
بسم الله الرحمن الرحيم

- **Name:** Bashair Hashim Ali
- **Age:**46 years
- **Residence:** Mosul/ Al-Samah Q.
- **Occupation:** non-employee.
- **Date of admission:** 17-8-2013
- **Date of exam:** 17-8-2013

Chief complaint: passing of black coloured stool for few days before admission.

- **Hx of present illness:** the story of the patient started before 5 years when she suddenly developed spontaneous bleeding from her nose which is profuse and stopped with local measures then she developed recurrent bleeding attacks from her nose and she went to hospital ENT department for local nasal cautery but the condition worsen then she consult to physician who made the final diagnosis , before more than 1 year she developed episodes of passing of black colour stool which is liquid in consistency and offensive odor for about 3times /day not associated with vomiting but mild abdominal pain no loss of appetite no loss of weight.

- She has also generalized weakness, pallor of skin and mucus membrane with dizziness.
- There were no other bleeding tendency , and normal gynecological period with usual bleeding.

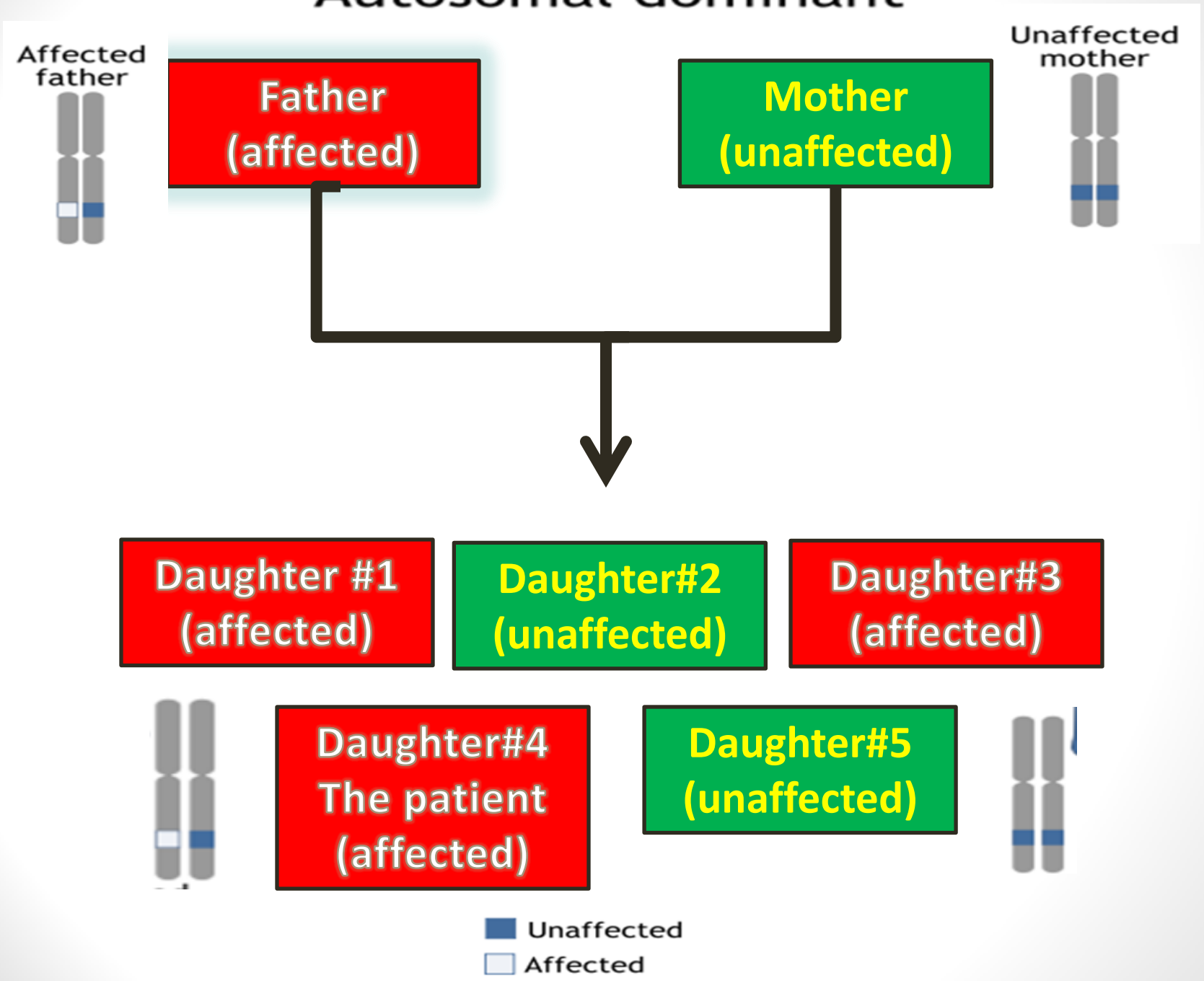
Review of systems:

- **GIT:** no loss of appetite, no vomiting, no hematemesis, chronic diarrhea passing of black color stool 3times/day.
- **CVS**: palpitation, no ankle swelling , mild chest pain , dyspnea on exertion and no orthopnea
- **Respiratory system**: S.O.B. on exertion ,mild cough with little whitish sputum, no hemoptysis, no chest pain.
- **G.U.S:** no hematuria, good urine output ,no dysuria or frequency, no loin pain.
- **CNS:** no fit, normal gait.
- **M. S.S:** nothing significant.

Past history:

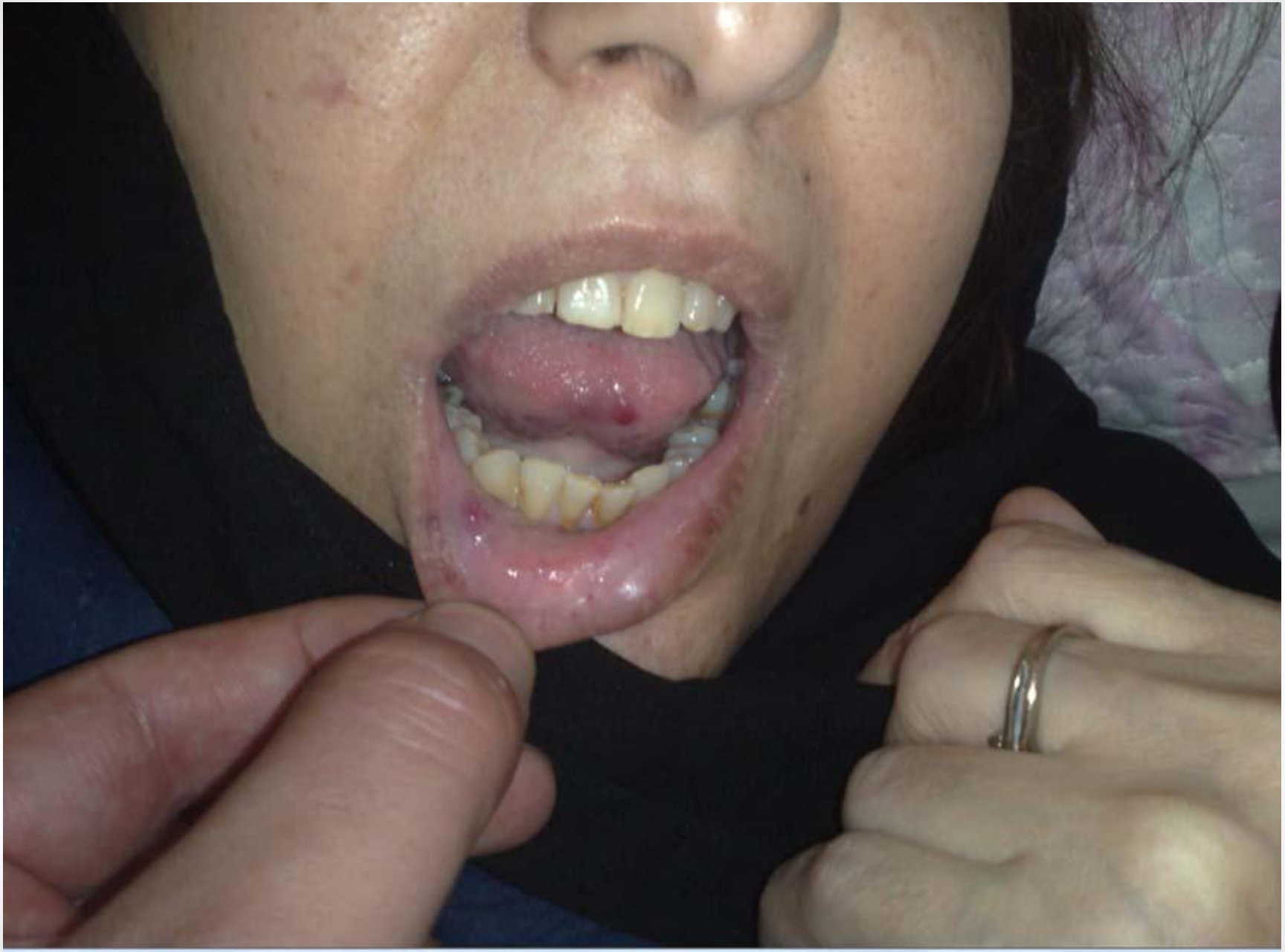
- **PMHx** diagnosed as Hereditary Hemorrhagic telangiectasia before 5 years on occasional oral and parental tonics(Iron and multivitamins)
- **PSHx**: endoscopy and laser coagulation for GIT.
- **Social hx:** Single, neither smoker nor alcoholic .
- **Family hx**: Her father and her 2 sisters have the same disease
- **Drug Hx**: No drug allergy

