COMPILED BY :

DR ATHOTA KAVITHA

ACAD MODERATOR

JOINT SECRETARY,IADVL2022-23

PEDIATRIC DERMATOLOGY

SIG PEDIATRIC DERMATOLOGY (IADVL ACADEMY)

(ACAD DISCUSSION 2022)

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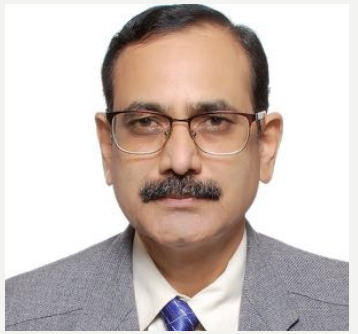

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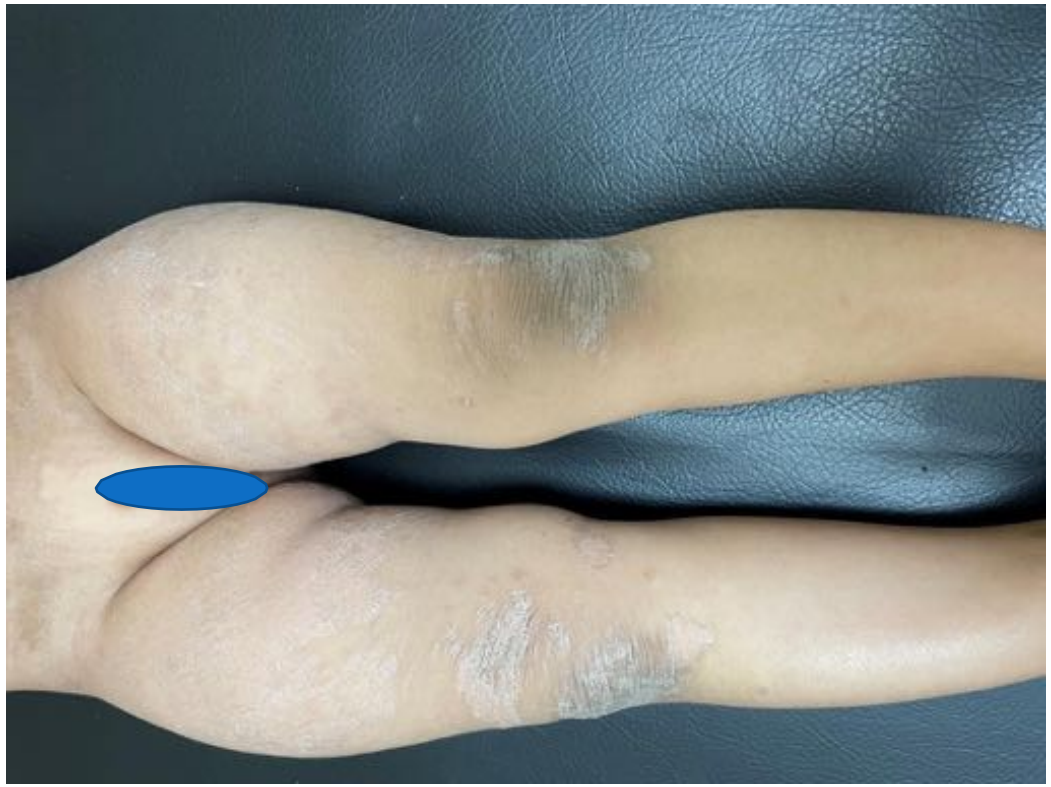
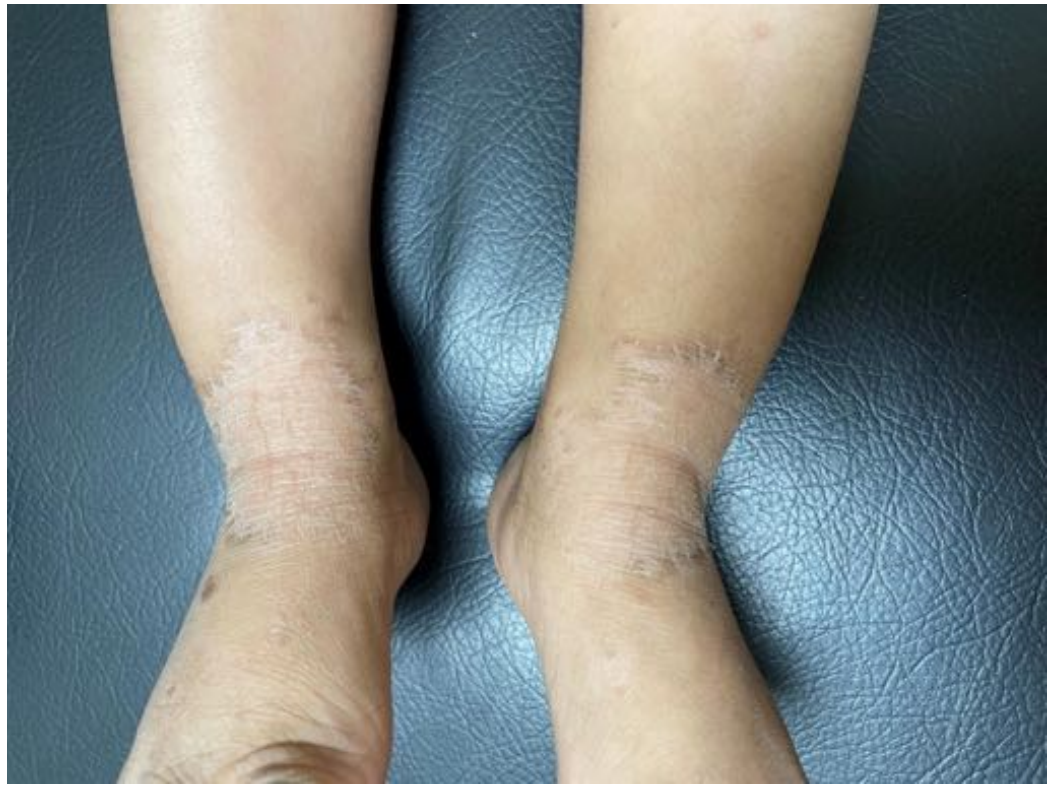
CASE 1

DR JIGNA PADHIYAR

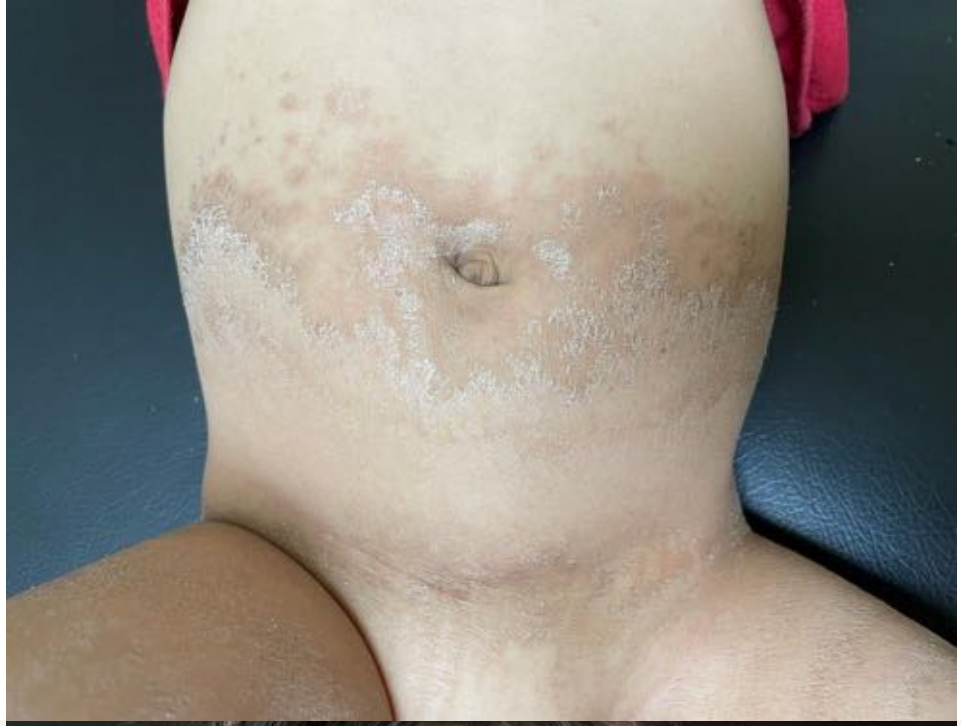
A child with symmetric papulo-squamous lesions

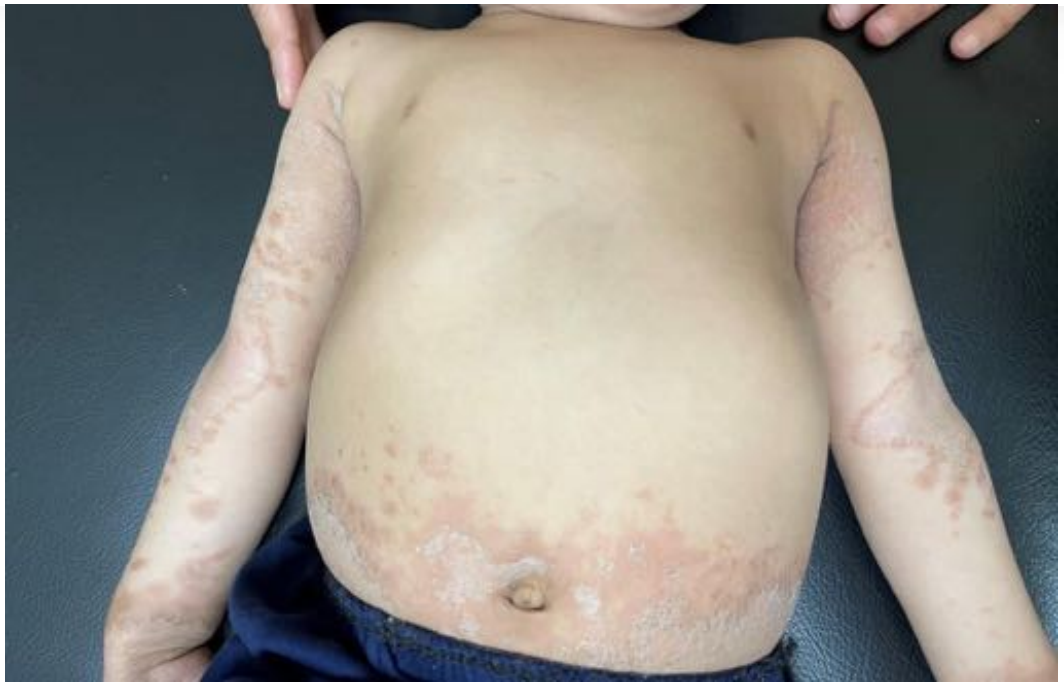
A 2-year-old female child born out of non- consanguineous marriage

