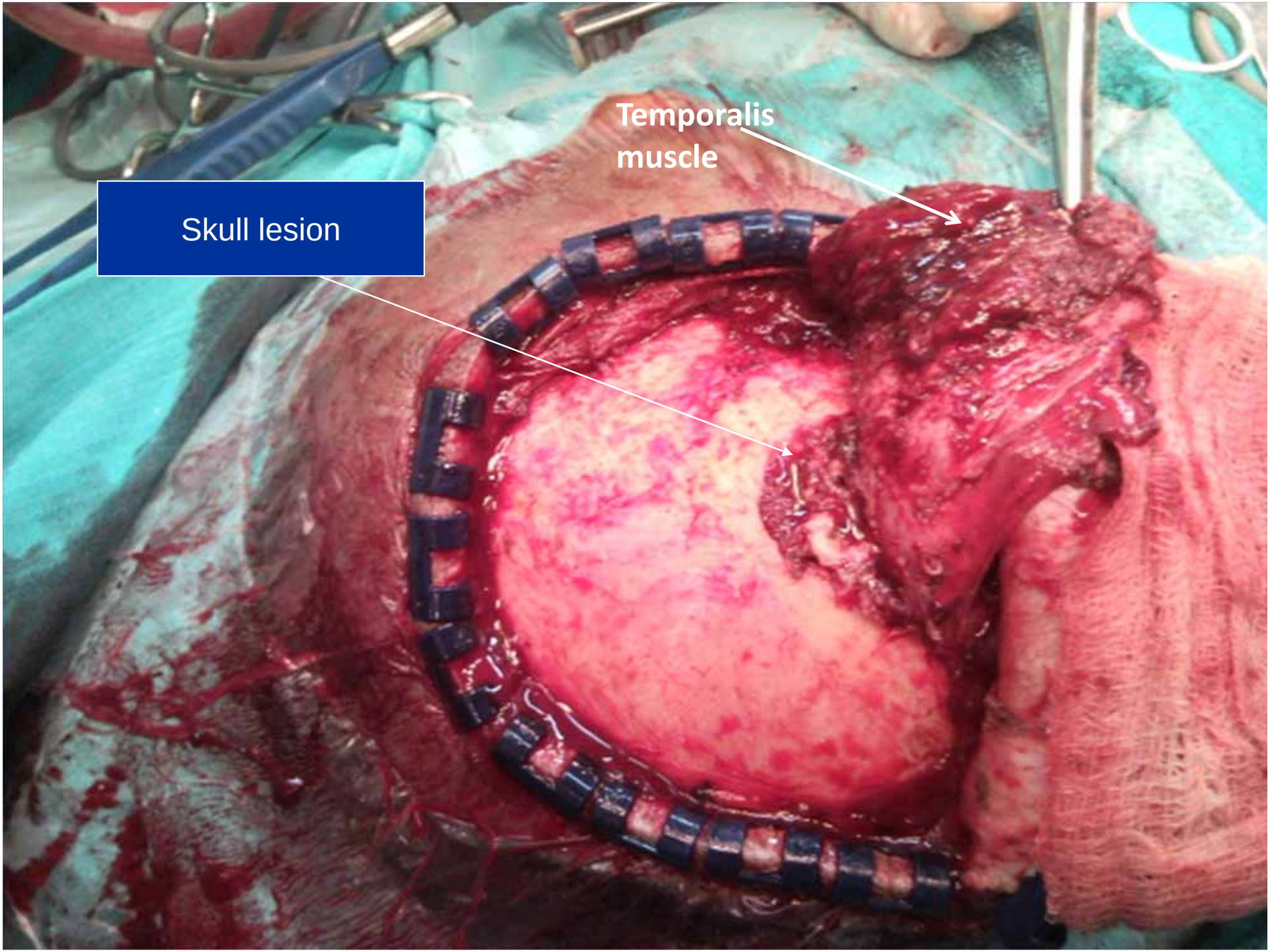
1) **HIV test** : Negative
2) **MRI BRAIN** : the main abnormality is multiple bony lesions affecting the skull vault? Metastatic disease? Lymphoma? Myeloma? Other types.
3) **CT-SCAN abdomen thorax pelvis** : normal