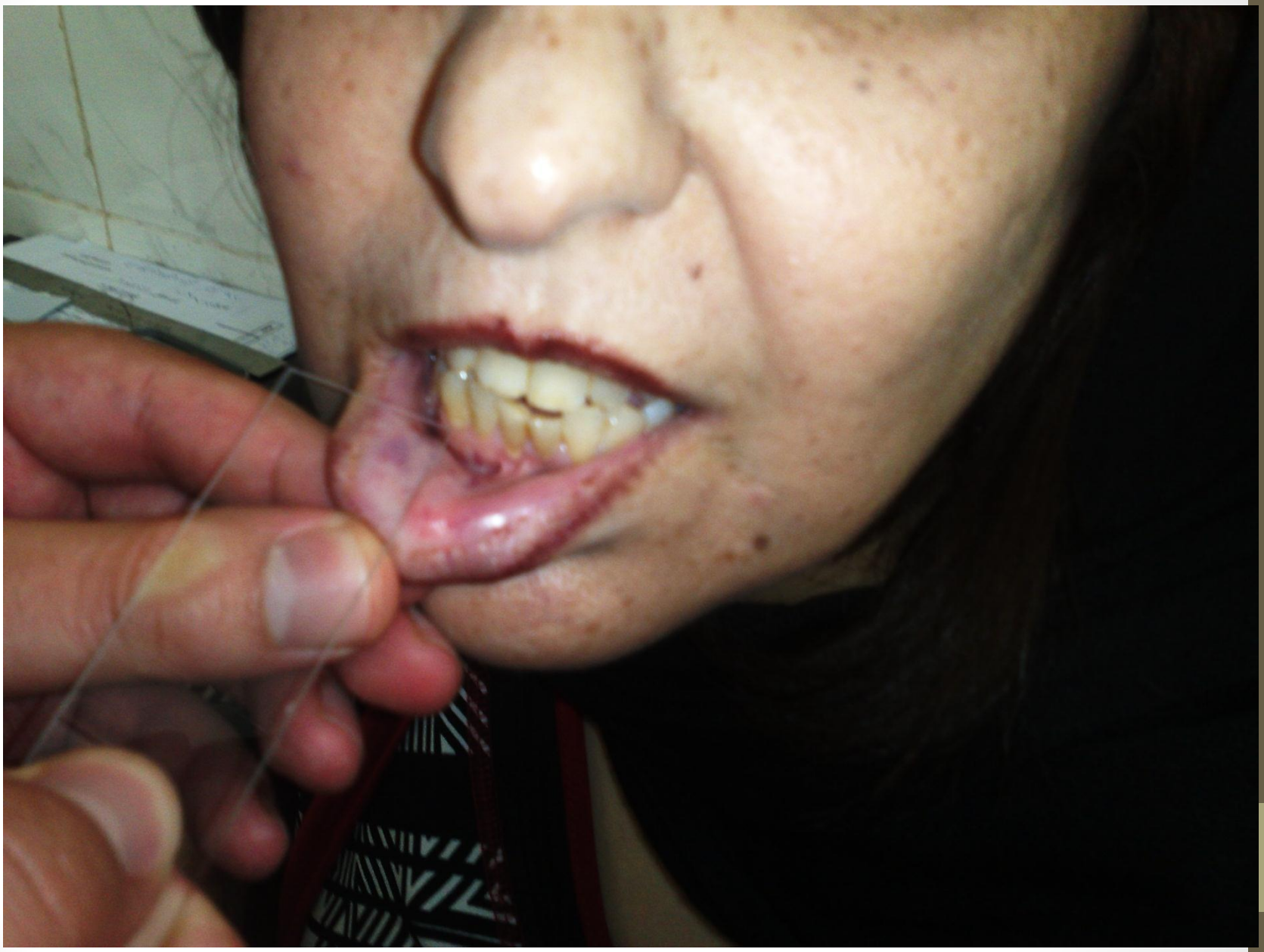
Autosomal dominant

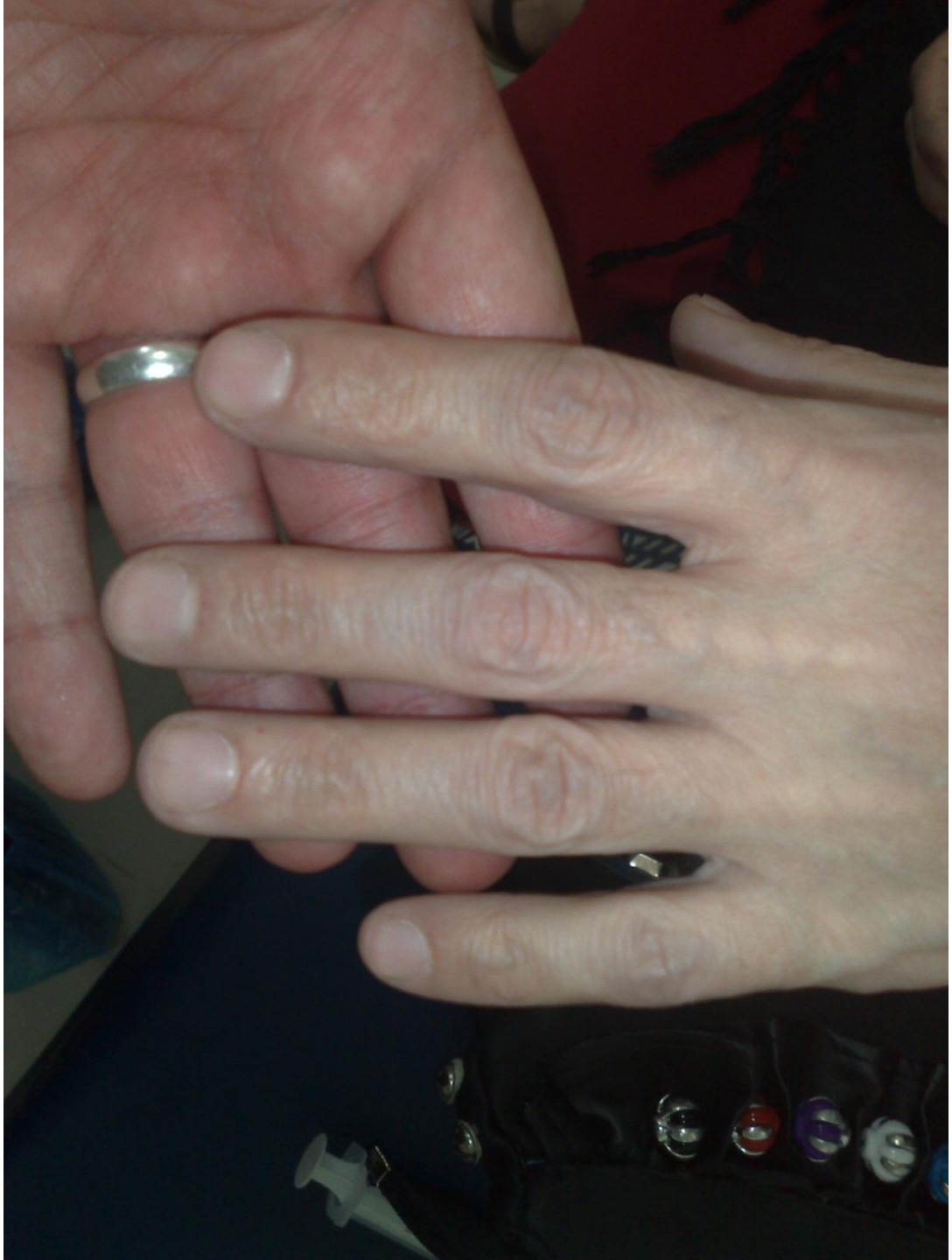


General examination:

- Middle aged female sitting in bed ,conscious, afebrile, not dyspnic, no cyanosis, pale, not jaundiced, normal hair distribution , no leg edema and no lymphadenopathy, no petechea or bruises but with telangictasia (small vascular malformations) in the skin of face and trunk and mucosal linings of the lips and tongue .
- There is no clubbing and no other lesions on her hands.