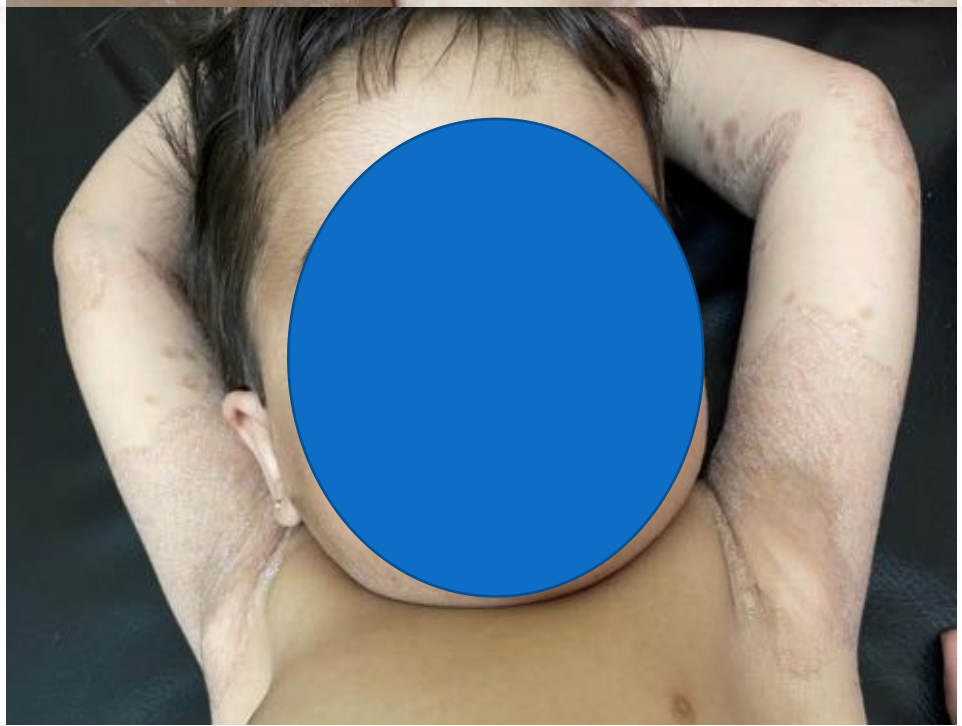


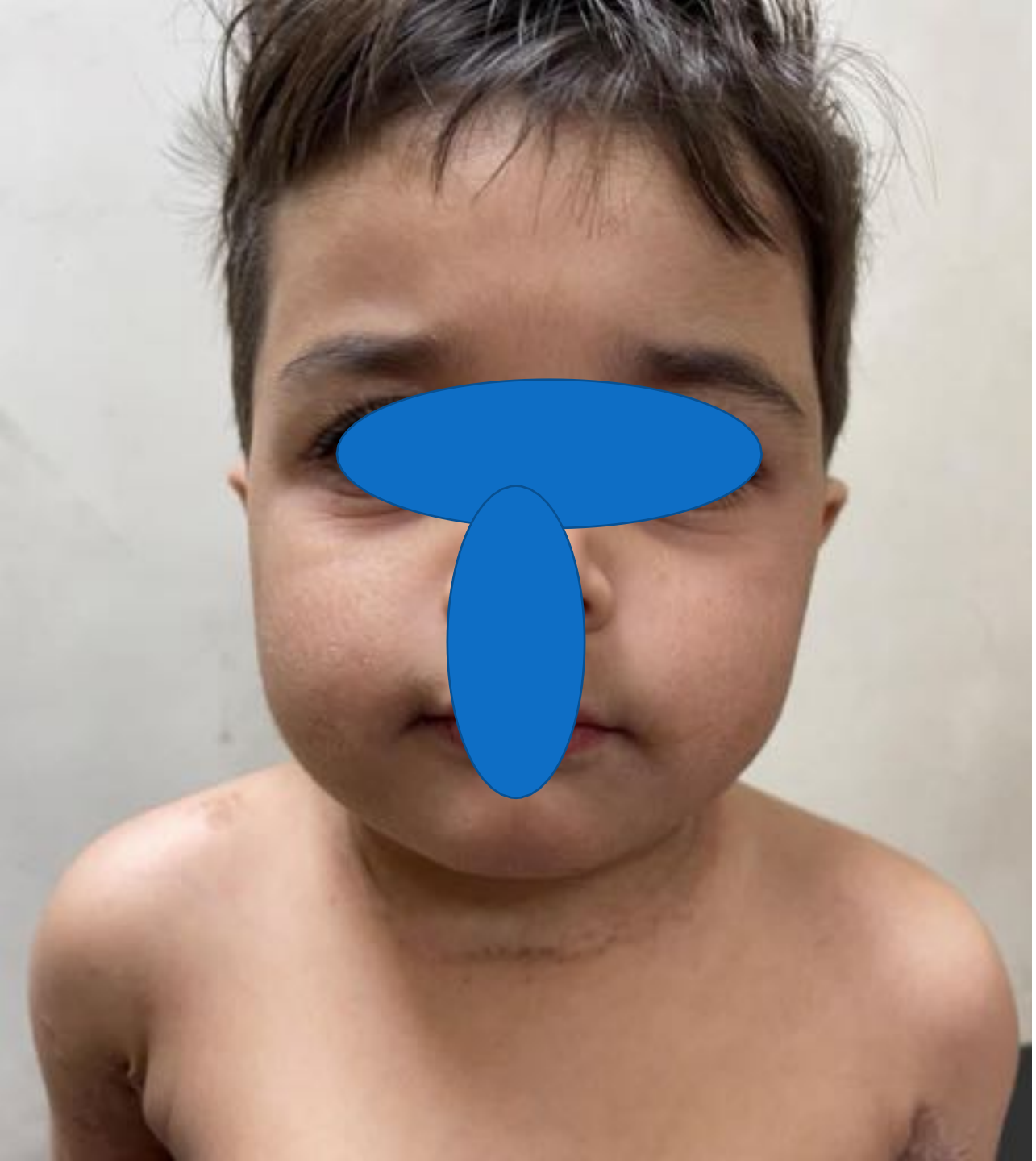


Bilateral symmetrical scaly plaques since the age of 2 months









Patient had xerosis over face and scaly plaque over neck

Other relevant history/examination points

- Patient was put on top-feed since the age of two months
- Lesions were asymptomatic
- No changes in oral mucosa and nails
- No hair abnormality found
- No family history of similar findings
- No history of atopy

Significant Basic Investigation

Haemoglobin: ranged from 6.8-8.6 on various occasions

SGPT: 175 (0-32 IU/L)

SGOT: 127.30 (<31 IU/L)

ALP: 473.00 (54-369 U/L)

Serum Zinc: 0.54 (0.60-1.20 microgram/ml)

USG: Mild Hepatomegaly (on multiple occasions)

Rest reports – Serum protein, TSH, HIV, HBsAg, HCV, RFT, Serum bilirubin were normal.

Differentials

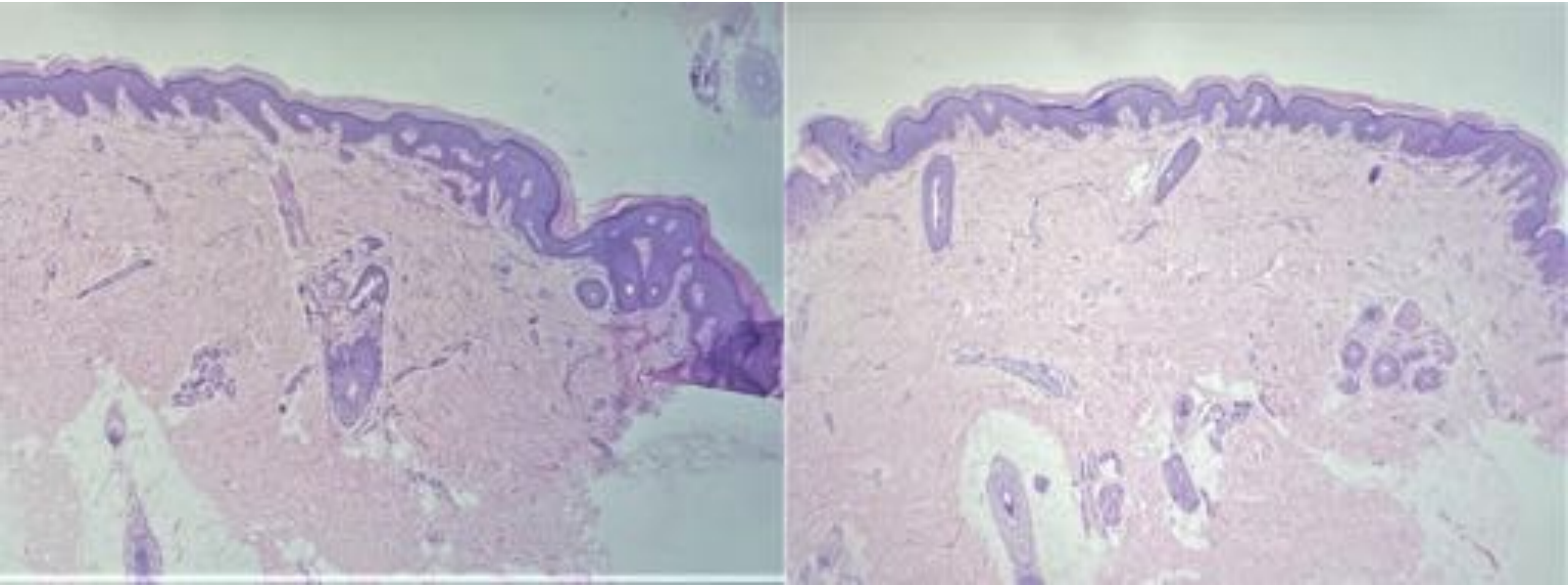
? Pityriasis rubra pilaris

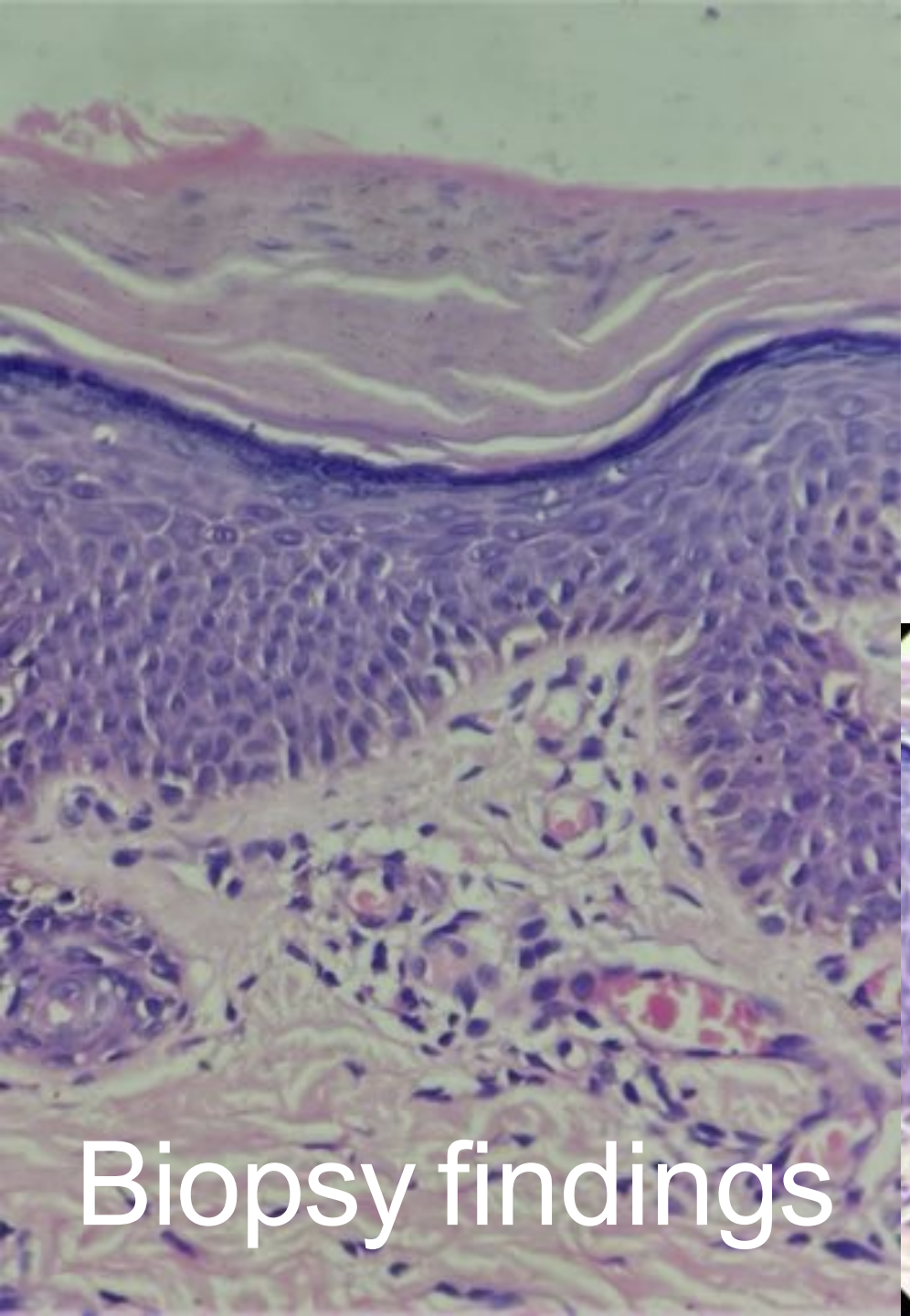
? Psoriasis

? Acrodermatitis enteropathica

? Netherton syndrome

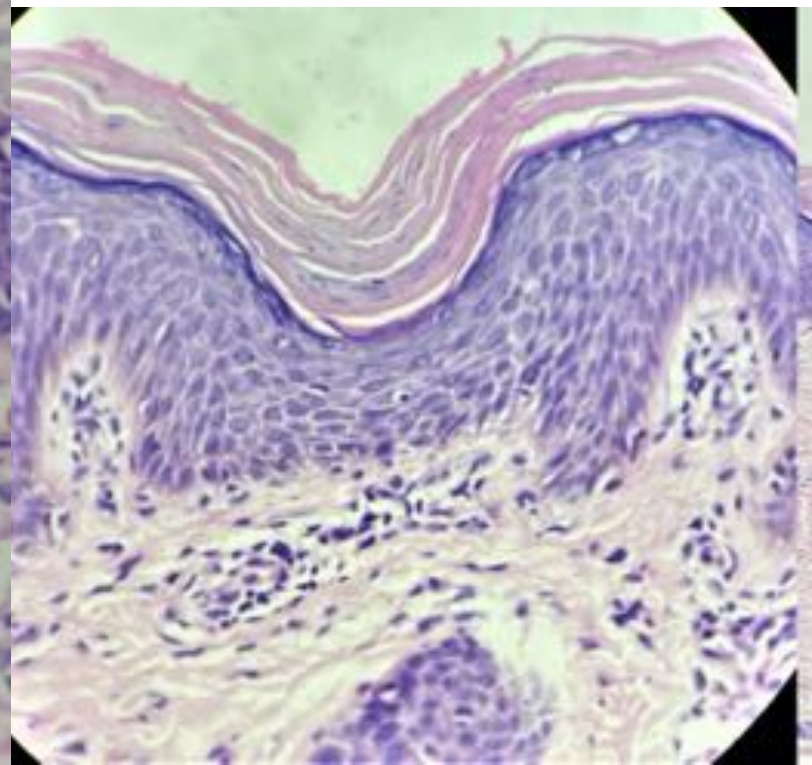
Biopsy findings: hyper keratosis, parakeratosis, mild hyper granulosis, subtle papillomatosis