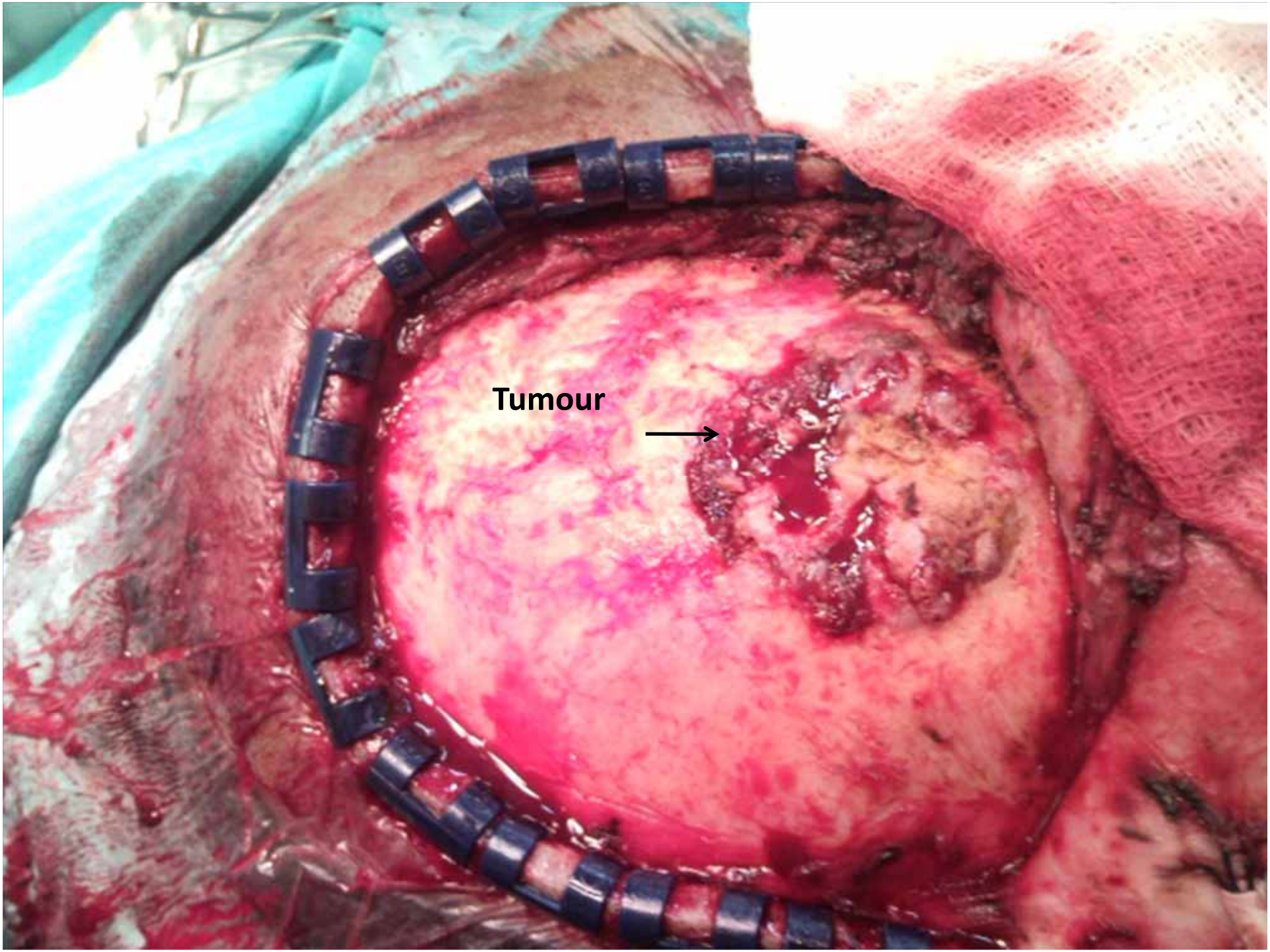
PROVISIONAL DIAGNOSIS

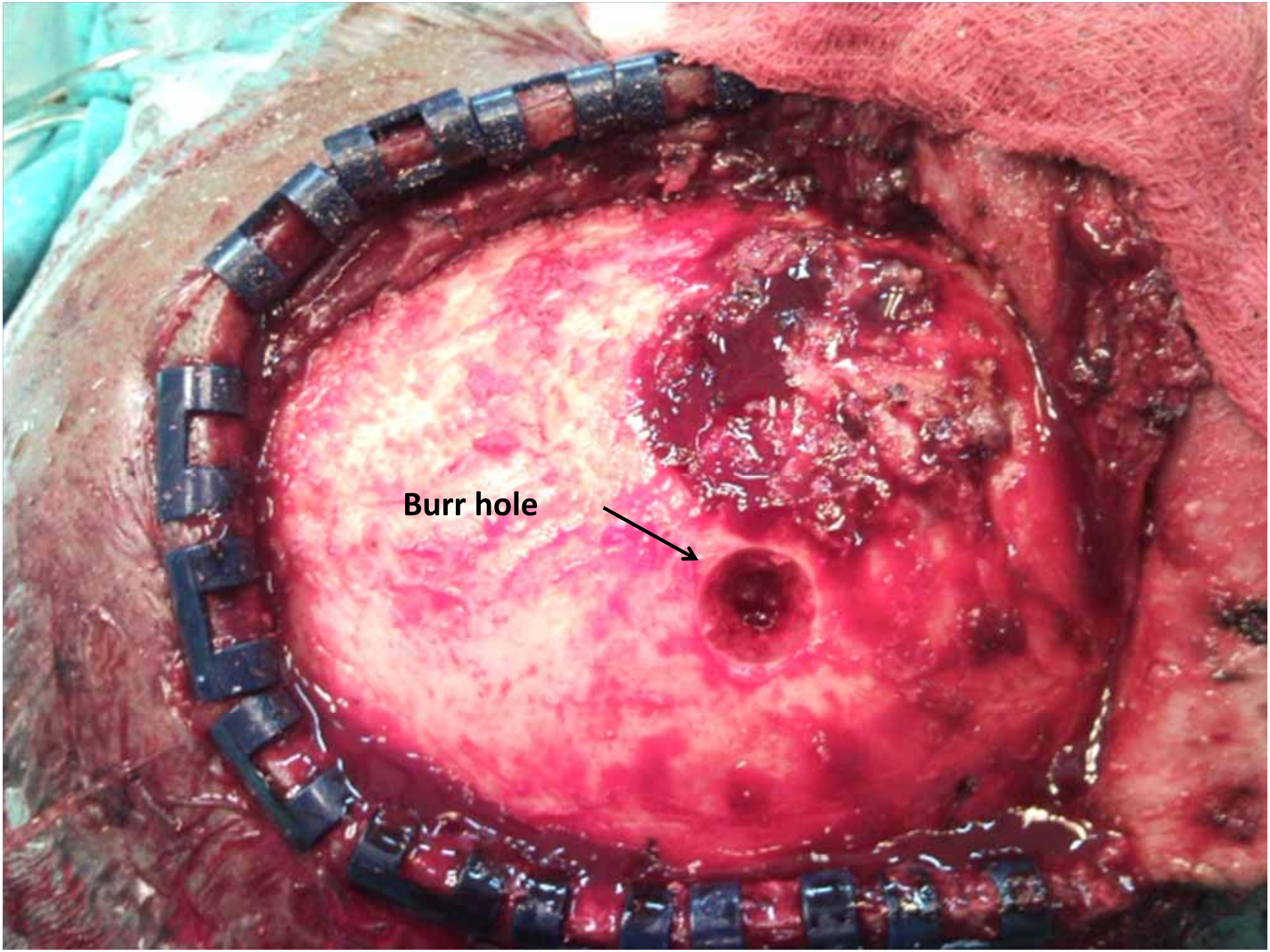
- LYMPHOMA
- MULTIPLE MYELOMA
- TUBERCULOMA
- TOXOPLASMOSIS
- MULTIPLE METASTASIS