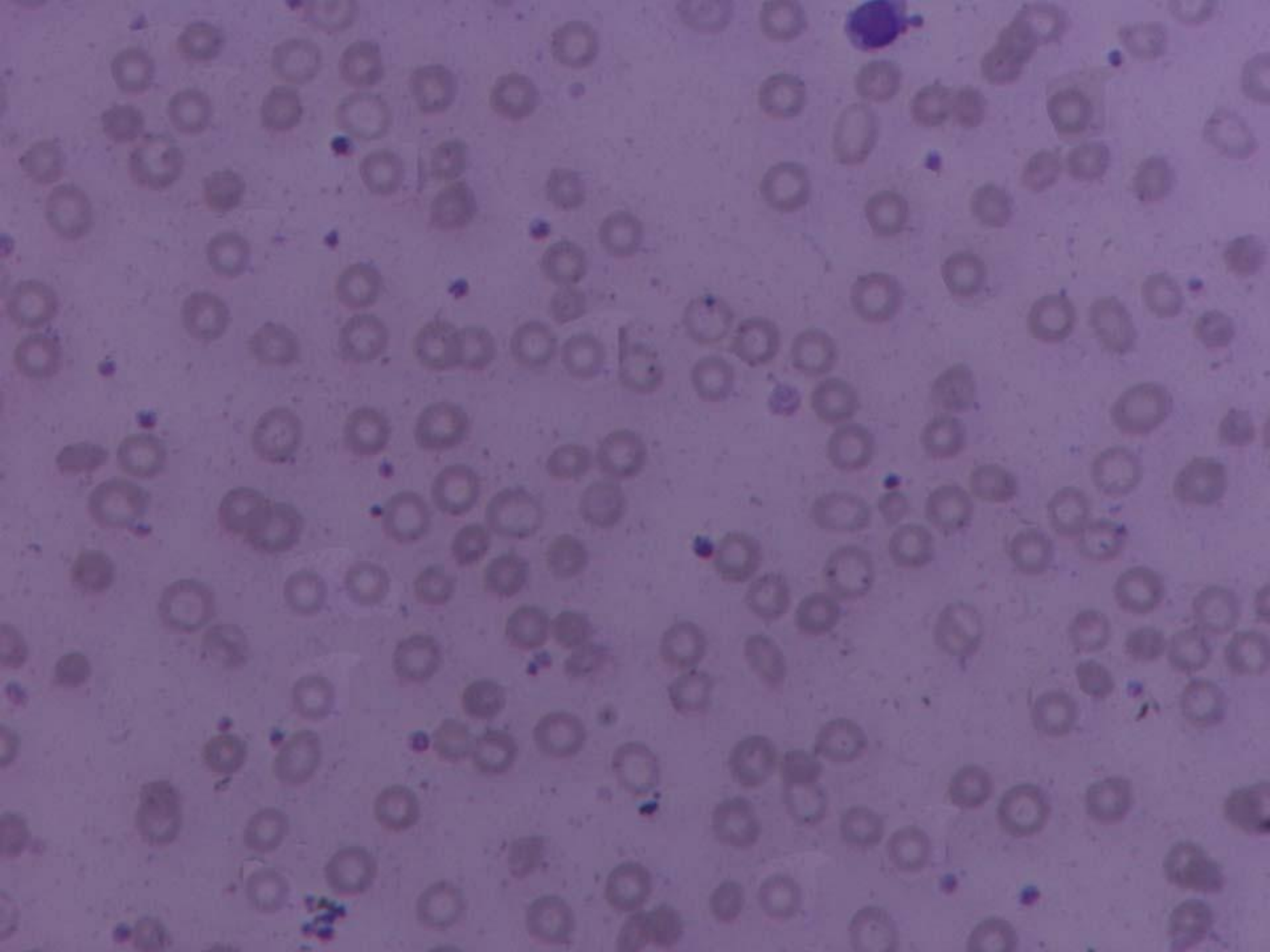
Vital Signs:

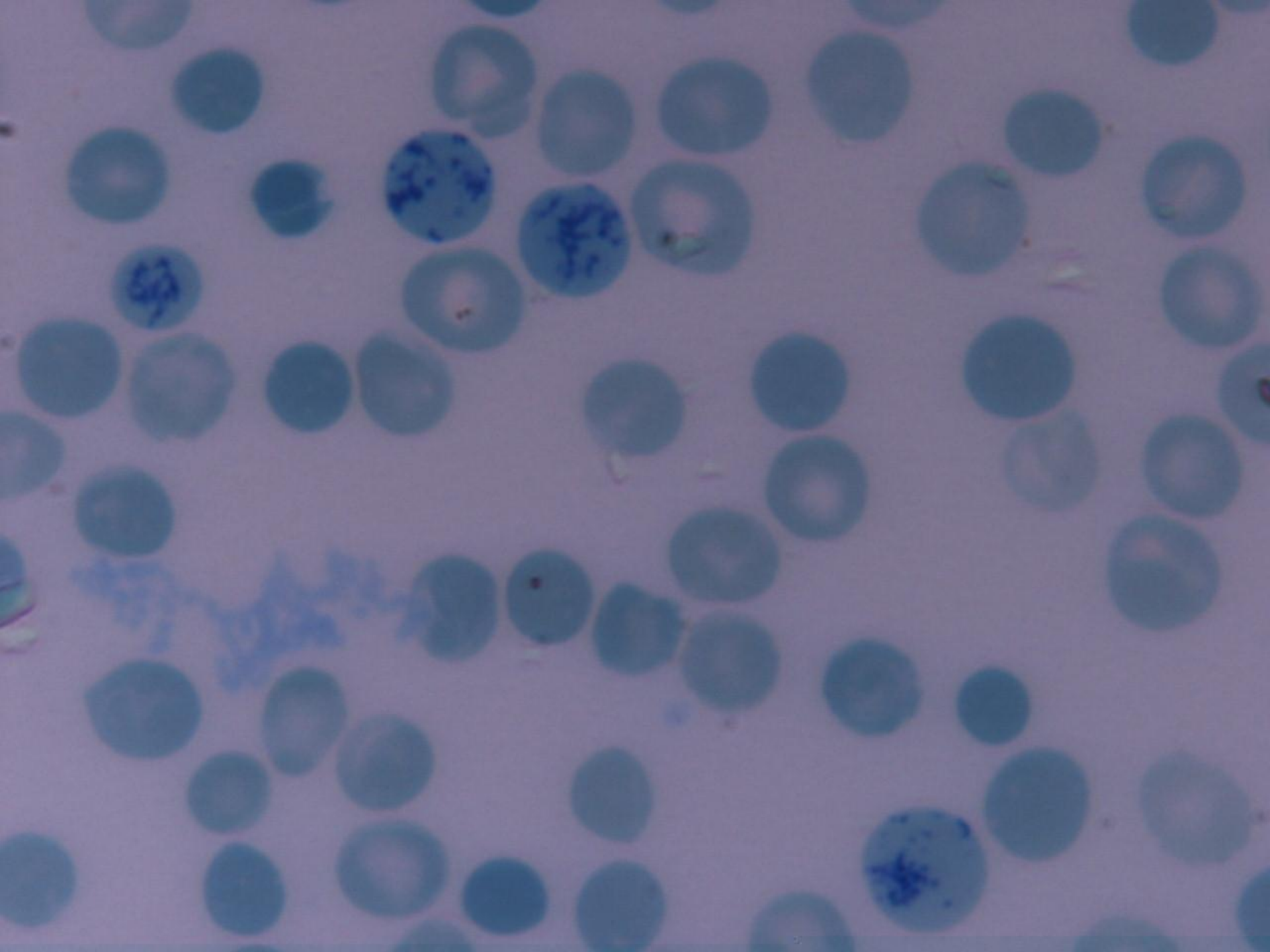
- **Temp**: 37.7^{o}C
- **RR:** 16/min
- **PR:** 94 b/m
- **BP:** 115/75 mmHg

- **Chest:** normal VB, clear . **Heart**: NDR.
- **Abdominal examination:** *Inspect:* ; no distend abdomen, move normal with respiration, invert umbilicus. No dilated veins.
- *palpation:* soft abdomen, No organomegaly.
- *Auscultation:* normal bowel sound.
- **Rectal examination***:* not done

CBC :(17-8-2013):