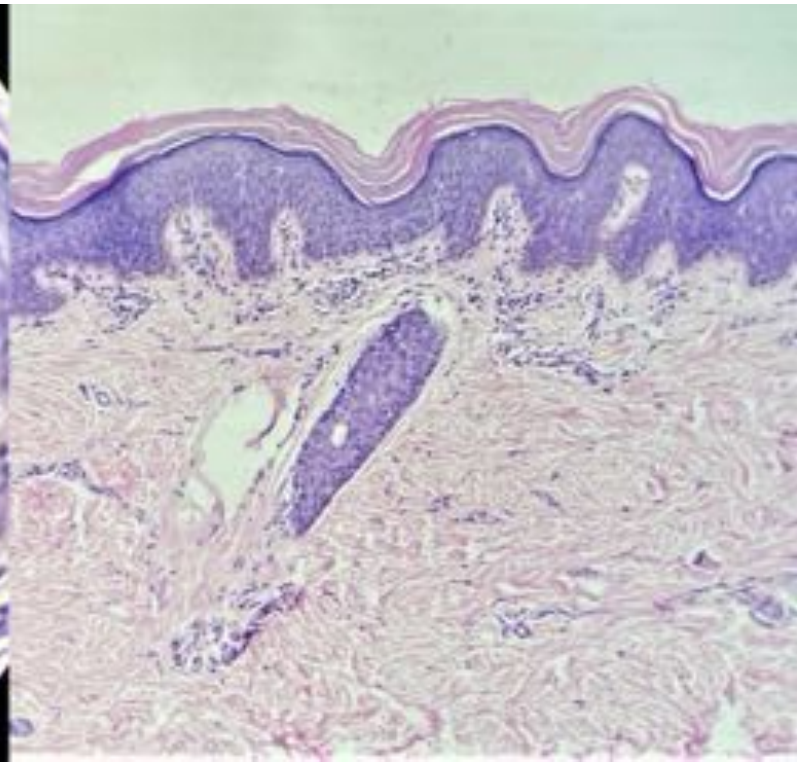




Biopsy findings

Hyper keratosis, parakeratosis, mild hyper granulosis, subtle papillomatosis





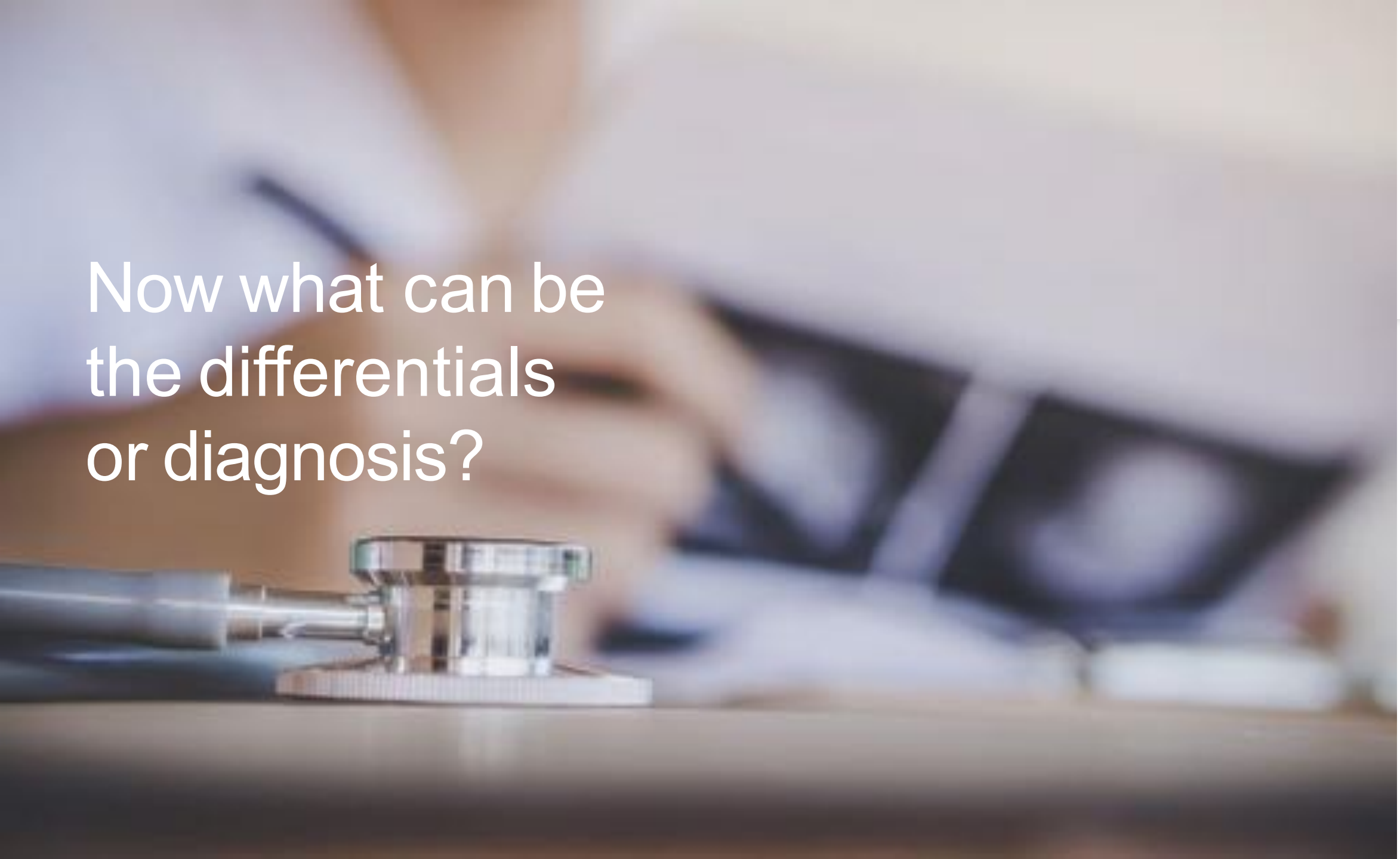
Based on these findings final diagnosis of erythrokeratoderma variabilis progressive (EKVP)/ Progressive symmetric erythrokeratoderma (PSEK) was made

EKVP/PSEK

Genetic analysis report

VARIANT OF UNCERTAIN SIGNIFICANCE RELATED TO THE GIVEN PHENOTYPE WAS DETECTED

Gene* (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
***ABHD5* (+)** (ENST00000644371.2)	Exon 6	c.893C>T **(p.Ser298Phe)**	Homozygous	Chanarin-Dorfman syndrome	Autosomal recessive	Uncertain Significance


Now what can be
the differentials
or diagnosis?

Ichthyosis and Hepatic disorders

- Omen Syndrome
- Neonatal ichthyosis-sclerosing cholangitis (NISCH syndrome)
- Mitochondrial disorder mutation in TRMU gene
- Chanarin- Dorfman syndrome
- Disorders of metabolism

- As we had genetic analysis with us, we did not advise peripheral smear to be done again. But his previous reports did not mention of Jordan anomaly.
- Staining for lipids were not available at our centre.

Chanarin Dorfman syndrome

- Multisystem, very rare, autosomal recessive lipid storage disorder
- Lipid is stored in the granulocytes at various sites in the human body, such as the muscle, liver, eye, ear, central nervous system, and bone marrow.
- Clinically, the disease is presented with ichthyosis, hearing loss, hepatomegaly, splenomegaly, cirrhosis, cataract, keratopathy, myopathy, and mental retardation.
- Lipid accumulation in the neutrophils (Jordan's anomaly)

Few of the reference which have reported EKV like lesions in Chanarin Dorfman syndrome

- Pujol RM, Gilaberte M, Toll A, Florensa L, Lloreta J, González-Enseñat MA, Fischer J, Azon A. Erythrokeratoderma variabilis-like ichthyosis in Chanarin-Dorfman syndrome. Br J Dermatol. 2005 Oct;153(4):838-41.
- Job, Anupa Mary; Aithal, Vijay; Lobo, Carol; Shanthala Devi, AM1. Dorfman-Chanarin Syndrome: An Unusual Presentation. Indian Journal of Paediatric Dermatology: Apr–Jun 2019 - Volume 20 - Issue 2 - p 163-165
- Chander, R., Varghese, B., Garg, T., Mittal, S., & Singh, S. (2011). Dorfman-Chanarin syndrome in two female siblings: Acase report and discussion on approach & management. Dermatology Online Journal, 17(4).

Final diagnosis of EKVP like lesions in case of Chanarin Dorfman syndrome was made

Treatment Chanarin Dorfman syndrome

- A multidisciplinary approach of various specialties, including internal medicine, dermatology, ophthalmology, nutrition, and gastroenterology.
- Diet is especially one of the most important components of therapy.
- Diet that lacks long-chain fatty acids and rich in medium-chain fatty acids.
- Ursodeoxycholic acid 250 mg 3×1/day per
- Treatment according to other systems involved

Treatment with retinoids

As this is a lipid storage disorder, we started patient on just topical therapy from our side.

What is opinion of experts on using retinoids in such patient?

Role of vitamin E, statins and ursodeoxycholic acid

- Will it improve skin lesions?
- Few studies have demonstrated that severity of ichthyosis corelates with triglyceride levels.

DISCUSSION THREAD….

- I think the real challenge starts after the diagnosis is made on how to treat such rare diseases.
- In my opinion, depending on functional disability due to disease, one should start with topical therapies like calcipotriol or tazatotene along with oral vitamin D 2000 i.u. per day.
- In case of inadequate response, oral acitretin can be added 0.5 mg per kg per day.

- An important issue will be monitoring for hepatotoxicity due to acitretin as these patients already have raised liver enzymes. Personally I have found acitretin to be safe in couple of my patients of chanarin Dorfman.

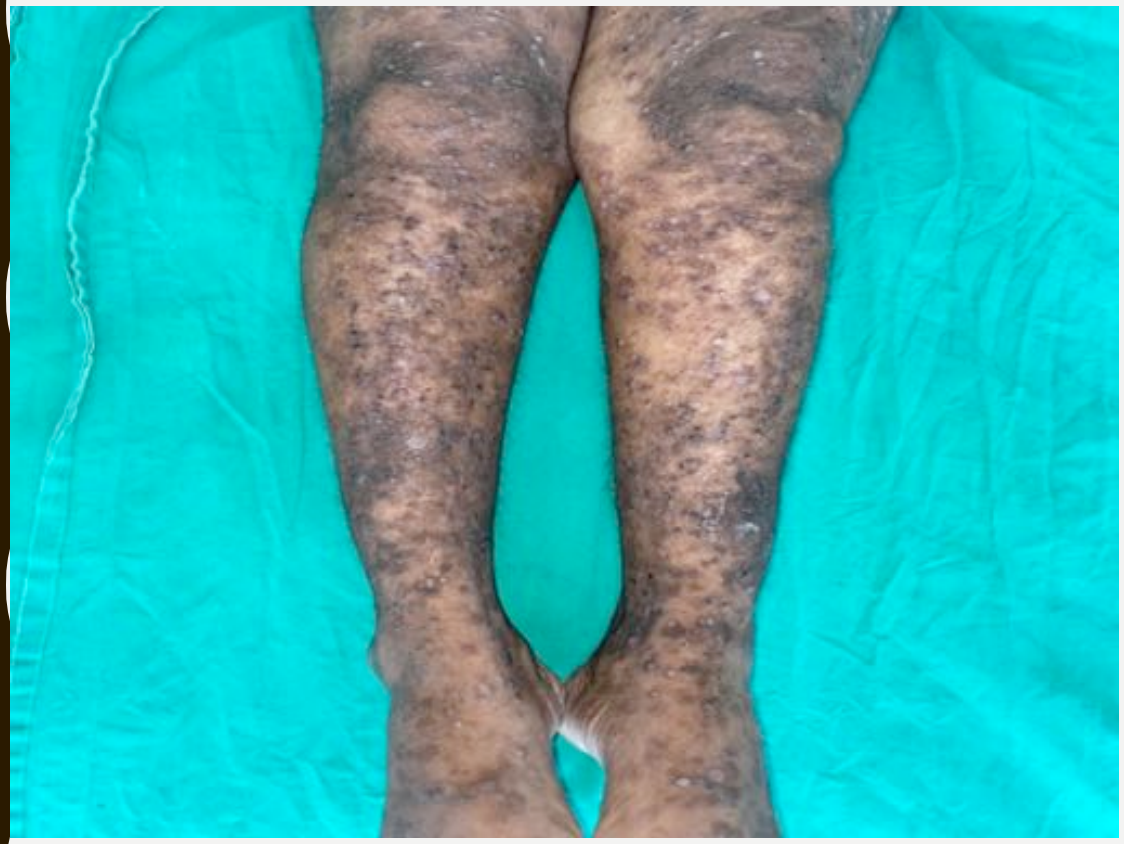
- Additionally, dietary modifications - low fat diet with little to no long-chain fatty acid/saturated fat.
- For hepatic issues, long term vitamin E and udiliv should be added. For hypertriglyceridemia, consider adding fibrates (only after consulting pediatric gastroenterologist)

