

AN UNUSUAL CAUSE OF STROKE IN A MIDDLE AGED FEMALE

Presenting Author: SHINES MARIYA

SHAJI P Guide : DR. KRISHNAN

BALAGOPAL

MEDICINE & ALLIED SUBJECTS

Department of Neurology



INTRODUCTION

Essential Thrombocythemia (ET) is an acquired myeloproliferative disorder characterized by a sustained elevation of platelet number

with a tendency for thrombosis and hemorrhage .The prevalence

is approximately **30/100,000**.The median age at diagnosis is 65-70

years but the disease can occur at any age. The female to male

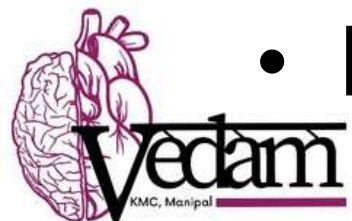
ratio is about 2:1. The clinical picture is dominated by a

occlusive events and
predisposition to vascular
hemorrhages.



CASE

- 50 year old female patient- no prior co morbidities**
- Y. Three month history of chest discomfort and exertional breathlessness**
- Evaluated locally- X ray chest done, ECG taken –reassured**
- On the day of admission, onset of dull aching headache from morning**
- One episode of generalized tonic clonic seizures followed by altered sensorium**
- Brought to ER with low GCS and respiratory difficulty**



EXAMINATION

- N** • **GCS-6/15**
- **HR-110/MIN**
- **BP-150/90mm, afebrile**
- **Tachypnoeic**
- **Left hemiparesis- Grade 2/5**
- **Gaze preference to right side**
- **Intubated and ventilated**
- **Shifted to Neurology ICU**

INVESTIGATION

- **S** • Blood reports- elevated platelet counts-**1178000/cu mm**
- Hemoglobin,TC,DC-normal
- ESR-normal
- Electrolytes, LFT,RFT –normal
- Serum iron, ferritin-normal
- ANA, Homocysteine, Protein C , Protein S , Anti Cardiolipin ,
Lupus anti coagulant-normal
- D dimer-elevated (>1000)

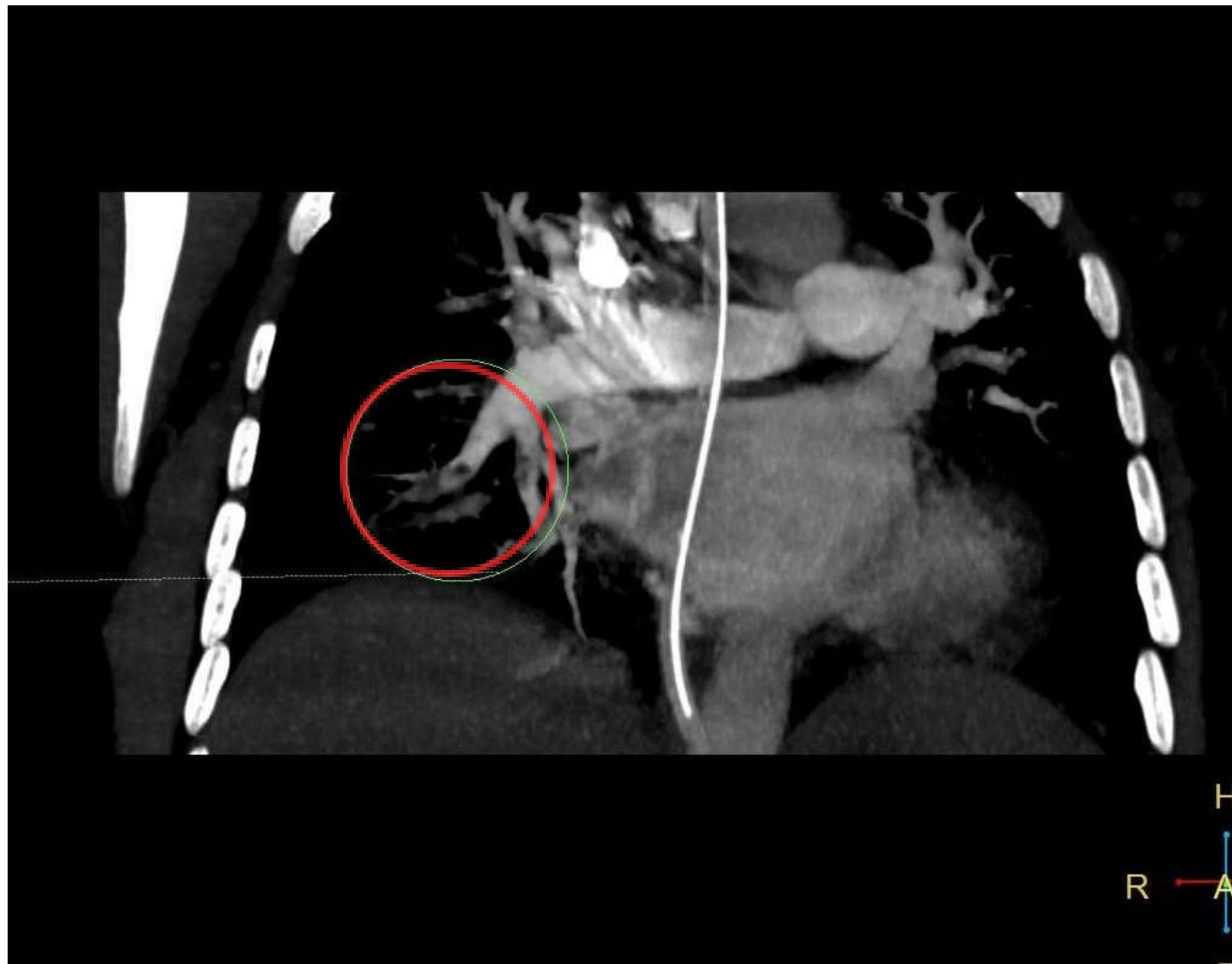
- **Chest X ray-normal**
- **ECG- sinus tachycardia**
- **Imaging-CT/MRI**
- **ECHO-Echocardiogram done showed dilatation of the Right Atrium and Right Ventricle with Tricuspid Regurgitation and severe**