	N.V.		• N.V.
Hb: 41 g/l	♀135± 15g/l	WBC: 4.5 *10^9/L	4.0 – 10.0*10^9/L
Hct: 0.14 l/l	♀0.41±0.05l/l	Neutrophils: 71.9 %	40 -80 %
RBC: 1.79 *10^{12}/L	♀4.3±0.5*10^{12}/L	Lymphocytes: 19.6%	20 -40 %
MCV: 68 fl	92 ± 9 fl	Monocytes: 5.5%	2 -10 %
MCH: 17.3 pg	29.5 ± 2.5 pg	Eosinophil: 1.8%	1 – 6%
MCHC: 252 g/l	330 ± 15 g/l	Basophil: 1.2%	<1%
Retics : 10 %	0.5 – 2.5 %	RDW : 19.5%	11.5-14%
Platelets: 230 *10^9/L	280 ± 130 *10^9/L	ESR: - mm/1st hr	<20 mm

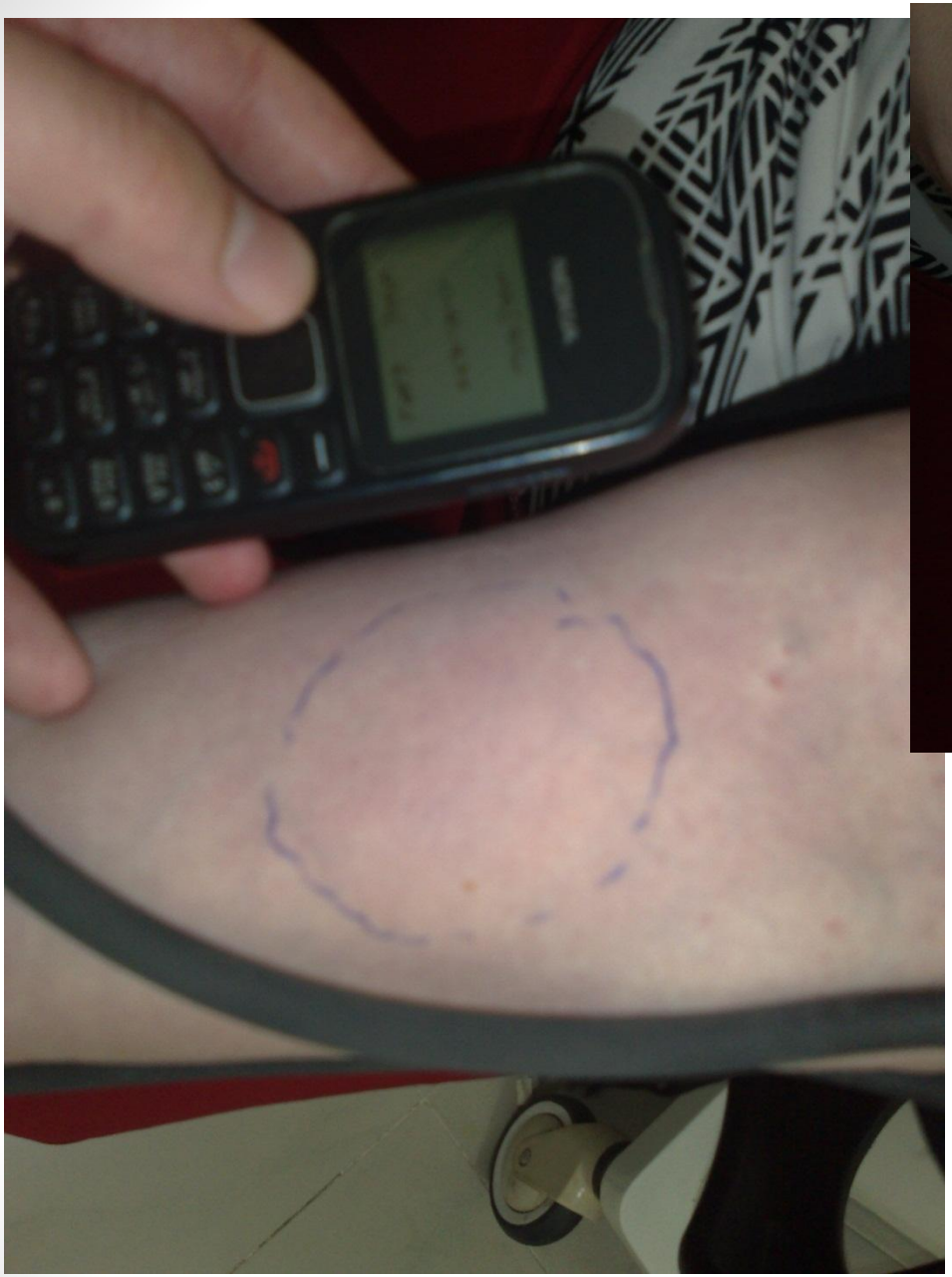


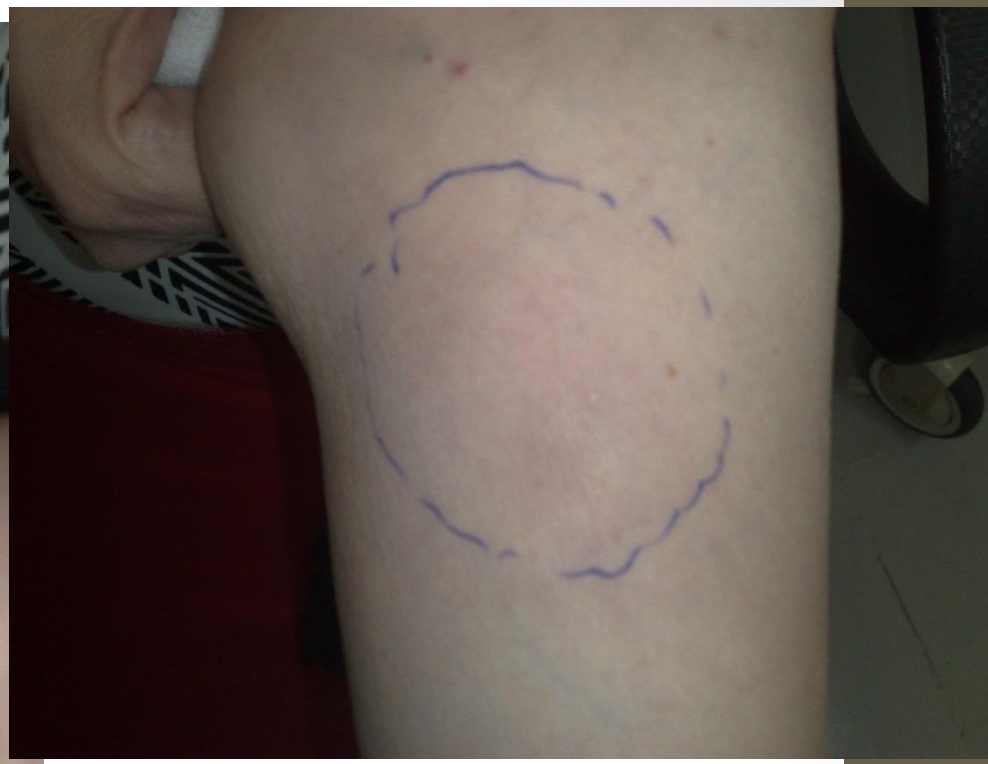


TEST	N.V.
S. Iron: 30 µg /dl	♀ 50-170 µg /dl
S. Ferritin: 2.12 ng/ ml	♀ 13-150 ng/ml
Total serum bilirubin: 11 µmol/L	< 17 µmol/L
S. A.L.T.: 12 u/L	♀ upto 31/L
S. A.S.T.: 18 u/L	♀ upto 32/L
S. Alkaline phosphatase 81 u/L	♀ 35 – 104 u/L
B. Urea: 4.5 mmol/L	3.3 -7.5 mmol/L
S. Creatinine: 61 µmol/L	♀ (53 – 99 µmol/L)
Blood group	A +ve

Haemostatic tests:

- **Bleeding time (Ivy's):** 4.5 min (N.V. 2 -7 min)
- **Hess test:** Normal
- **Prothrombin time:** 14.0 sec C:13.5 sec
- **aPTT:** 32 sec. C: 32 sec.





CBC :(23-9-2013):

	N.V.		• N.V.
Hb: 76 g/l	♀135± 15g/l	WBC: 3.9 *10^9/L	4.0 – 10.0*10^9/L
Hct: 0.26 l/l	♀0.41±0.05l/l	Neutrophils: 70.1 %	40 -80 %
RBC: 2.86 *10^{12}/L	♀4.3±0.5*10^{12}/L	Lymphocytes: 21.3%	20 -40 %
MCV: 92 fl	92 ± 9 fl	Monocytes: 5.4%	2 -10 %
MCH: 26.6 pg	29.5 ± 2.5 pg	Eosinophil: 2.3%	1 – 6%
MCHC: 287 g/l	330 ± 15 g/l	Basophil: 0.9%	<1%
Retics : 7.5 %	0.5 – 2.5 %	RDW : 16.7%	11.5-14%
Platelets: 150 *10^9/L	280 ± 130 *10^9/L	ESR: 12 mm/1st hr	<20 mm

- **<u>Ultrasound of the abdomen:</u>**
- Normal liver size and echogenicity ,normal biliary passages and portal veins
- Normal GB no stones.
- Both kidneys are normal size and echogenicity ,no stones .
- Normal pancreases
- Normal spleen
- Normal urinary bladder
- No masses or enlarged L.N.