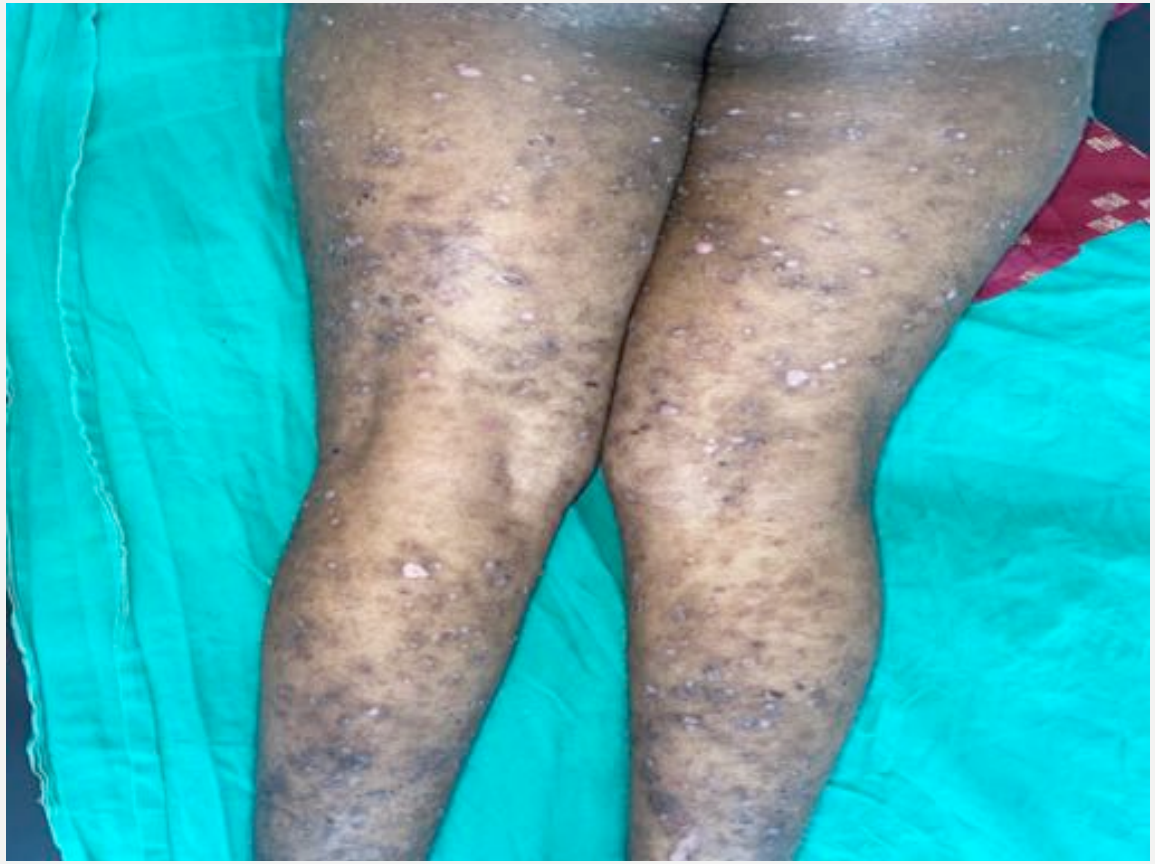
CASE 2

DR LIZA MOHAPATRA

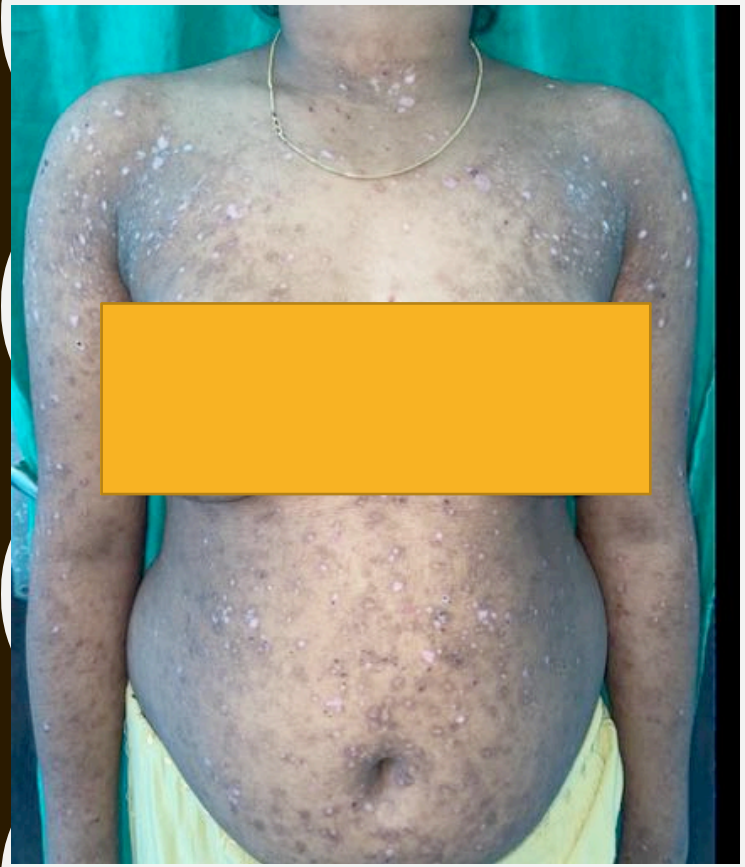
CASE 2

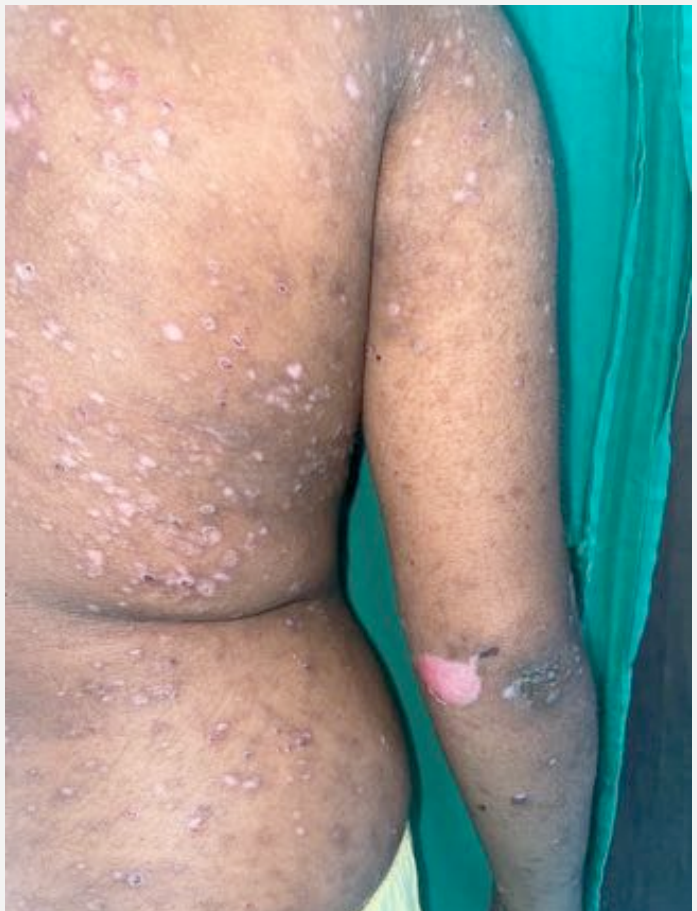
- 14-year-old female presented with multiple tense pruritic blisters all over her body since childhood.



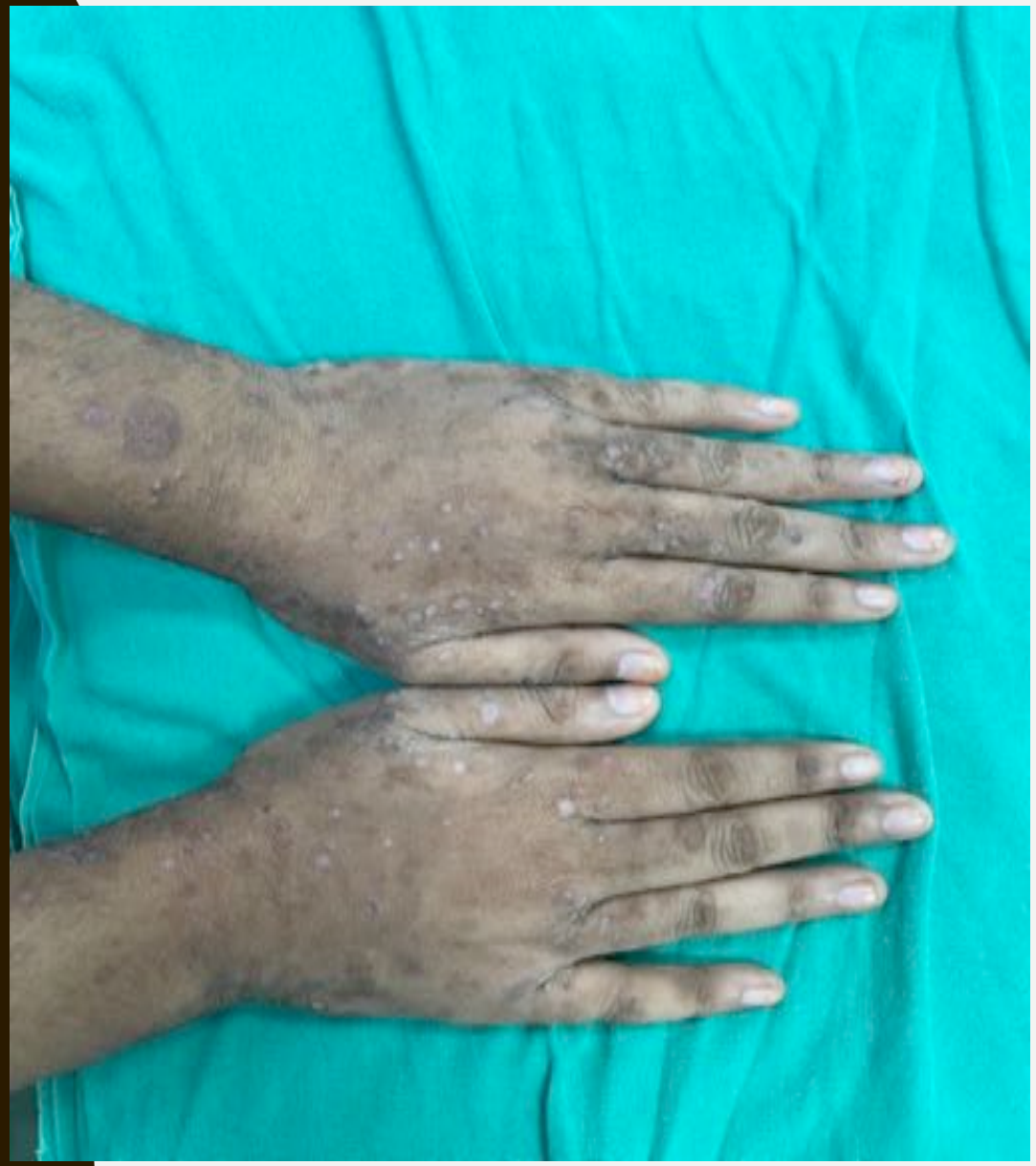


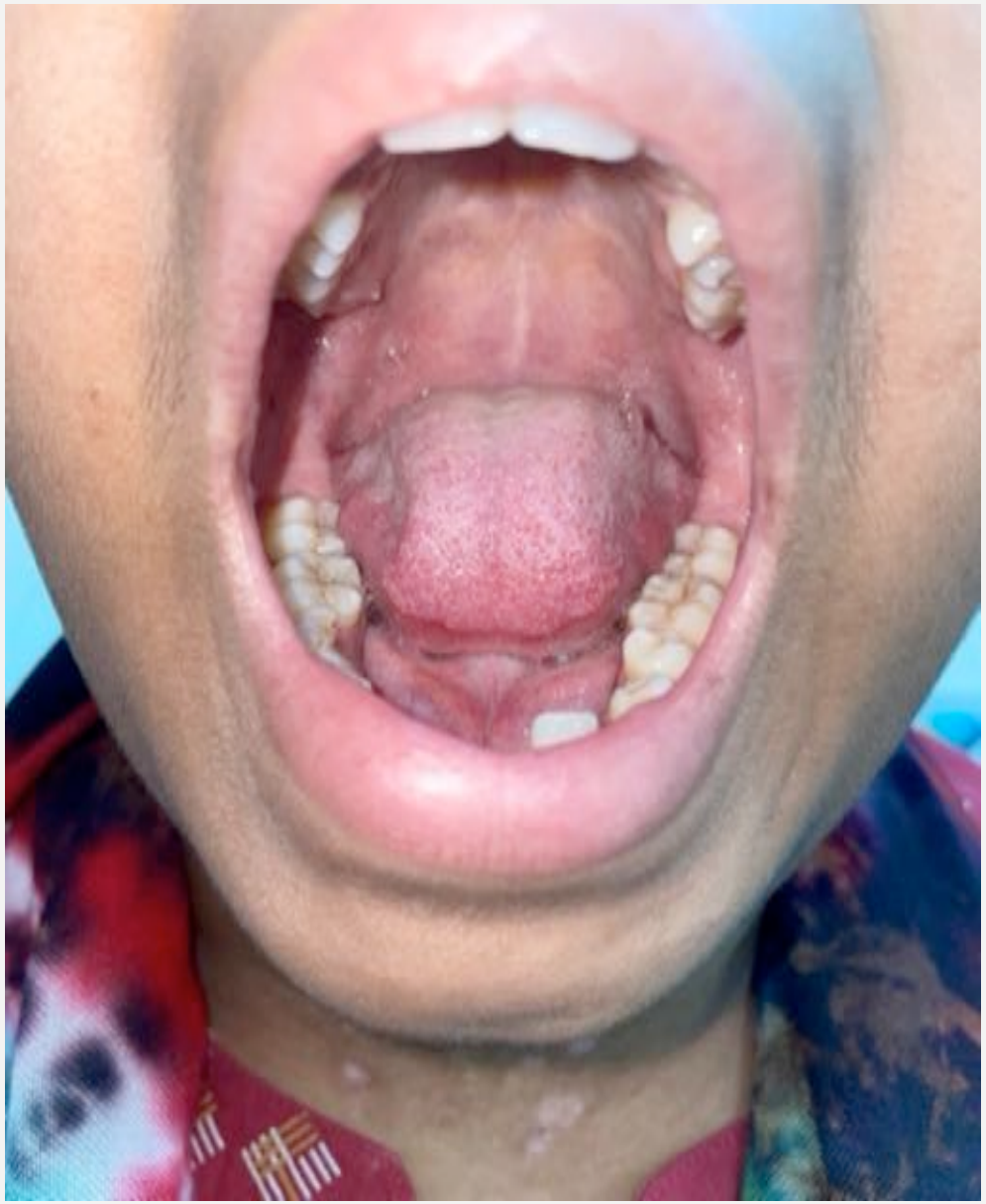
- Started since 2 years of age with blistering limited to face and trauma prone areas.Generalized distribution in last 2 years





- Similar presentation in elder brother
- Born out of consanguineous marriage.
- Hair, Nails, Teeth –normal
- No mucosal lesions.
- No H/O wheals before appearance of lesions.
- Scarring minimal , No Milia
- Palm sole involvement-NIL