Pulmonary Arterial Hypertension



CTPA

This showed multiple filling defects in the segmental branches
of the Right Interlobal arteries suggestive of a
Pulmonary segmental thromboembolism



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MRI done-DWI/ADC

Multiple infarcts in the Right Middle Cerebral Artery territory



MRA

Thrombotic occlusion of Right MCA M1 segment



COURS

E

- Hematology opinion was taken and the possibility of an **Essential Thrombocythemia (ET)** was considered

- A bone marrow examination was suggested but the family of the patient declined providing consent for the procedure

- Testing for Myeloproliferative Neoplasm gene alterations in peripheral blood revealed a 5 bp insertion in Exon 9 of

CALR(Calreticulin) gene

- **CALR- one of the mutations seen in ET-confirming diagnosis**

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(10/20/2021)

MANAGEMENT

I Cardiology/Neurology/Hematology consults taken.

- Started on Low Molecular Weight Heparin and transitioned to
ora

I anticoagulation with Warfarin with periodic monitoring .

- Double antiplatelet agents and anti convulsants were also started.
- Sildenafil was started in view of high Pulmonary Artery pressures.
- In view of high platelet counts and positive genetic mutation, she was started on **Hydroxyurea** with a view to reducing elevated platelet counts (cytoreductive therapy).

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COURS

- E** • **She was weaned off the ventilator and shifted to the ward.**
- After physiotherapy, her muscle strength improved.**
- Repeat platelet counts showed reduction in number.**
- She was discharged with regular follow up with both Neurology and Hematology Departments.**
- Last follow up , power-5/5, walking normally.**
- On oral anti coagulants and hydroxyurea- platelet counts normalized.**