- **<u>Serological tests for HIV & Hepatitis:</u>**
- **HBsAg:** Negative
- **Anti HCV Test:** Negative
- **HIV 1&2** : Negative

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Ministry of Health
D.O.H Suleiman

KCGH

Kurdistan Center for
Gastroenterology & Hepatology
OGD/NBI

Name: Bashaer Hashm Ali | Age: 46 years | Gender: ♀
Date: 16/10/2011 | Ref. by: Dr. *Muhammad Yasin* | Tel: 0770[redacted]5253699

Clinical data: Case of Hereditary Hemorrhagic telangiectasia with repeated GIT Bleeding presented for Argon plasma coagulation

Premedication: Pethidine: 50 mg, Midazolam:5 mg

Endoscopic Findings:

Esophagus: Normal mucosa, no varix
GEJ: at 40cm, Lax cardia

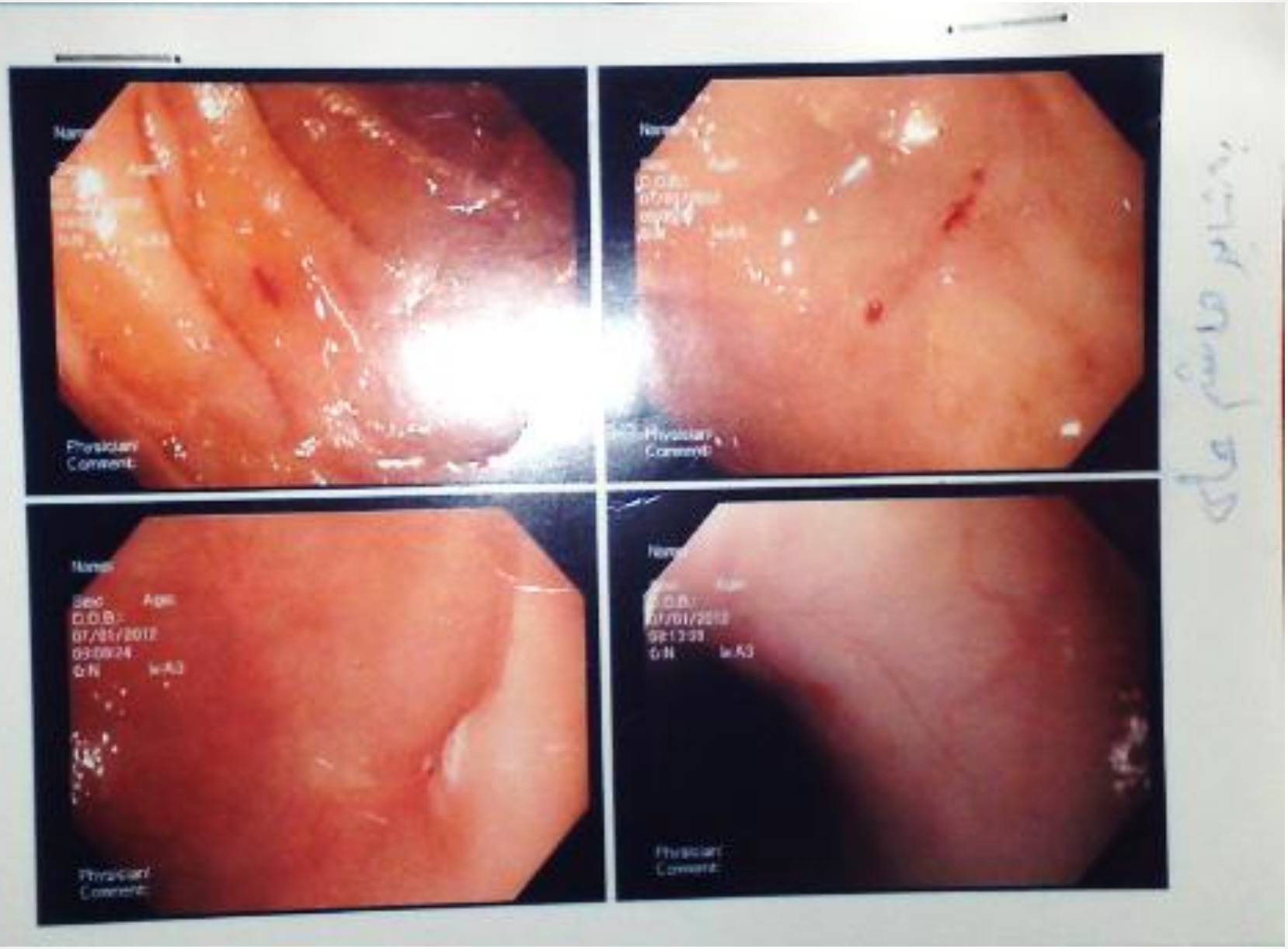
Stomach: Normal gastric mucosa and folds
Multiple variable size of Telangiectasias (AVM) with stigmata of recent bleeding, argon plasma coagulation applied
Single 10mm sessile polyp in the gastric body, polypectomy done by hot snare

Duodenum:
D1: Normal
D2: Normal

Conclusion: HHT
First session of ARP done

Recommendation: please for follow up endoscopy and second session after one month

Name & signature of Endoscopist: Dr.Taha Alkarbuli, Dr.Hiwa Abubakr
Assistance: Abdullah & Sarwar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D.O.H Sulaimani — Colonoscopy /NBI

Name: Basharr Hashm Ali	Age: 46 years	Gender: ♀
Date: 16/10/2011	Ref. by: Dr. Muhammad Yasin	Tel: 07705253699

Clinical data: Case of Hereditary Hemorrhagic telangiectasia with repeated GIT Bleeding presented for Argon plasma coagulation

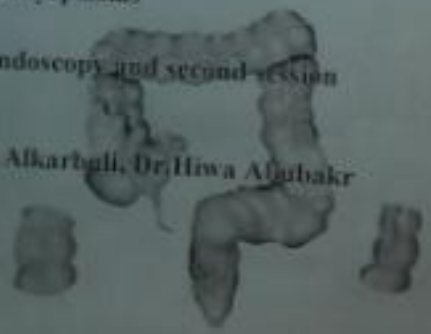
Premedication: Pethidine 50 mg

Anal inspection Normal

DRE: Normal

Preparation: Fair preparation

COLONOSCOPY: Complete colonoscopy up to cecum revealed normal vascular pattern
Multiple angiodyplasia seen in the cecum, ascending colon and transverse colon APC done