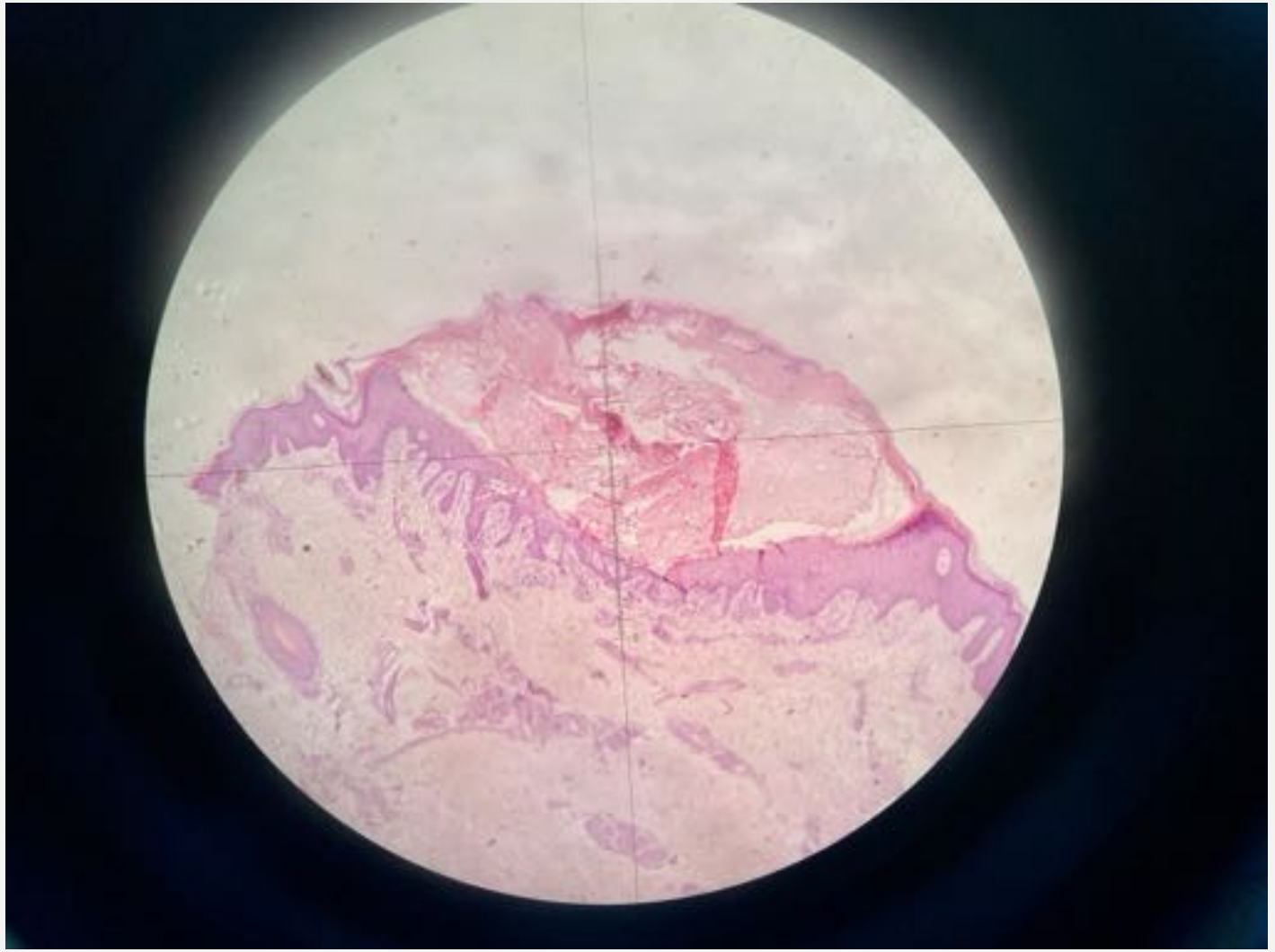




- Investigations:
- CBC-Hb-9.2gm%, TLC-12.03, TPC-2.2 lakhs
- Other blood investigations within normal limits
- Differentials?
- CBDC
- Bullous pemphigoid
- EBS

HISTOPATHOLOGY

- 2021,December – biopsy showed subepidermal blistering
- 2022 June- Intraepidermal split



- IFM report shows-

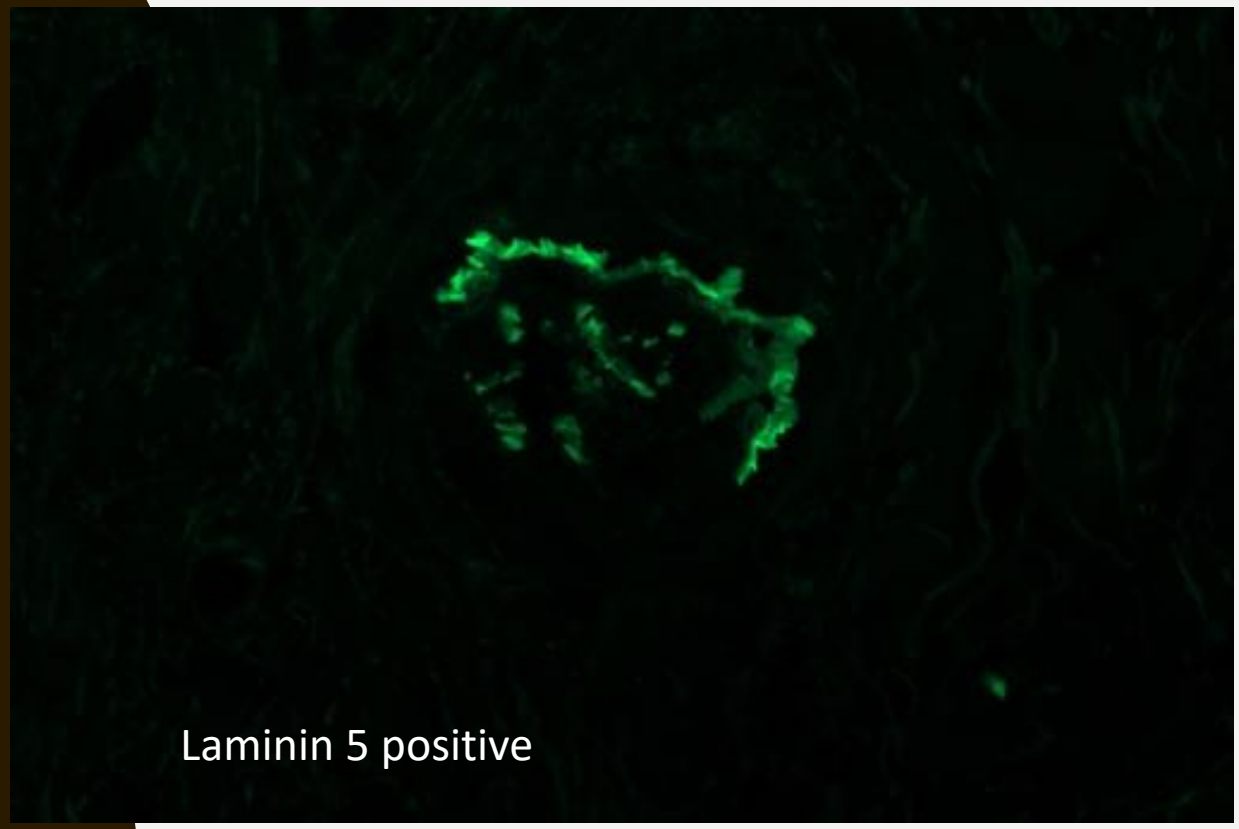
  Collagen 7 Positive rules out Dystrophic EB

  Laminin 5 Positive rules out dominant type of Herlitz EB

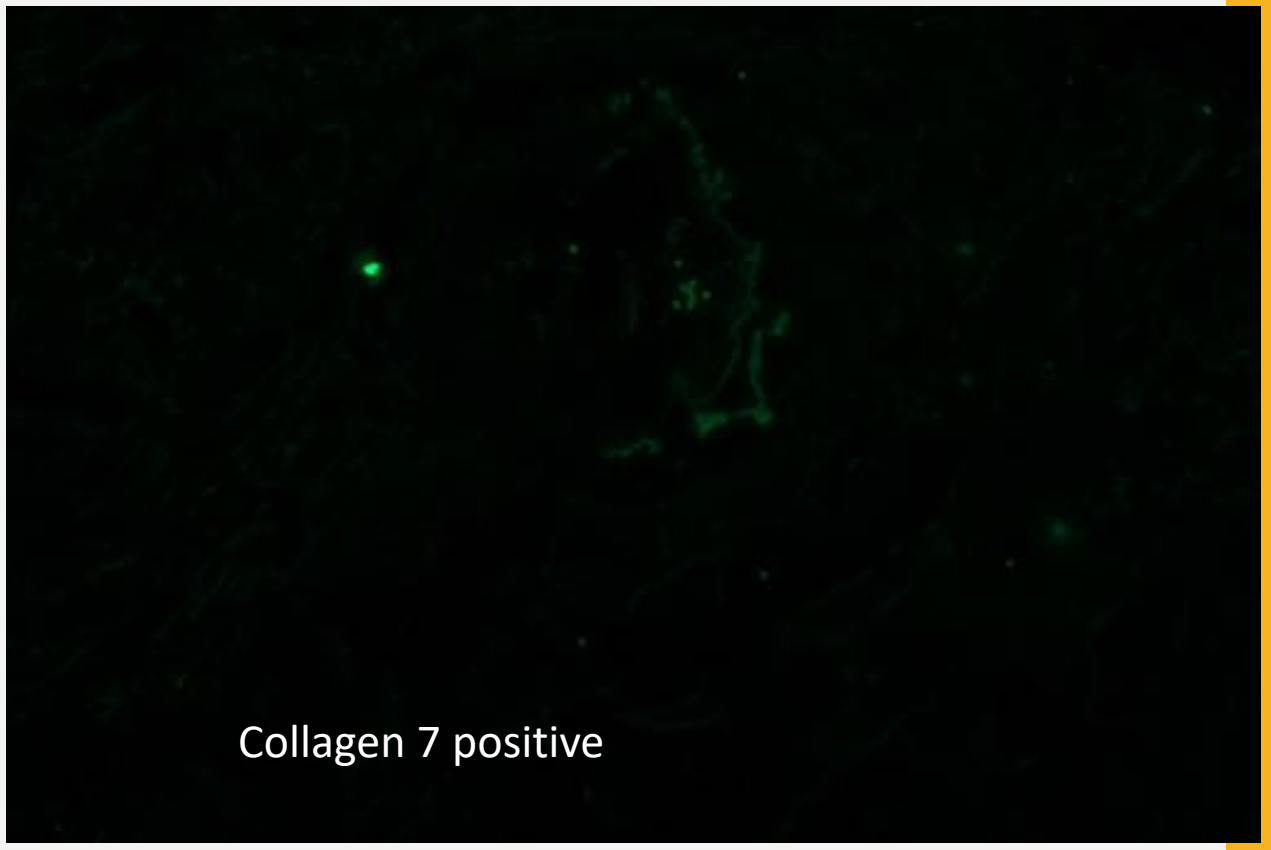
  Collagen 17 Negative – suggests junctional EB subtype

  Keratin 5 and 14 – not interpretable due to damaged epidermis

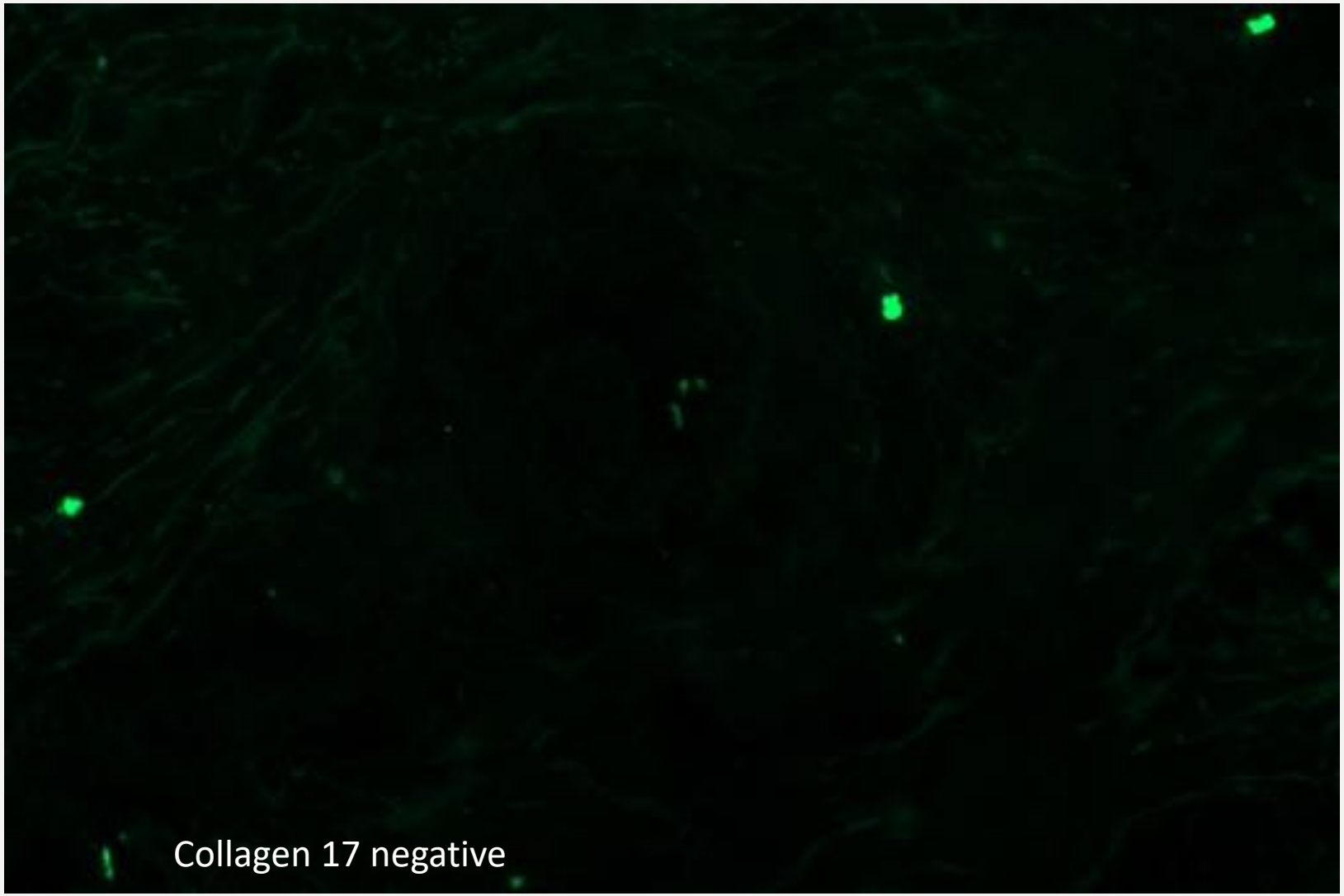
Since biopsy tissue had little epidermal tissue, staining is shown only for dermal tissue around hair follicles



Laminin 5 positive



Collagen 7 positive


Collagen 17 negative

- Diagnosis ??- Collagen 17 JEB / EBS
- Final diagnosis – Mutation analysis (sample sent for the same)
- Management ?