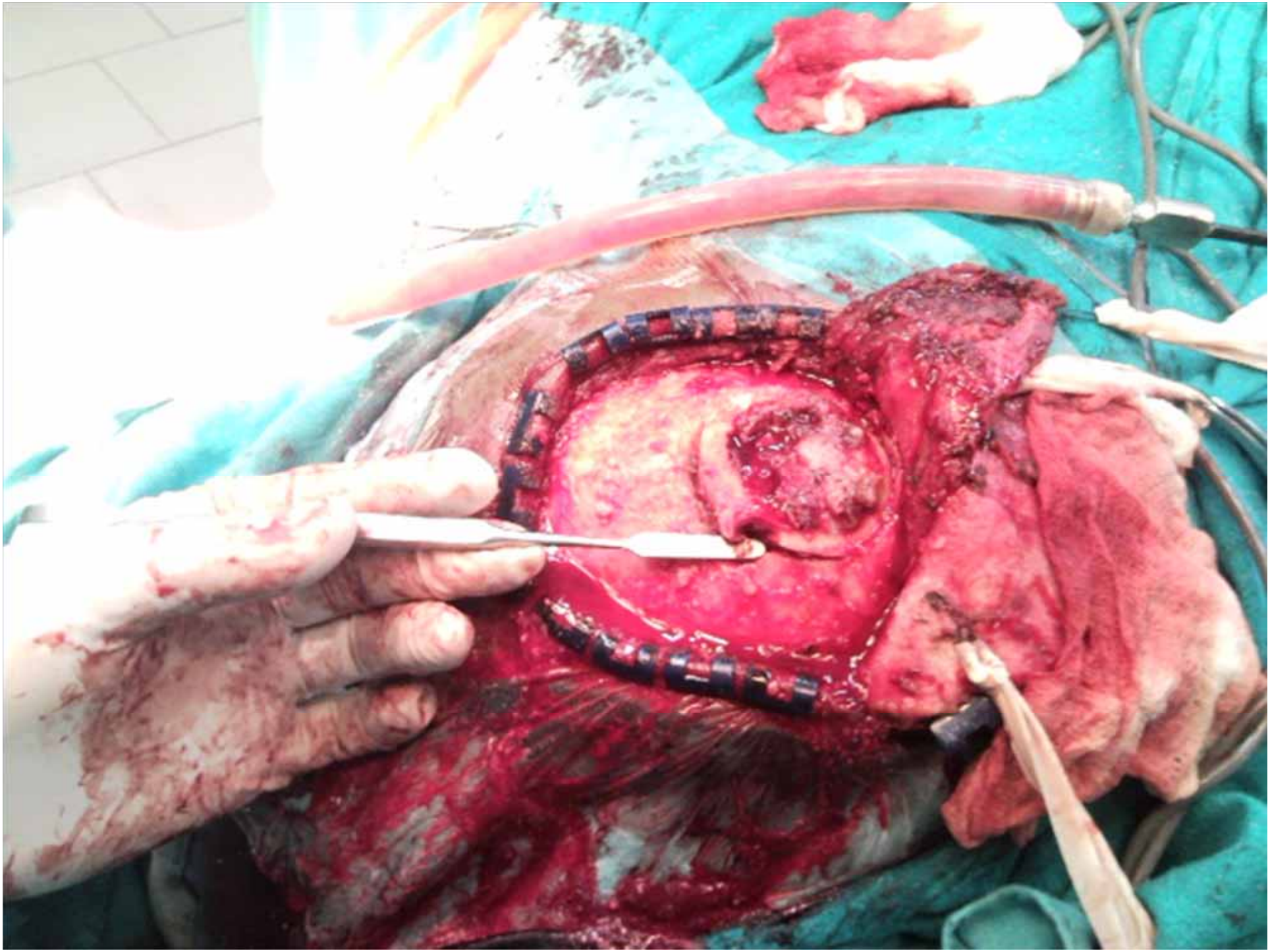
MANAGEMENT

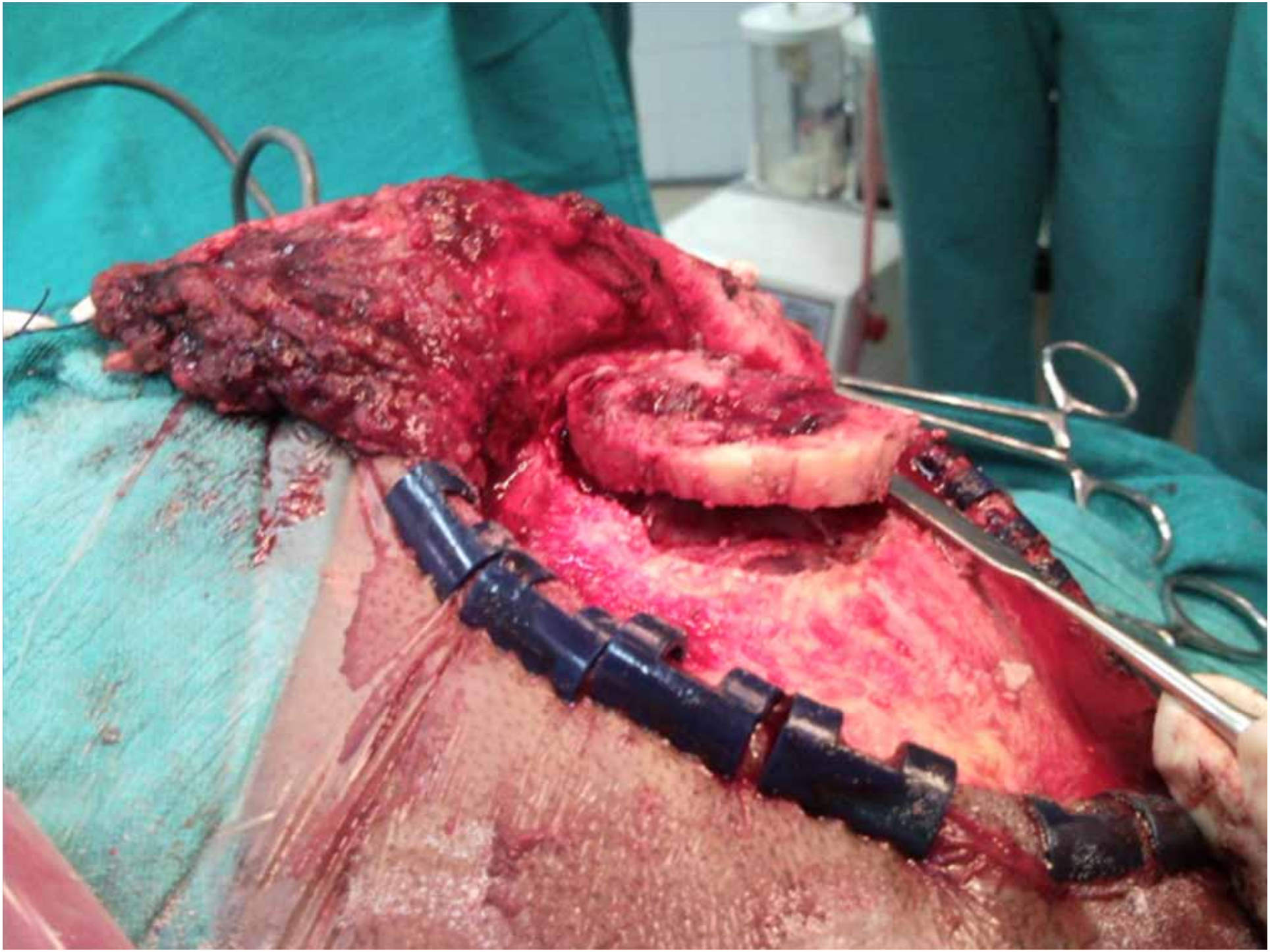
Left frontal craniotomy under general anaesthesia with excision of skull lesion