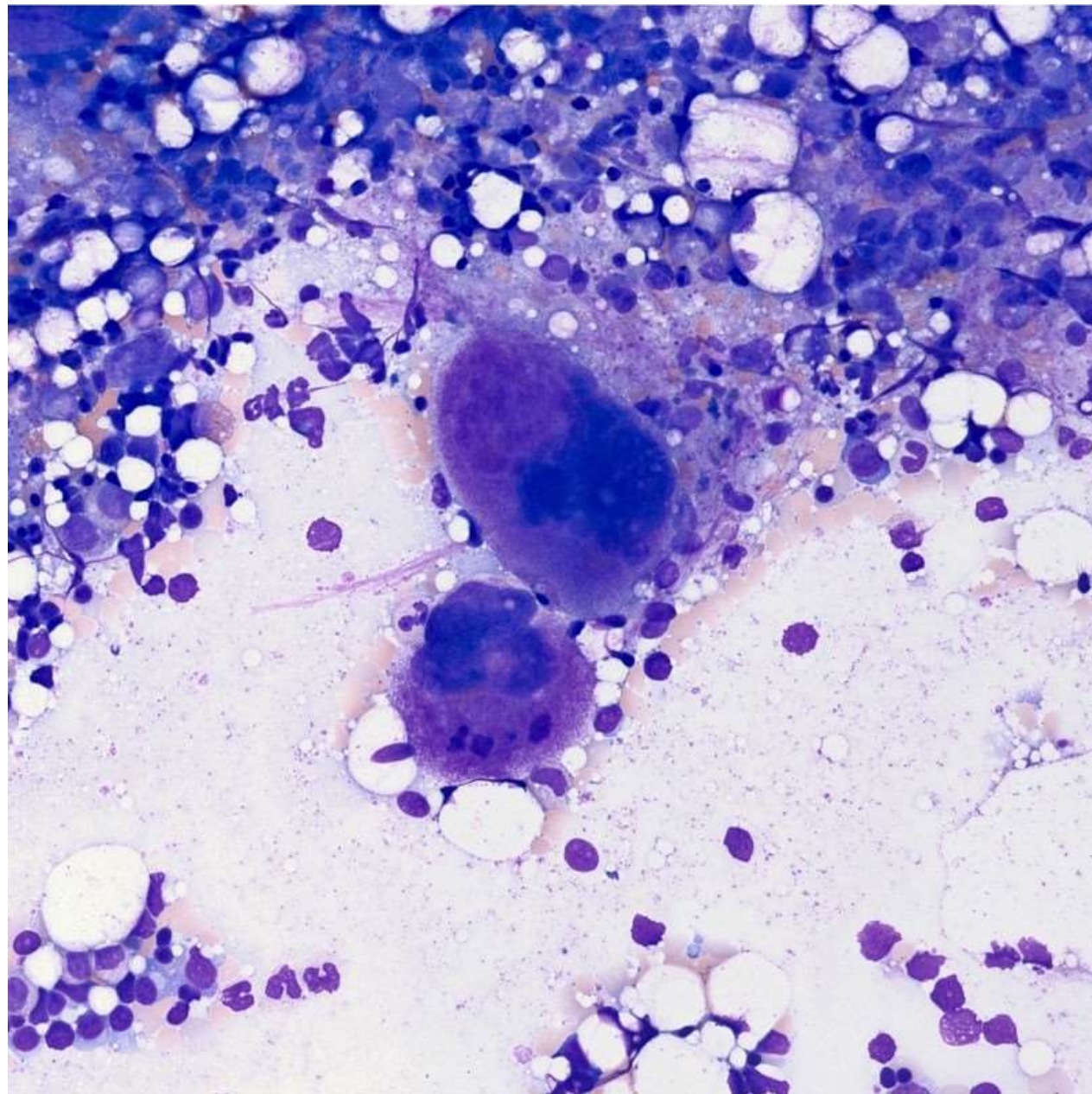
DISCUSSIO

N Essential Thrombocythemia is a **chronic myeloproliferative disorder** in which sustained megakaryocyte proliferation leads to an increase in the number of circulating platelets