- The patient responded to prednisolone at a dose of 1mg/kg body weight tapered slowly .
- With few episodic blistering.
- She was also prescribed dapsone – but was discontinued on the basis of falling Hb and raised Reticulocyte count.
- The disease activity in her brother in contrary was quiescent.
- Along with prednisolone she was also given methotrexate 10mg/week

Take home message from the above case

- Interpretation of the immuno fluorescent antigen mapping shows that laminin 5 positivity around the hair follicle area and collagen 7 positivity. This means laminin 5 and collagen 7 structure are intact or normal.Collagen 17 negative in IMF suggests either absence of the structure or mutated collagen 17. Keratin 5 and 14 could not be interpreted due to damaged epidermis.
- There is a subgroup of hemidesmosomal variants of EB that includes Plectin mutation leading to EB with MD, BPAG2/ Collagen 17 mutation leading to junctional EB, and alpha 6 beta4 integrin mutation resulting EB with PA
    - GABEB(Generalized atrophic benign EB) is a variant of junctional EB where collagen 17 expression is absent. It usually presents with generalized blistering with atrophy, pigmentary changes and tooth and nail abnormalities.
    - Our case had such a similar generalised presentation but without atrophy and nail defects.

The learning point here is always to look for **DIF positivity** in childhood blistering cases if we suspect CBDC.

- Some response to steroids and dapsone again can be explained by the fact that some inflammatory variants of EBS respond to steroids

CASE 3

DR RAHUL MAHAJAN

- 15- years- old Male Student ,R/O Bijnour, UP
- Chief complaints
- Painful oral erosions for 3 months
- Loss of weight in last 3 months
- Weakness and malaise for 3 months
- Pain, bleeding, erosions on fingers for 1.5 months

History of present illness:

- Painful oral erosions started from left buccal
- mucosa- involved all oral mucosal surfaces in next 3 weeks
- Associated with gradually progressive difficulty in eating due to pain and bleeding from erosions significantly
- decreased oral intake

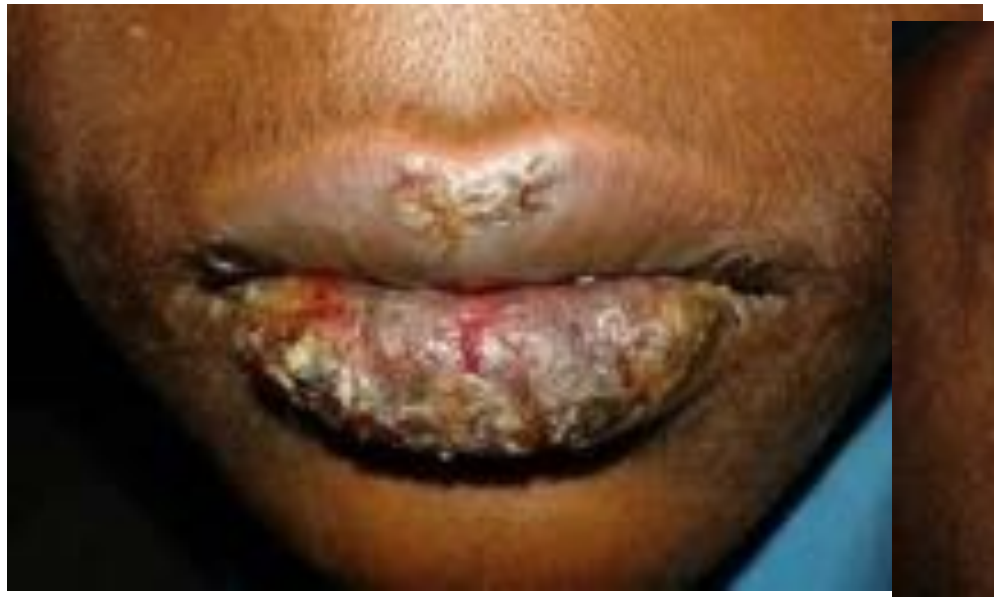
- Though there was no decrease in appetite, there was significant weight loss of 10 Kgs in 3 months
- Progressive erosions on periungual areas with associated pain
- Generalized weakness and malaise
- No history of prior drug intake
- No history of fever, night sweats
- No history of recurrent infections or abnormal bleeding
- No h/o cough, chest pain, breathlessness
- No altered bowel or urinary habits, pain abdomen or jaundice

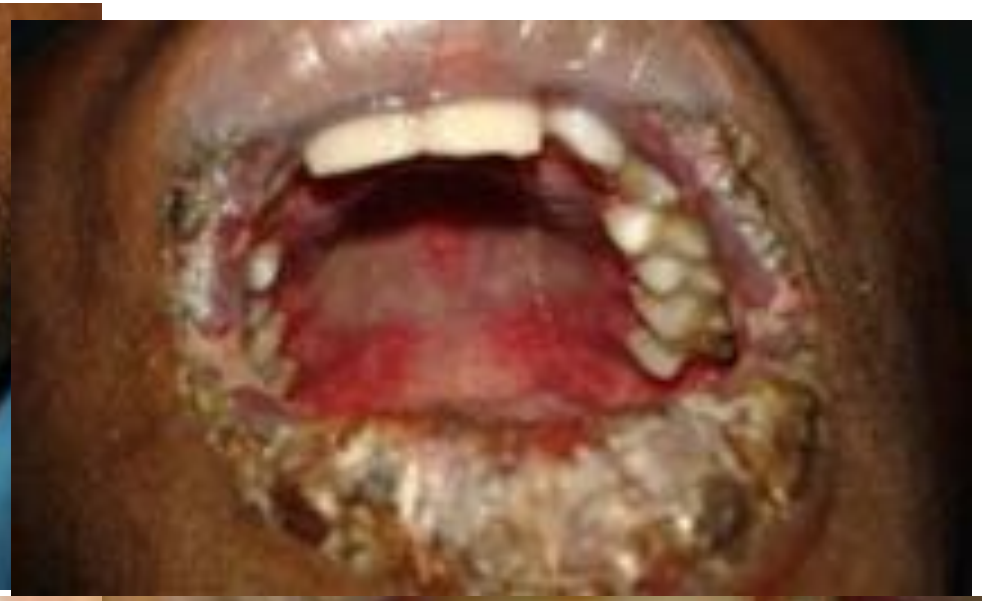
- Past history: non contributory
- Treatment history: nothing significant
- Family history:
- Parents and two brothers did not have malignancy or significant oral erosions
- Personal history:
- Non contributory

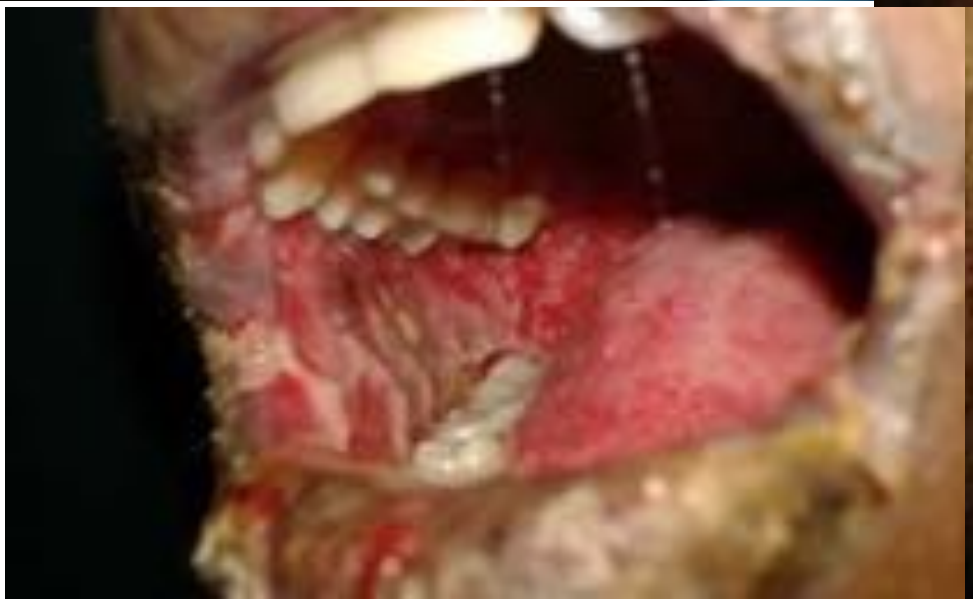
General physical examination:

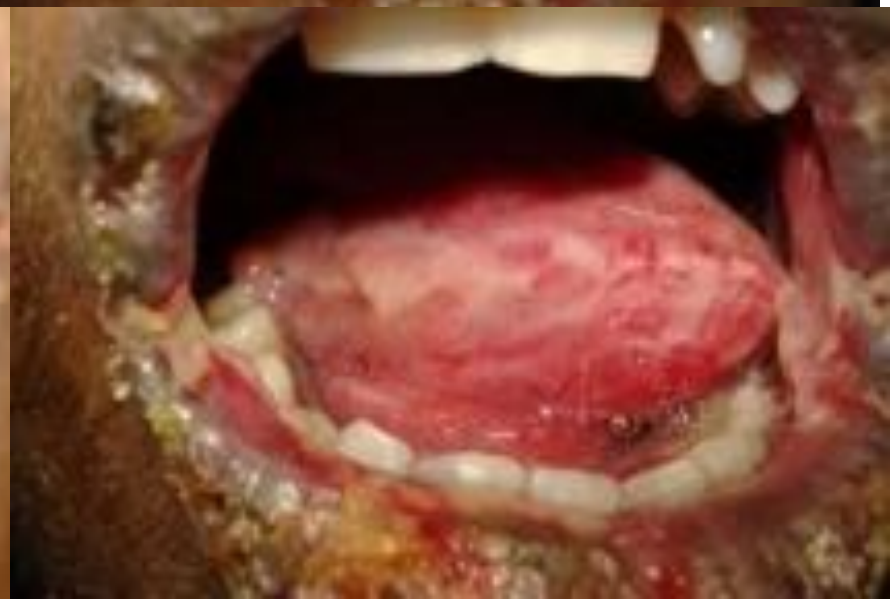
- Thin built, malnourished
- Wt: 30 Kgs
- Pulse 90/ min, regular
- BP 114/74 mm hg
- Temperature 98.60 F
- A+ CY0J0CI0 Oe0LNE (axillary, inguinal, lower
- cervical, firm, discrete, mobile, 0.5-1.5 cm in size)

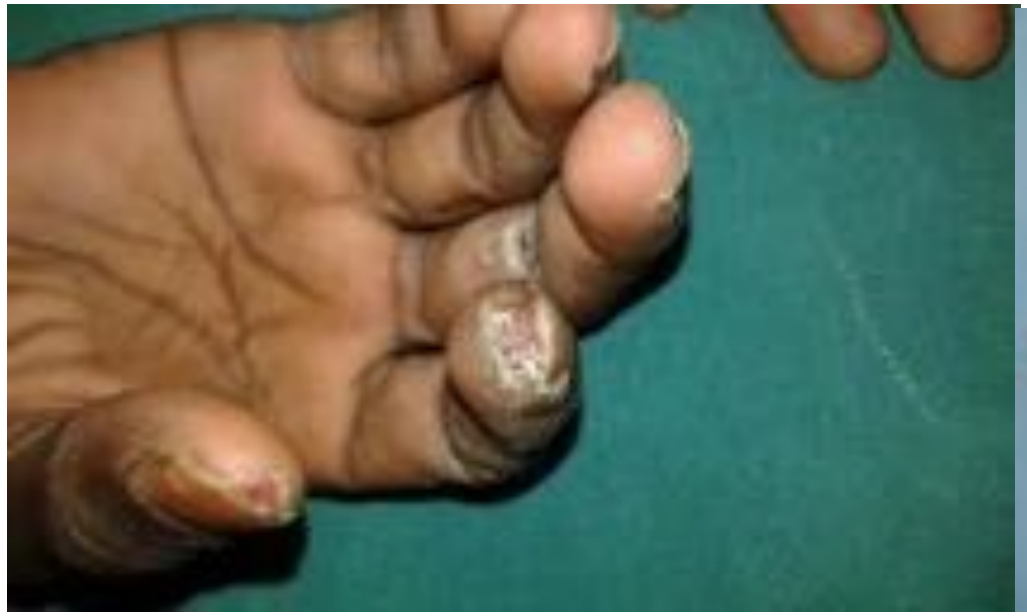
- Systemic examination
- CNS-WNL
- CVS-WNL
- Respiratory system-WNL
- Per abdomen- soft tender, no abdominal mass