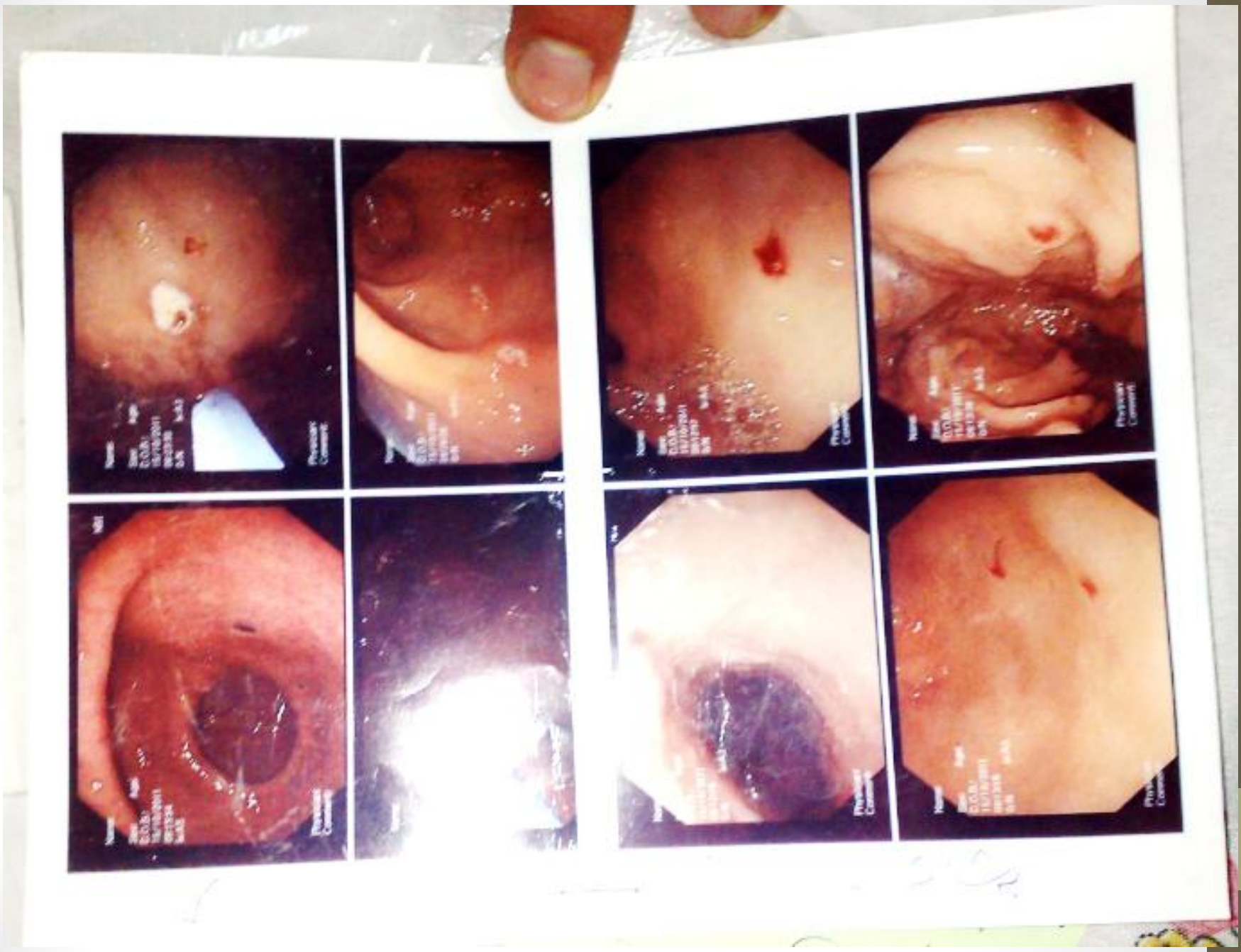


Conclusion: HHT, Multiple Colonic Angiodysplasias
APC done

Recommendation: please for follow up endoscopy and second session after one month

Name & signature of Endoscopist: Dr.Taha Alkarboli, Dr.Hiwa Abubakr

Assistance: Abdulla & Sarwar



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Name: Bashaer Hashm Ali
Date: 29/10/2011

Age: 46 years
Ref. by: Dr. Muhammad Yasin

Gender: ♀
Tel: 07700525369

Clinical data: Case of Hereditary Hemorrhagic telangiectasia with repeated GIT Bleeding presented for SECOND SESSION OF Argon plasma coagulation to obliterate the gastro duodenal angioectasias

Premedication: Pethidine: 50 mg, Midazolam:5 mg

Endoscopic Findings:

Esophagus: Multiple small angioectasias involving esophageal wall
GEJ: at 40cm, Lax cardia

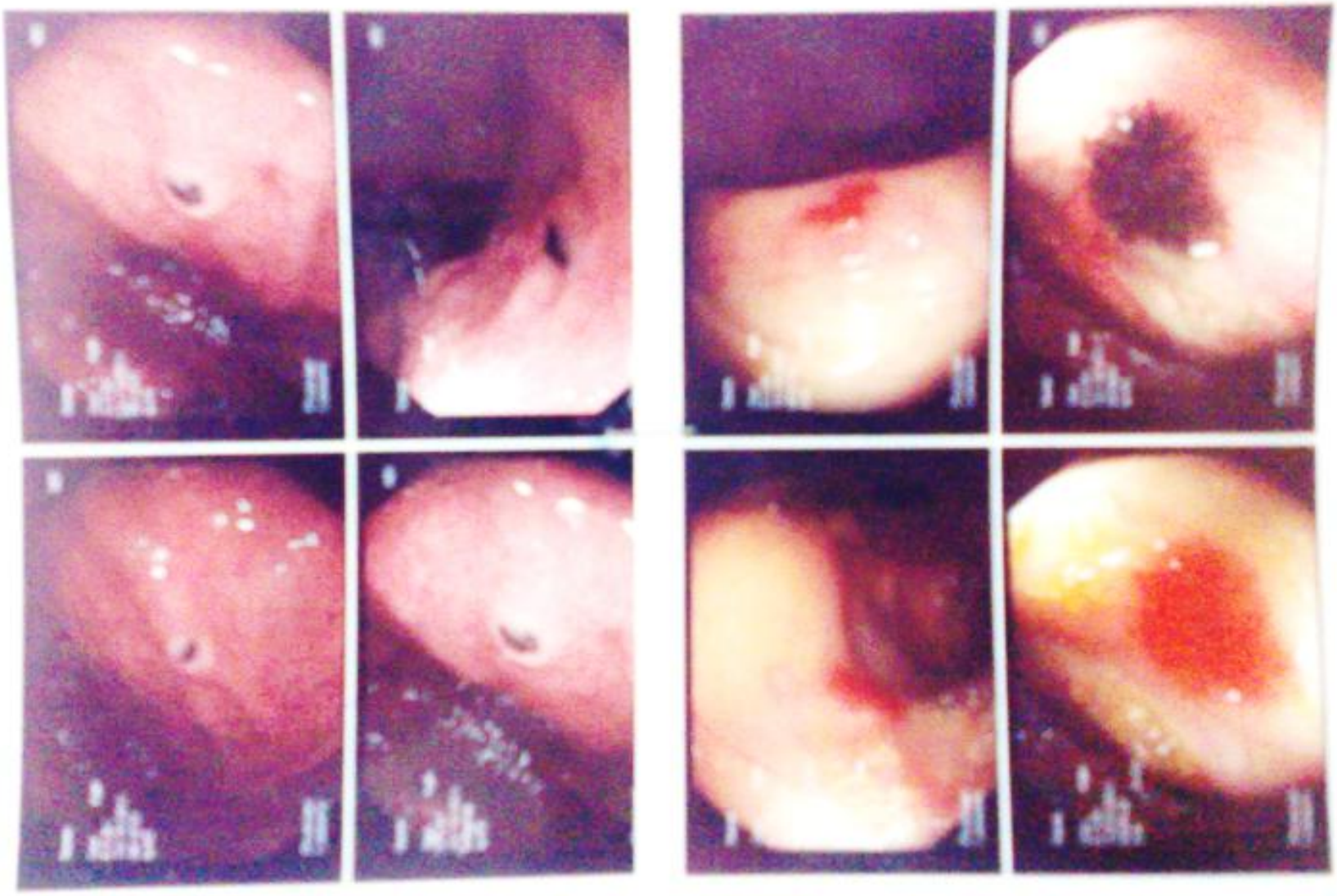
Stomach: Normal gastric mucosa and folds
Multiple variable sizes of Angioectasias (AVM) with stigmata of recent bleeding, second session of argon plasma coagulation applied
Scar and ulcer seen at the site of previous polypectomy and Angioectasias obliteration

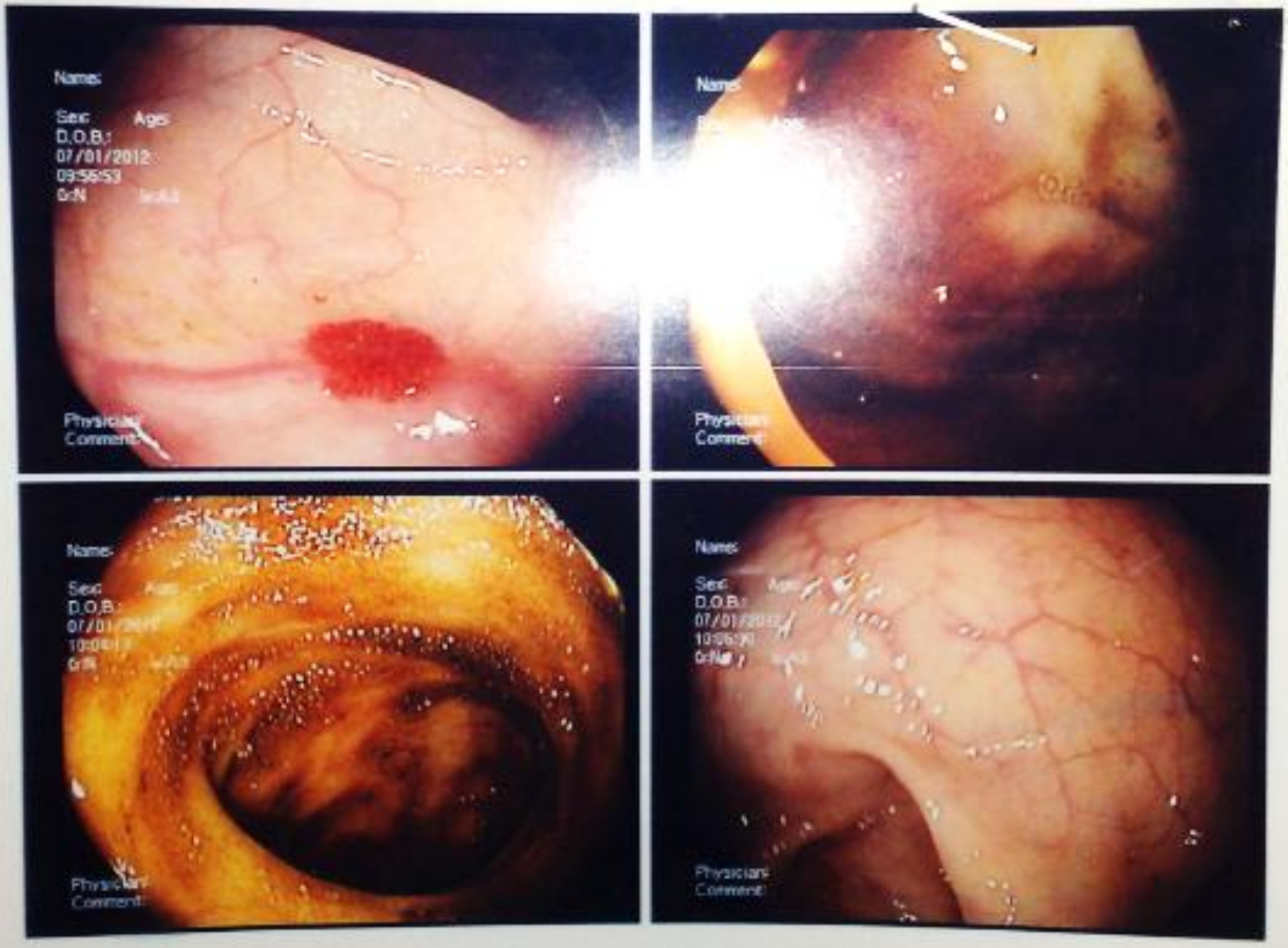
Duodenum:
Multiple Angioectasias of duodenal mucosa, obliteration done by APC