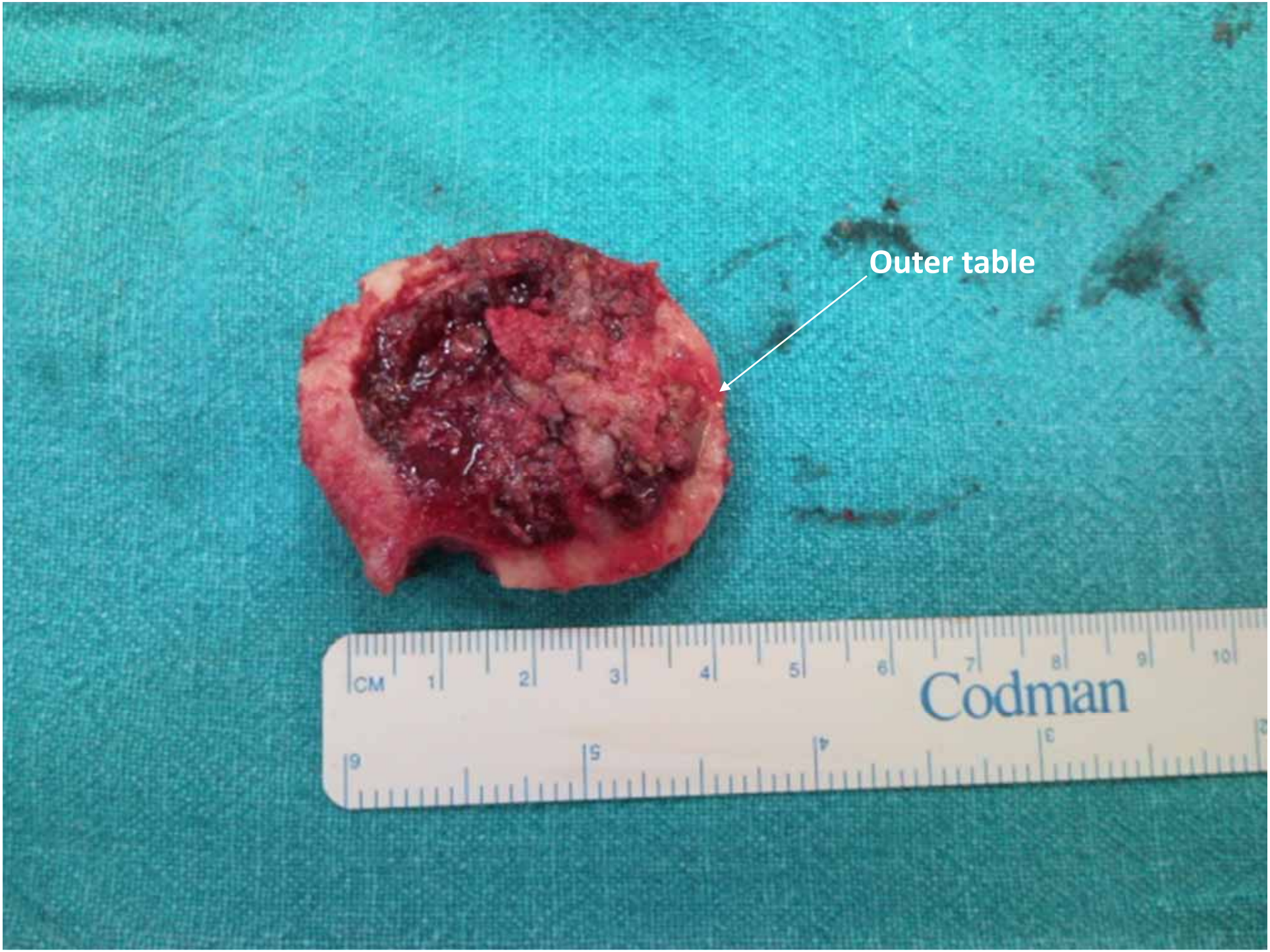

Temporalis muscle
Skull lesion

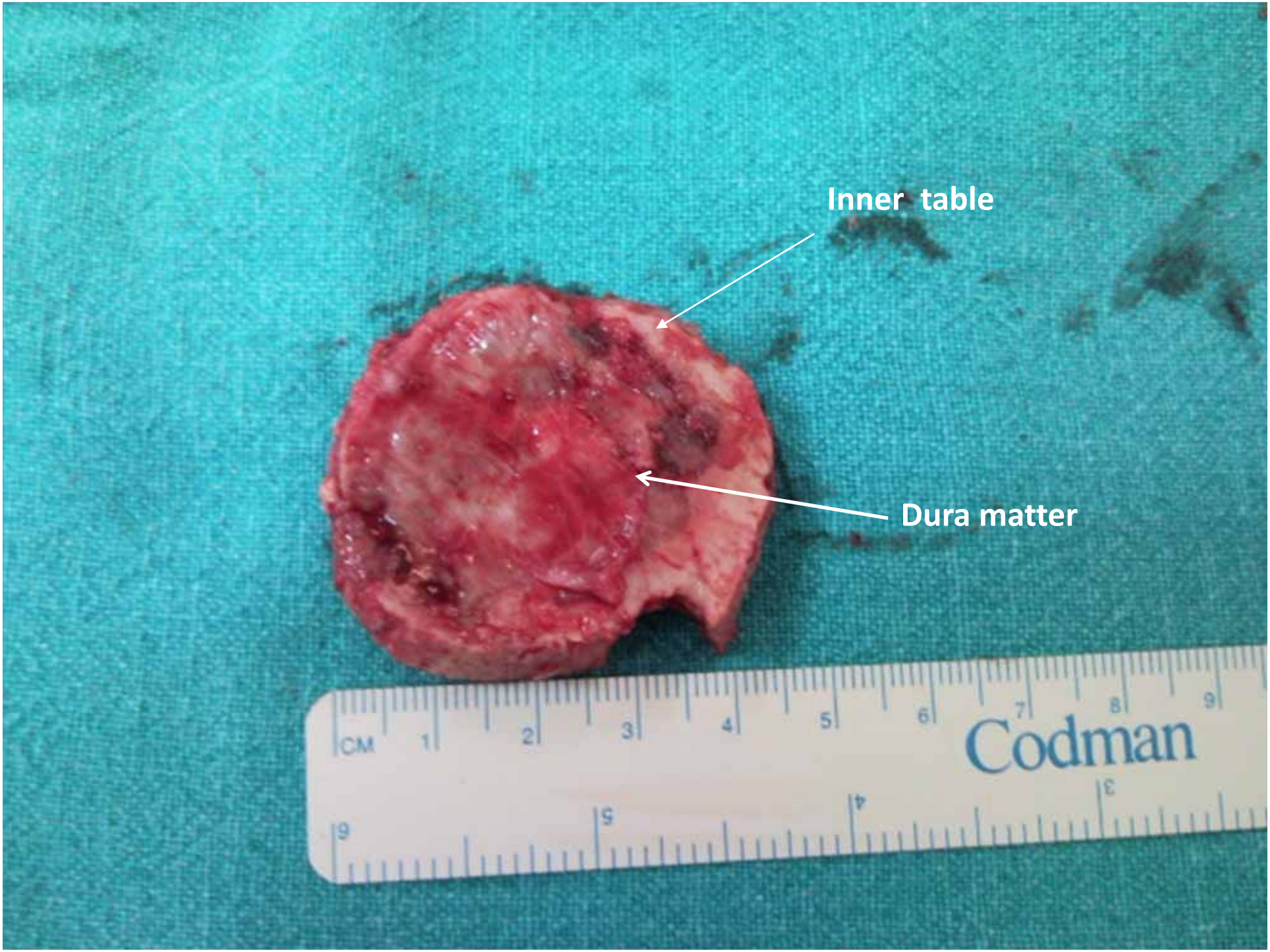

Tumour

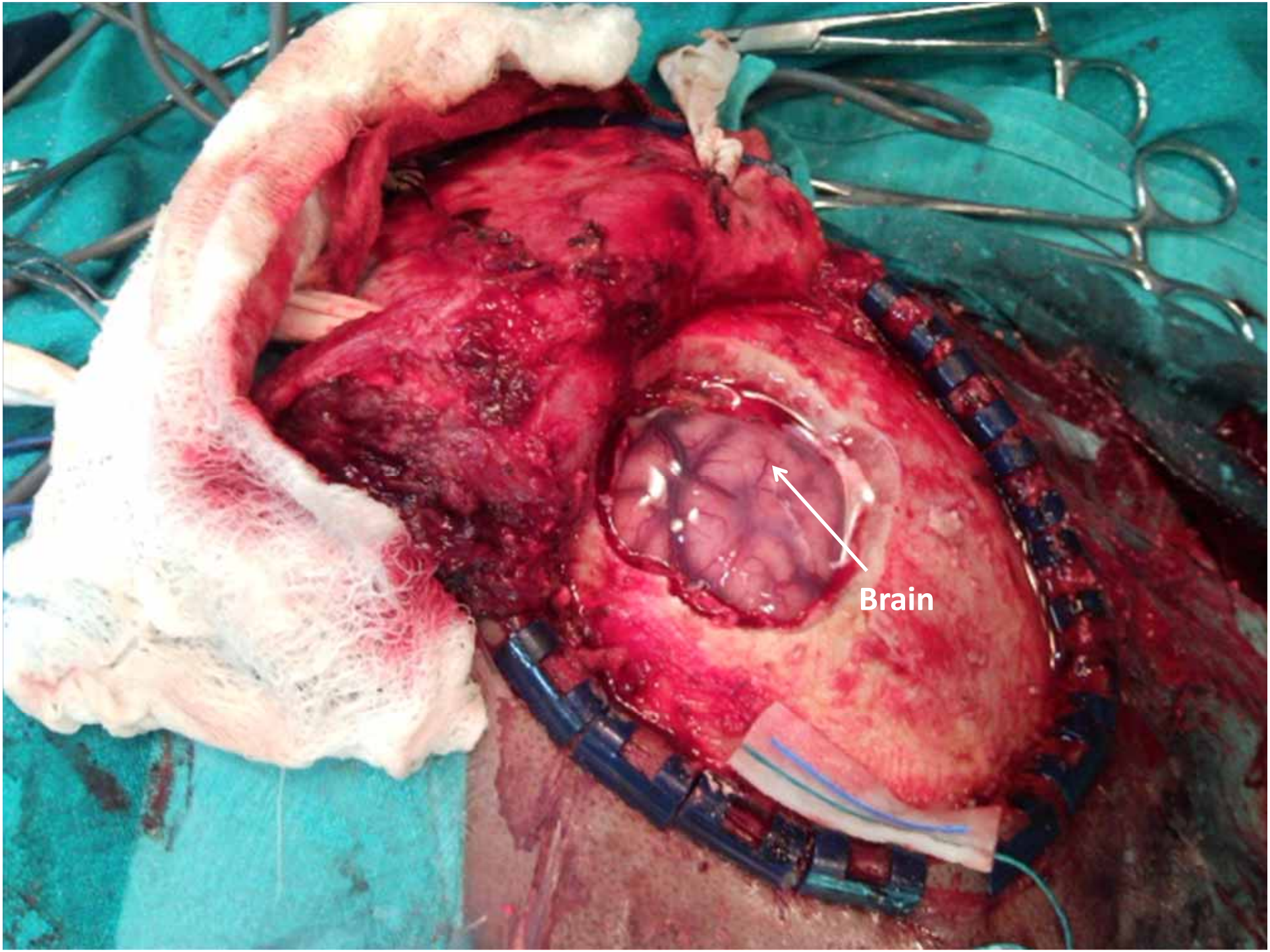

Burr hole

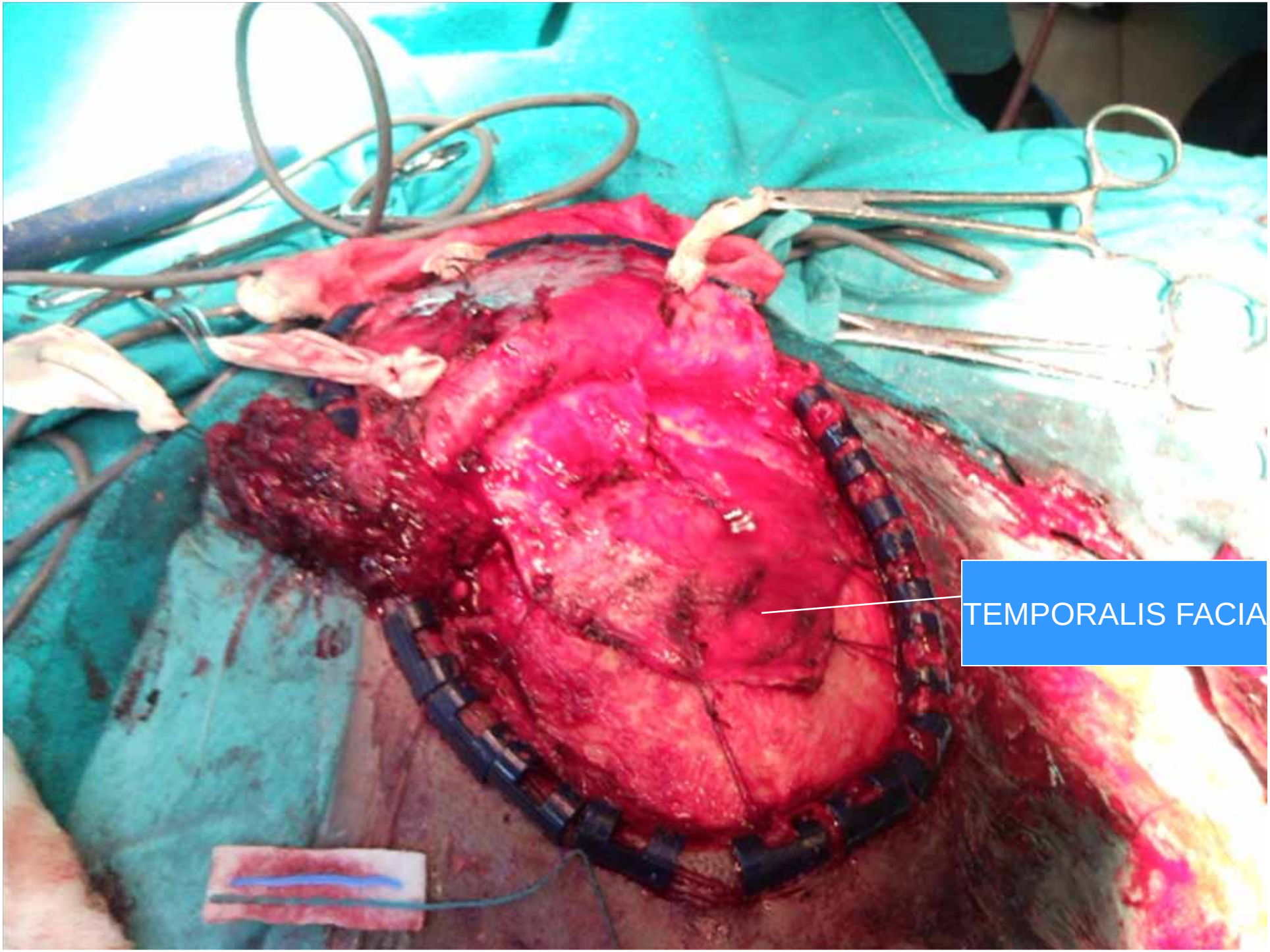





Outer table
CM
Codman


Inner table
Dura matter
CM
Codman


Brain


TEMPORALIS FACIA

Definition

Langerhans cell Histiocytosis (Histiocytosis X)

-a term that encompasses a spectrum of clinical conditions, ranging from a single, sometimes self limiting osteolytic bone lesion to a fulminant, disseminated process that may be fatal