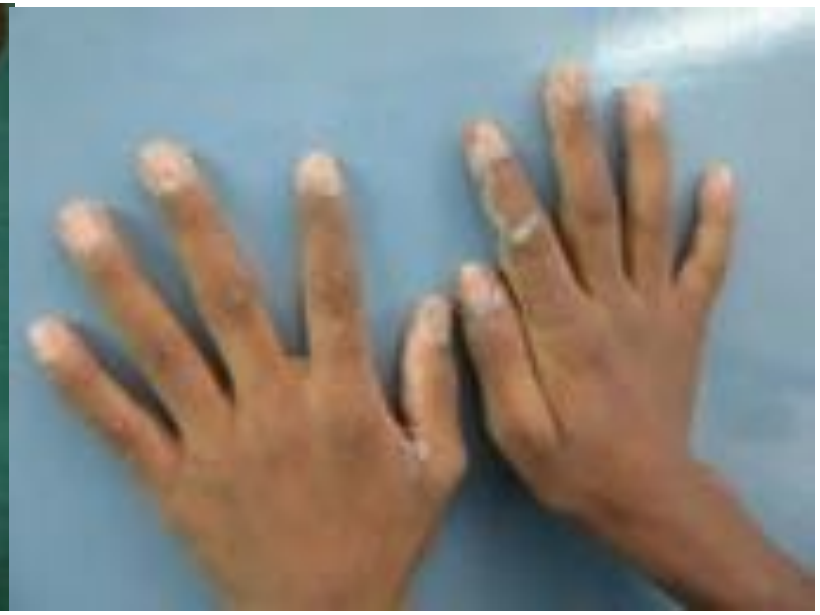


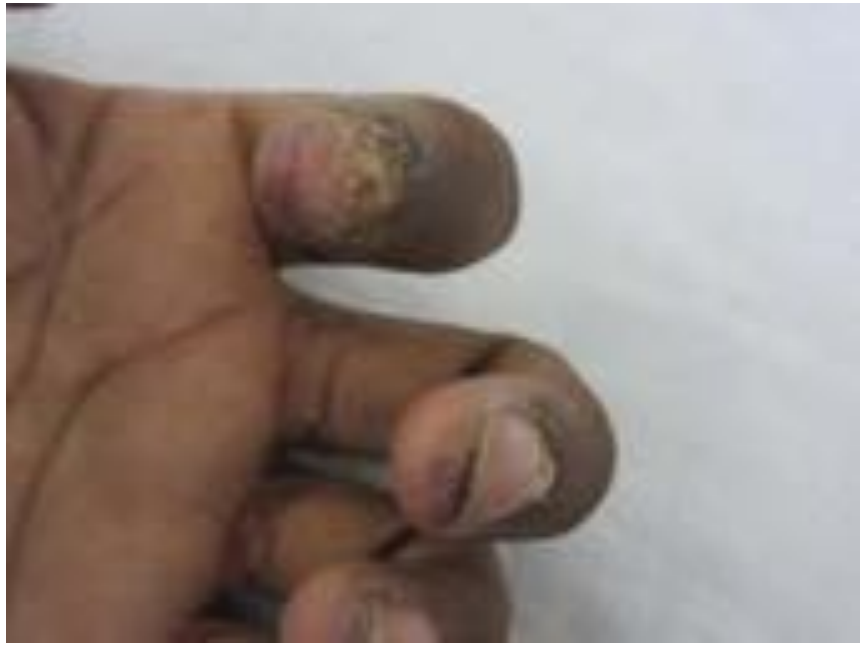








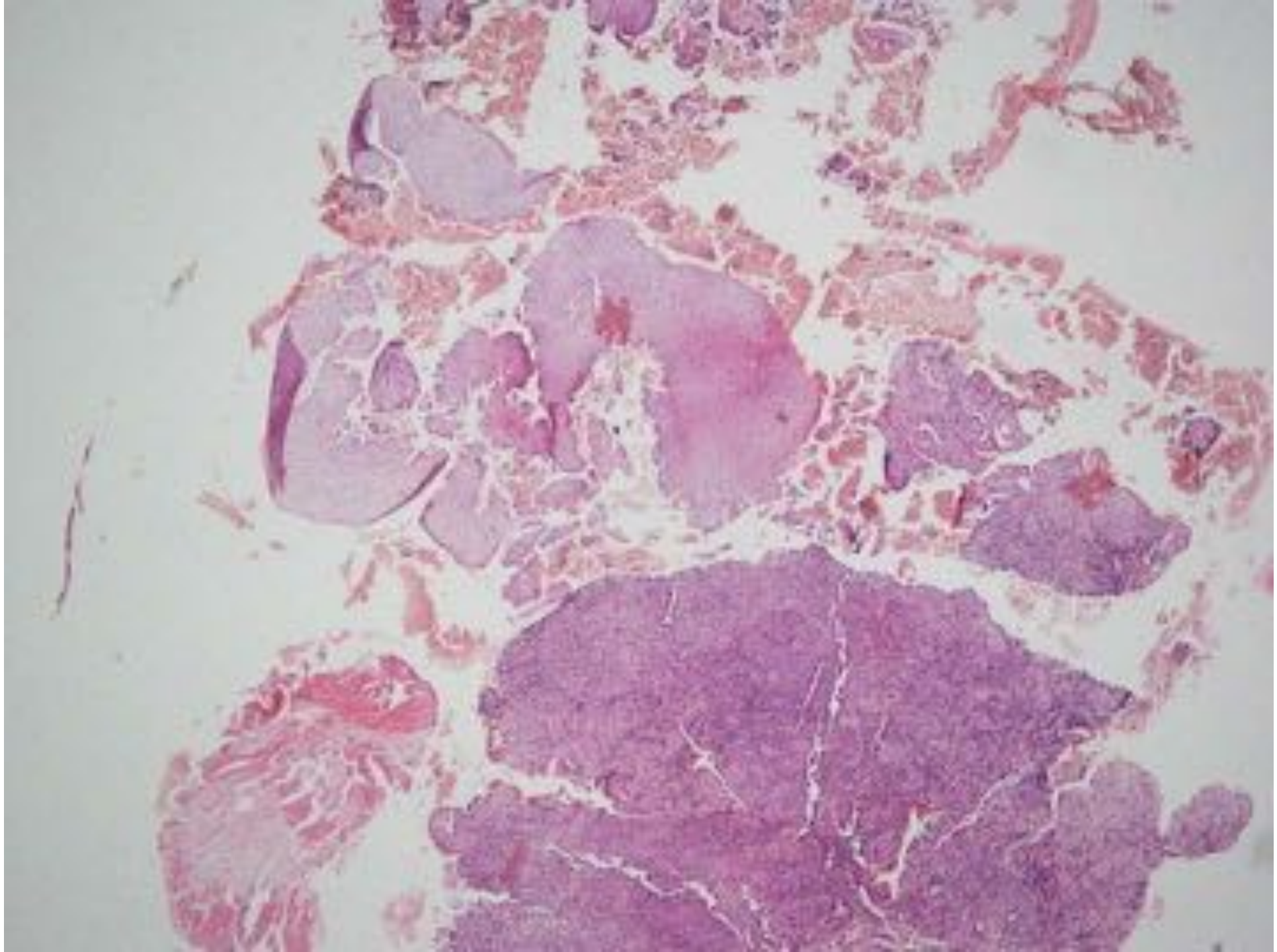


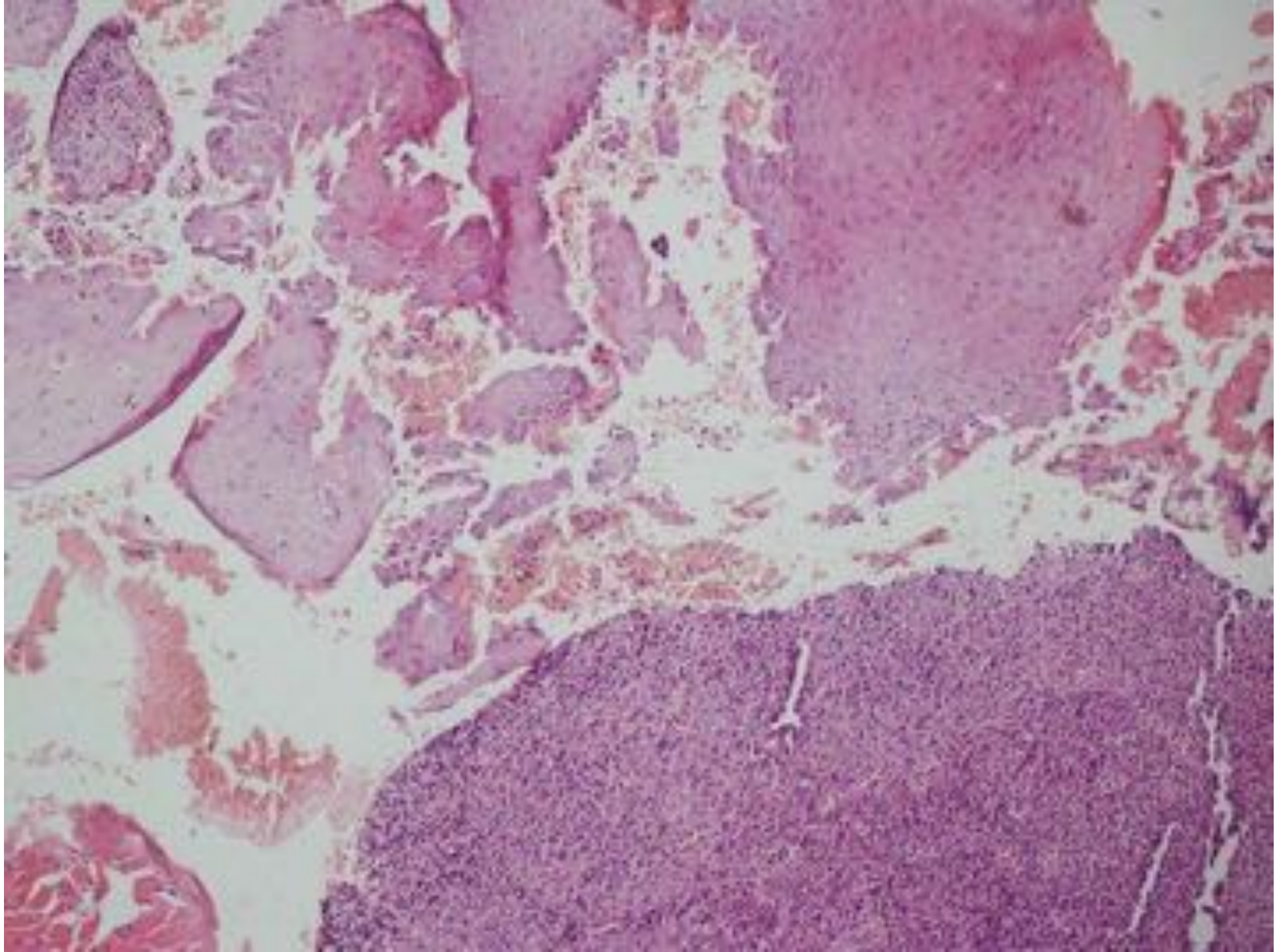


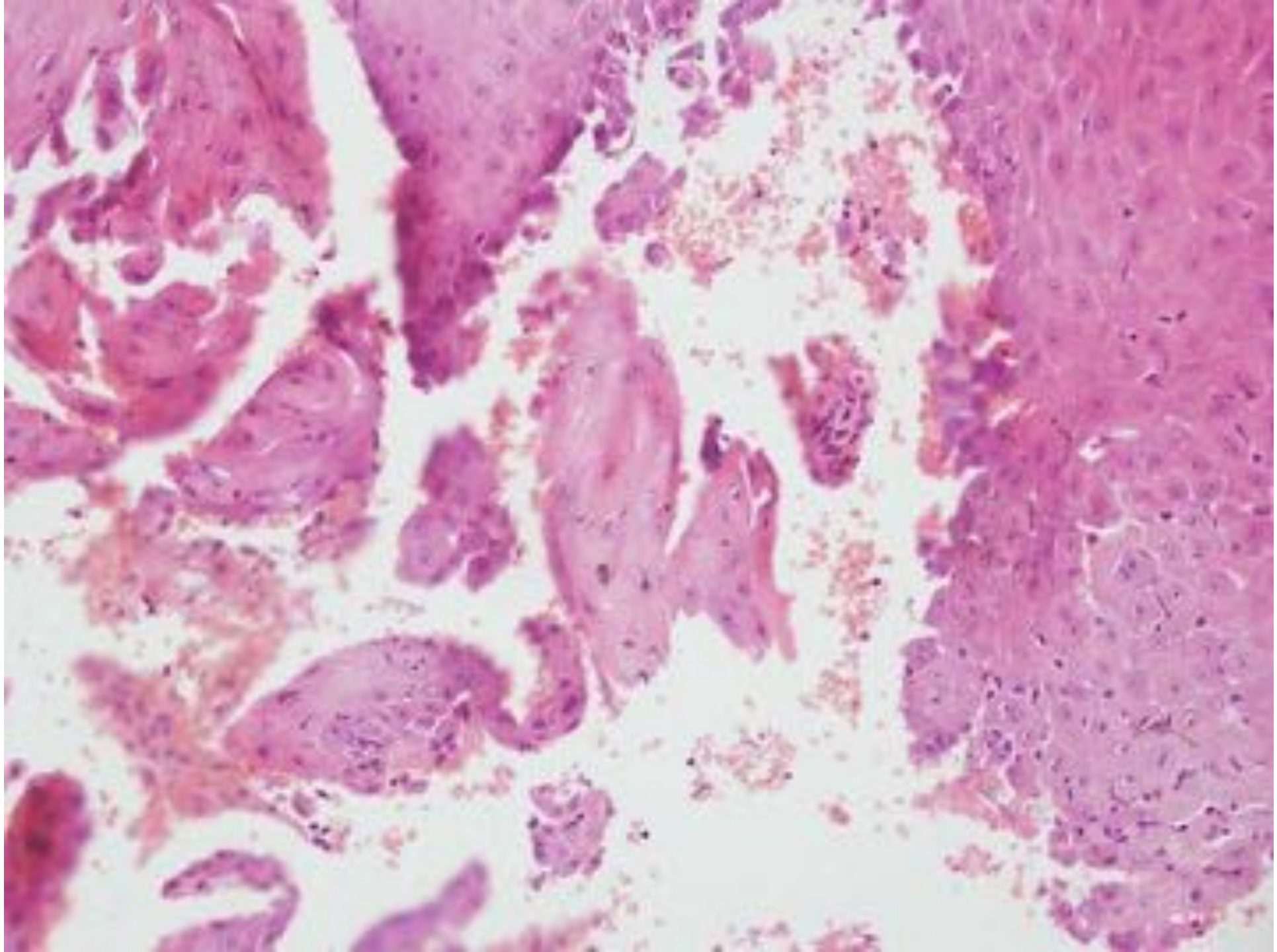
- Investigations:
- ► Hb- 13.4 gm%
- ► TLC- 8300/mm³
- ► DLC- N60L30M8E2
- ► Na/K- 140/4.24
- ► Ur/ Cr- 33/0.76
- ► TSB- 0.54
- ► TSP/Alb- 6.7/3.54
- ► OT/PT/AlkP-34/18/167
- ► Uric Acid- 5
- ► LDH-781
- ► ECG- WNL
- ► CXR- WNL
- ► ESR- 30
- ► CRP- 4.95
- ► Serum electrophoresis- mild Hypergammaglobulinemia along with mild alpha-2 prominence
- ► HIV- NR
- ► HBsAg/ Anti-HCV- negative
- ► HSV1/2 IgG and IgMnegative
- ► FNAC from enlarged lymph nodes- Reactive lymphoid hyperplasia