Conclusion: Hereditary Hemorrhagic Telangiectasia
Second session of ARP done to obliterate Gastro duodenal AVM

Recommendation: Please for follow up endoscopy with arrangement of Enteroscopy and second session Colonoscopy

Name & signature of Endoscopist: Dr.Taha Alkarbuli, Dr.Hiwa Abubakr
Assistance: Abdullah & Sarwar

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OGD/NBI

Name: Bashair Hashm Ali
Date: 07/01/ 2012

Age: 46 years
Ref. by: Dr. *Hiwa Abubakr*

Gender: ♀
Tel: 07700525369 99

Clinical data: Case of Hereditary Hemorrhagic telangiectasia presented with recurrent attacks of melena and bleeding per rectum, first session of ablation done by APC, Presented for Second session

Endoscopic Findings:

Esophagus: Normal mucosa, two small Angioectasia seen

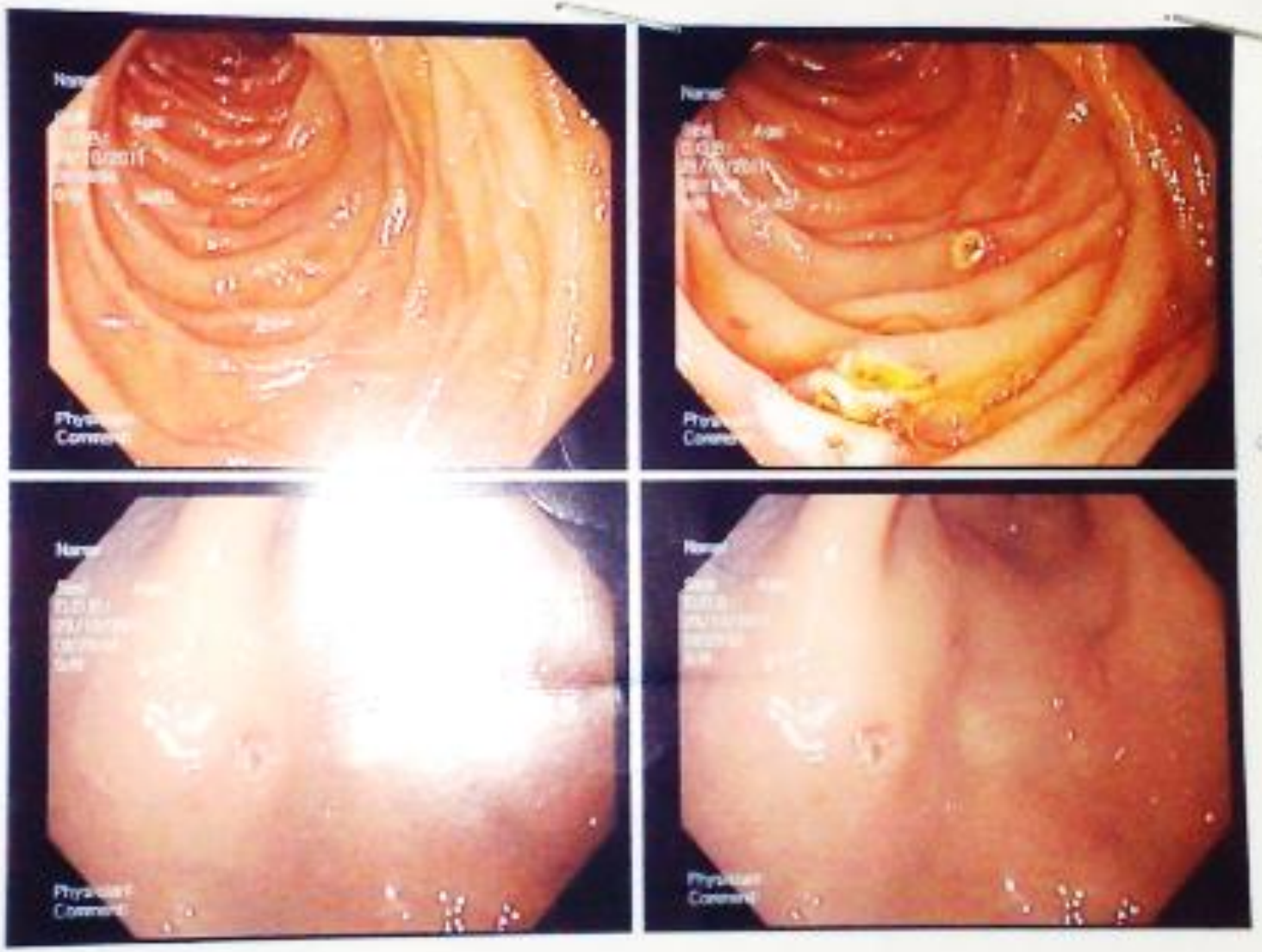
Stomach: Multiple variable size vascular ectasias seen, ablation done by Argon plasma coagulation

Duodenum:
D1: Normal
D2: Normal

Conclusion: HHT, Gastric vascular ectasias
Ablation done by APC

Recommendation: For follow up please with starting Treatment with Thalidomide 200mg daily and long acting Octreotide analogue

Name & signature of Endoscopist: Dr.Hiwa Abubakr
Assistance: Abdullah & Sarwar



Hereditary Hemorrhagic Telangiectasia

- also known as Osler-Weber-Rendu syndrome.
- Onset of disease
  - 50% diagnosed by age of 16years , 90% by age of 40 years.
- It is an autosomal dominant disorder, high penetrance, characterized by multiple telangiectatic lesions involving the skin and mucous membranes associated with epistaxis and other bleeding complications.
- HHT :prevalence of 1 in 8,000 in other members of a family with HHT (USA"Wintrob") .
- Mutations in the endoglin gene on chromosome 9 (HHT1) or in the activin receptor–like kinase gene (ALK1) on chromosome 12 (HHT2) account for most of the cases. These genes encode proteins that are involved in signalling by the transforming growth factor (TGF) – β which affect VEGF.