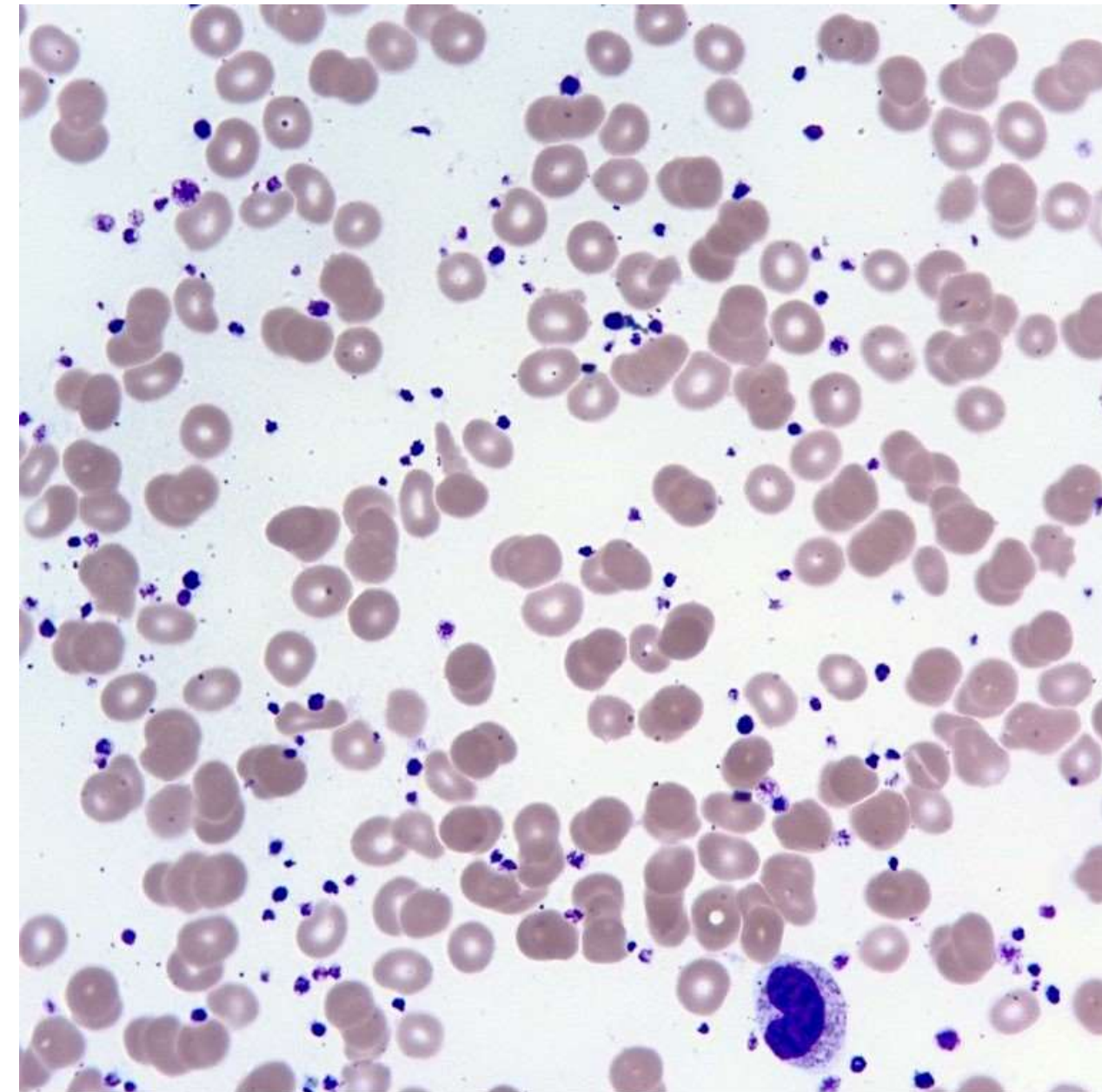
- Mutations in *JAK2*, *CALR*, or *MPL* -found in approximately 90% of patients with ET
- *JAK2* mutation is seen in approximately 50-60% of patients
- Somatic mutations in *CALR* -detected in peripheral blood in the approximately 25% of ET cases
- Most symptomatic patients present with symptoms that relate to small- or large- vessel thrombosis



- **Headache and stroke** - most common neurologic symptoms
- **Venous thrombosis of the splenic, hepatic, or leg and pelvic veins**
- **Pulmonary hypertension may result from pulmonary vasculature occlusion**
- **Bleeding complications seen -the gastrointestinal tract is the primary site of bleeding complications-functional platelet disorder**
- **CALR - Risk of thrombosis was twice as high in patients with JAK2 mutations than in those with CALR mutations**
- **Transformation to polycythemia was not observed in patients with CALR mutations -Better prognosis**



Bone marrow aspirate showing atypical megakaryocytes in a case of essential thrombocytosis. The atypical megakaryocytes are enlarged, with abundant mature cytoplasm and deeply lobed, hypersegmented (staghorn-like) nuclei.



Peripheral blood smear in a case of essential thrombocytosis showing marked thrombocytosis. The platelets show anisocytosis and bizarre forms.

Histopathology of Essential Thrombocythemia

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CONCLUSION

- N**• Essential Thrombocythemia is one of the causes of stroke and the presence of a **high platelet count** should alert the clinician to this possibility.
- Do not neglect to perform complete blood counts and investigate further if abnormal in all stroke cases .
- Genetic mutation analysis should be considered in **ALL** such cases
- There are very few case reports of patients of ET presenting with **both** stroke and pulmonary embolism in the same patient and this **is what we highlight in this case.**

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**THANK
YOU**