Common feature

- A clonal proliferation of a histiocytic cell types known as the LANGERHANS CELL

Clinical entities

1. Hand Schuller- Christian disease

- Calvarial defects
- Exophthalmos
- Diabetes Insipidus

2. Letterer-Siwe disease

3. Eosinophilic granuloma

-acutely progressive course, fever, Pancytopenia, hepatomegaly, diffuse pulmonary, infiltrates and a cutaneous eruption

Prognosis

1. **Single bone-(monostotic granuloma)**

   → excellent Prognosis

2. More than one site but lesions limited to bone- polyostotic **ensinophilic**

   → good

3. **Multifocal + extra skeletal**

- Disseminated langerhans cell histiocytosis

Clinical Presentations

- **Monostotic eosinophilic granuloma**

- Local tenderness or a small mass lesion in a child
- Pathological Fracture
- Incidental findings

Polyostotic esoniphilic

- Pain, mass lessions over scalp
- Local effects at base of skull
- Diabetes Insipidus
- Proptosis
- Recurrent otitis media
- Pituitary hypothalamus dysfunction
- Ammenohorrea
- Growth retardation

Disseminated Langerhans Cell histiocytosis

- Fever, hepatosplenomegaly ,

Anemia, leukopenia, thrombocytopenia,

Purpura , lymphadenopathy ,

Skin macules and papules with osteolytic bone lesions

Radiological Features

Skull

- Osteolytic lesion without a sclerotic rim
- Complete absence of bony trabeculae
- Chest XRay

  - Reticulo nodular pattern

  -Honeycomb appearance
- CT

` -Low density lesion

Management

- Biopsy
- Observations
- Systemic multiagent chemotherapy
- Radiation
- Inteferon
- Cyclosporin
- Bone marrow transplantation


Thank you