Pathophysiology:

- Ultrastructural analysis of cutaneous HHT lesions suggests that post-capillary venule dilation is the earliest identifiable morphologic abnormality.
- As the venules enlarge, they become convoluted and interconnect with arterioles through capillary segments. The capillary segments eventually disappear, and direct arteriolar-venular communications are established
- An infiltrate of mononuclear cells appears in the perivascular region of the HHT lesions.
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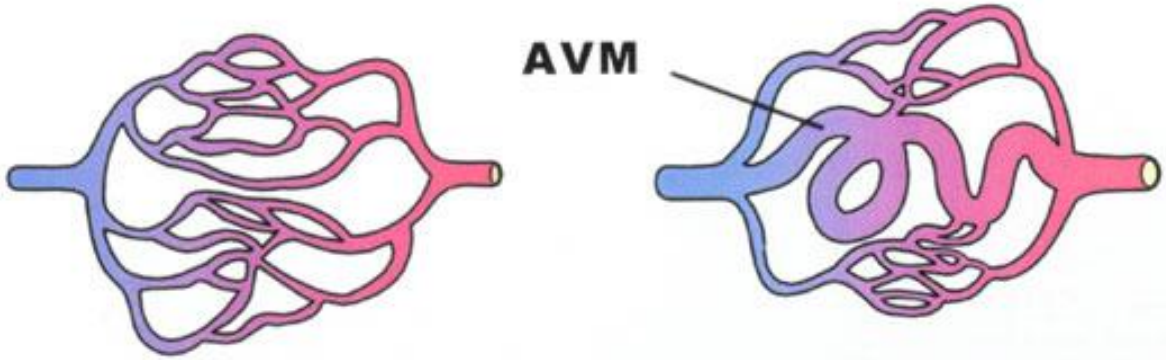


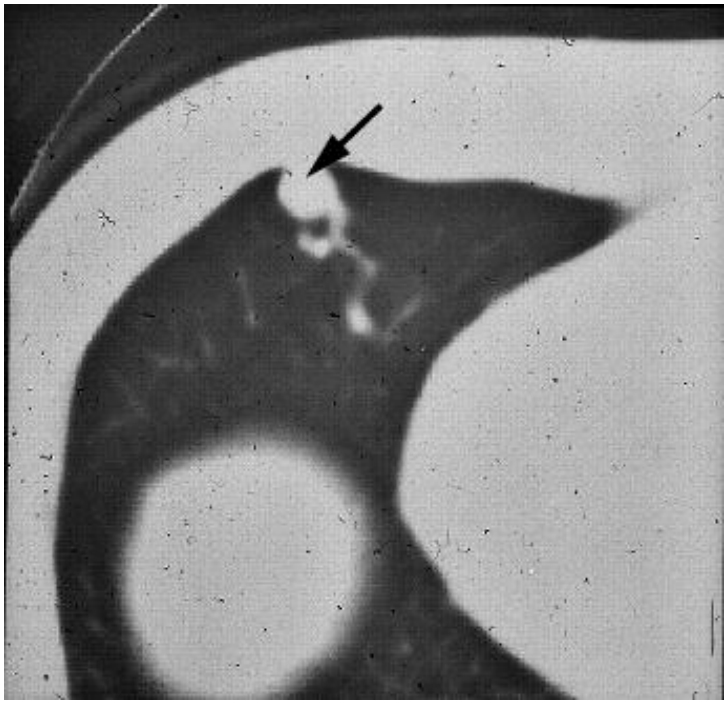
- The bleeding manifestations are thought to occur because of mechanical fragility of these vessels. Common abnormalities in the hemostatic system do not seem to represent a major factor in the underlying bleeding tendency. HHT patients manifest a variety of other complications including shunting, emboli, and thrombosis.

Clinical Manifestations

- The cutaneous lesions usually appear in affected persons by 40 years of age, and they increase in number with age. The lesions measure 1 to3 mm in diameter and are sharply demarcated in appearance .They blanch with pressure, but the blanching may be incomplete as a result of “strangulation” of coiled loops of vessels .
- The telangiectatic lesions are most commonly found on the face, lips, nares, tongue, nail beds, and hands. Some patients have only a few lesions necessitating a thorough search in anyone suspected of having HHT. Bleeding from these cutaneous telangiectasias is uncommon and rarely of clinical importance.
- Virtually all internal organs can be affected.
- Cerebral, pulmonary and neurological involvement is described more commonly with ENG mutations, whereas liver involvement has been associated with ALK1 mutations.

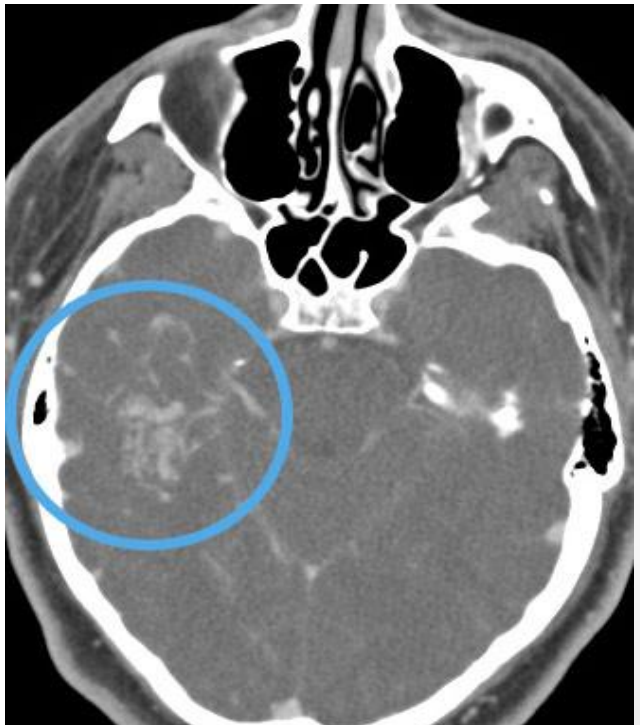
Pulmonary AVMs

- Majority of patients with pulmonary AVMs have no symptoms
  - 1/3 of patients show signs of right-to-left shunt (cyanosis, polycythemia)
- Pulmonary hemorrhage is rare, except in pregnancy (1.4%)



Cerebral AVMs

- Affect ~10% of HHT patients
- Majority are silent
- Hemorrhage is less likely with HHT than other causes of cerebral AVMs because of the lower association with aneurysms
- However, HHT patients are 23x more likely to have a hemorrhagic stroke than the general population



GIT Bleeding:

- Approximately 20% of patients with HHT develop significant upper and lower gastrointestinal (GI) tract hemorrhage. Bleeding is rare before the fifth decade of life. Approximately 40% of the bleeding episodes occur from upper GI tract lesions, whereas only 10% occur in the colon, and a full one half (50%) are indeterminate.
- Hepatic involvement may occur , but symptoms and complications are rare(cirrhosis, portal hypertension and high - output cardiac failure.)

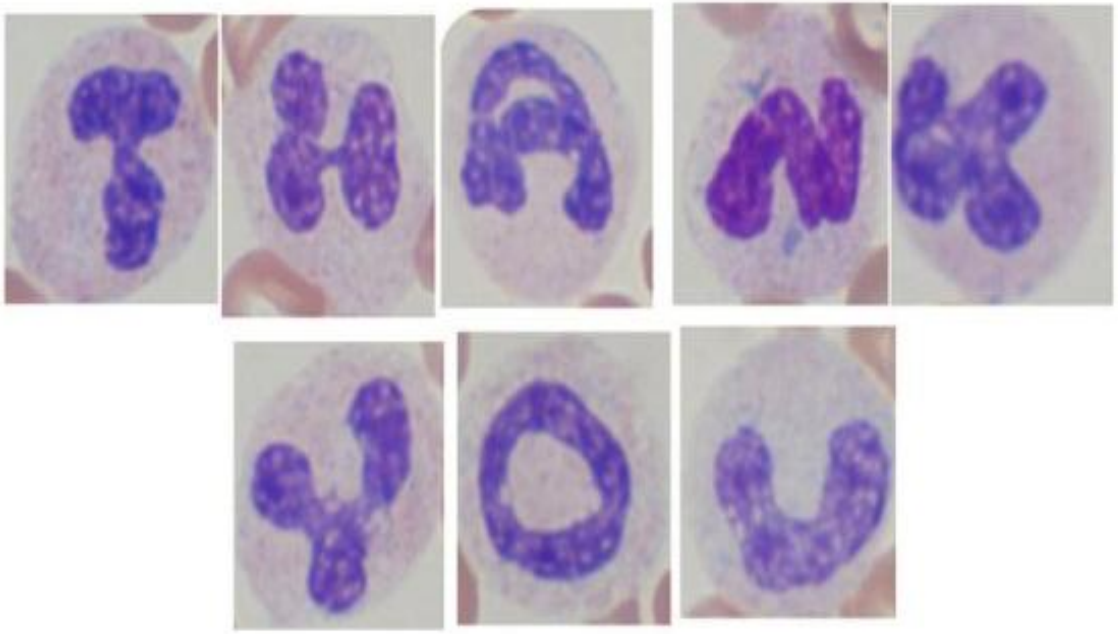
Curacao Criteria for the Diagnosis of Hereditary Hemorrhagic Telangiectasia

- **Epistaxis—spontaneous and recurrent**
- **Telangiectasias—multiple, at characteristic sites (lips, oral cavity, fingers, nose)**
- **Visceral lesions—with or without bleeding (gastrointestinal, pulmonary, cerebral, hepatic)**
- **Positive family history—a first-degree relative with HHT**

- The presence of three of the above criteria indicate definite HHT; the presence of two criteria are suspicious for HHT, if one criterion HHT is unlikely.

Management:

- Approximately **one third** of patients have mild symptoms requiring **no treatment**, and **another third** have moderate symptoms requiring only **outpatient treatment**. The **remaining third** have severe symptoms often requiring **inpatient treatment**, transfusions or chronic iron replacement therapy, and surgery.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