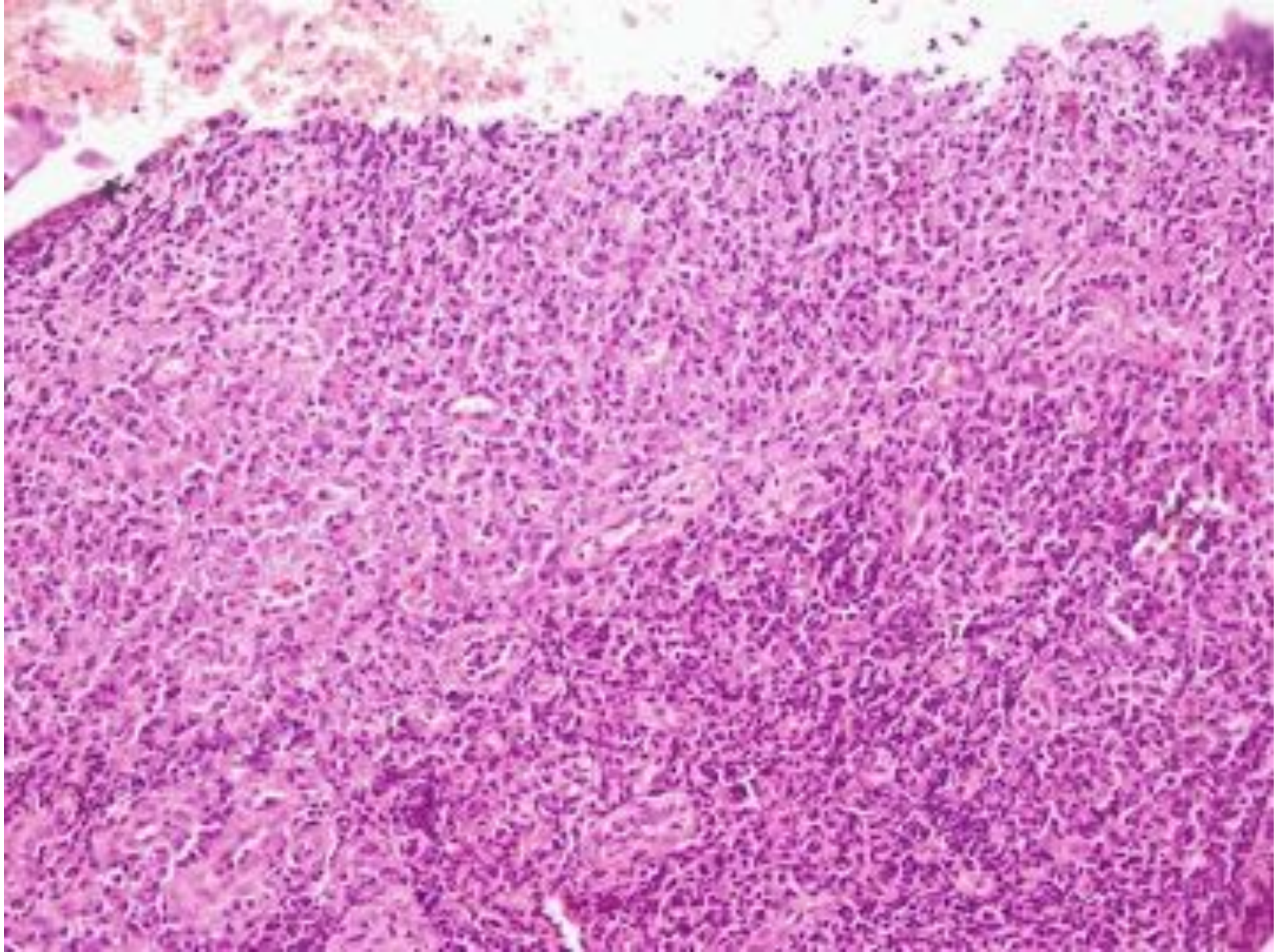
- ►USG Abdomen- Well defined ovoid hypoechoic mass with multiple internal vascular channels in right hemipelvis
- ►CE MRI- Well defined enhancing mass lesion in right hemipelvis with loco-regional
- lymphadenopathy
- ►PET/CT- Faintly FDG avid heterogeneously enhancing soft tissue mass (6.4×4.8 ×5.8 cm) in
- right side of pelvic mesentery. Focal calcification. Adjoining fat planes preserved. Few non FDG avid enlarged (1.2 cm×1.4 cm) right external iliac nodes,

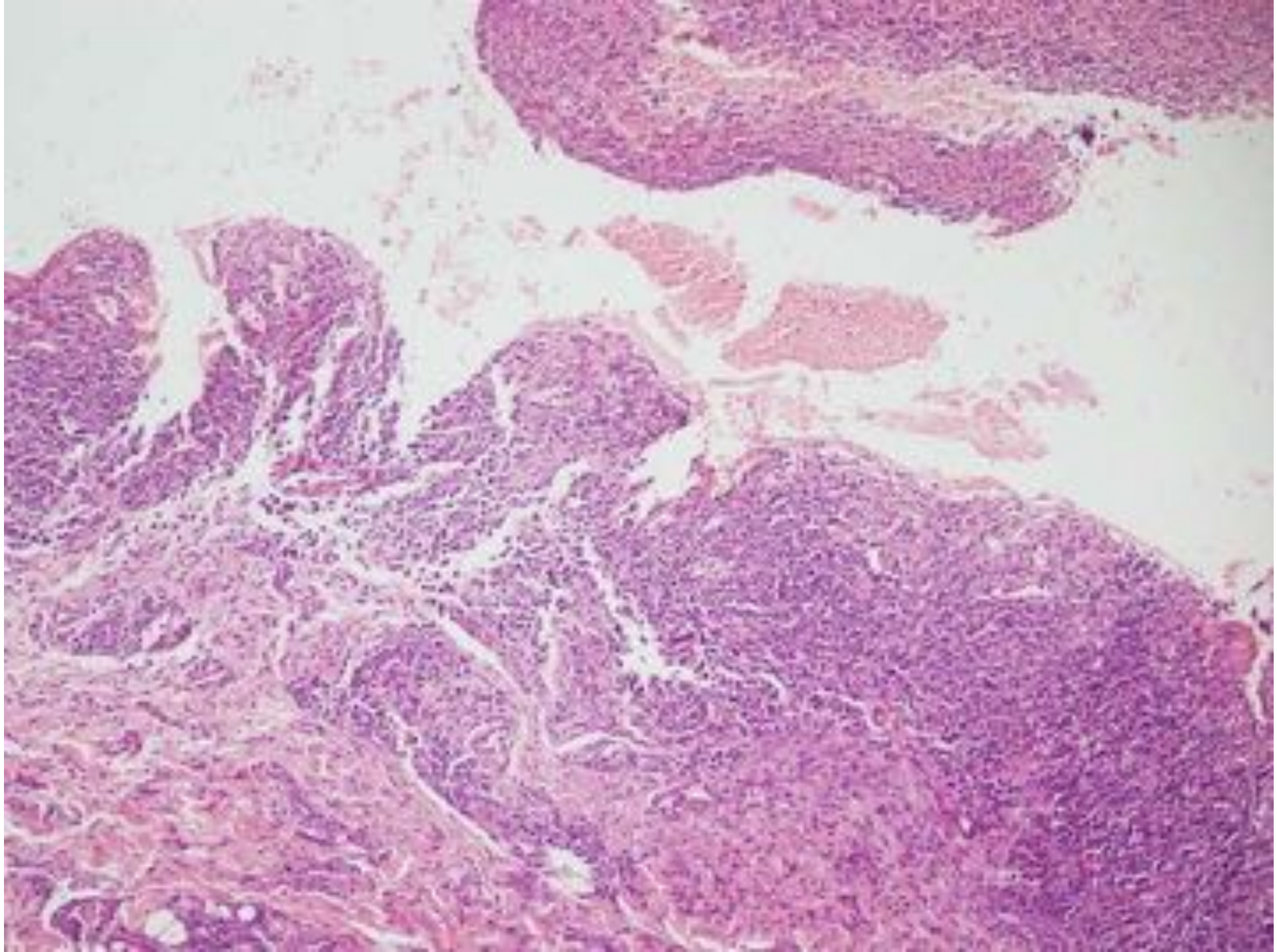
Mucosal & Skin biopsies

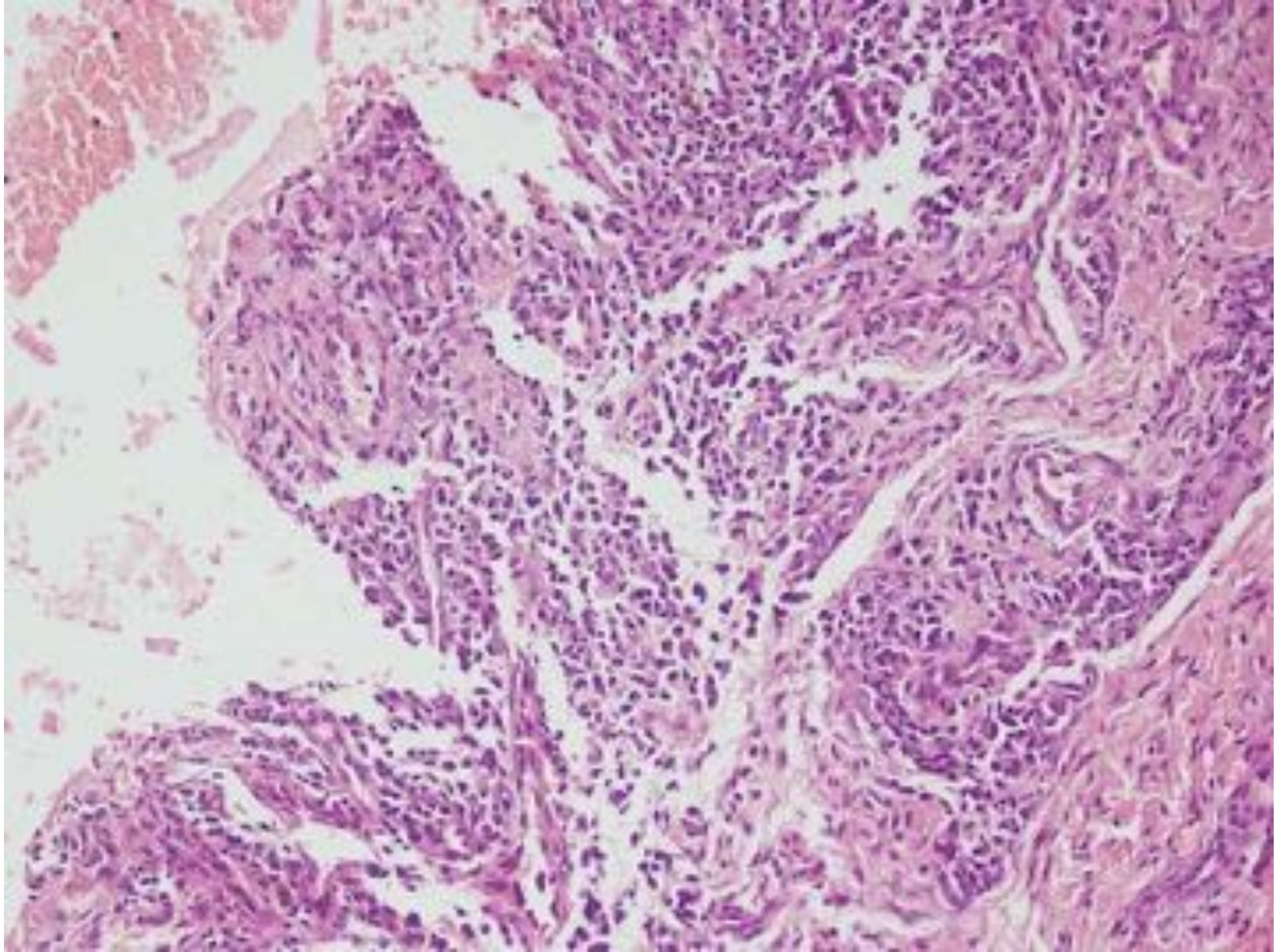


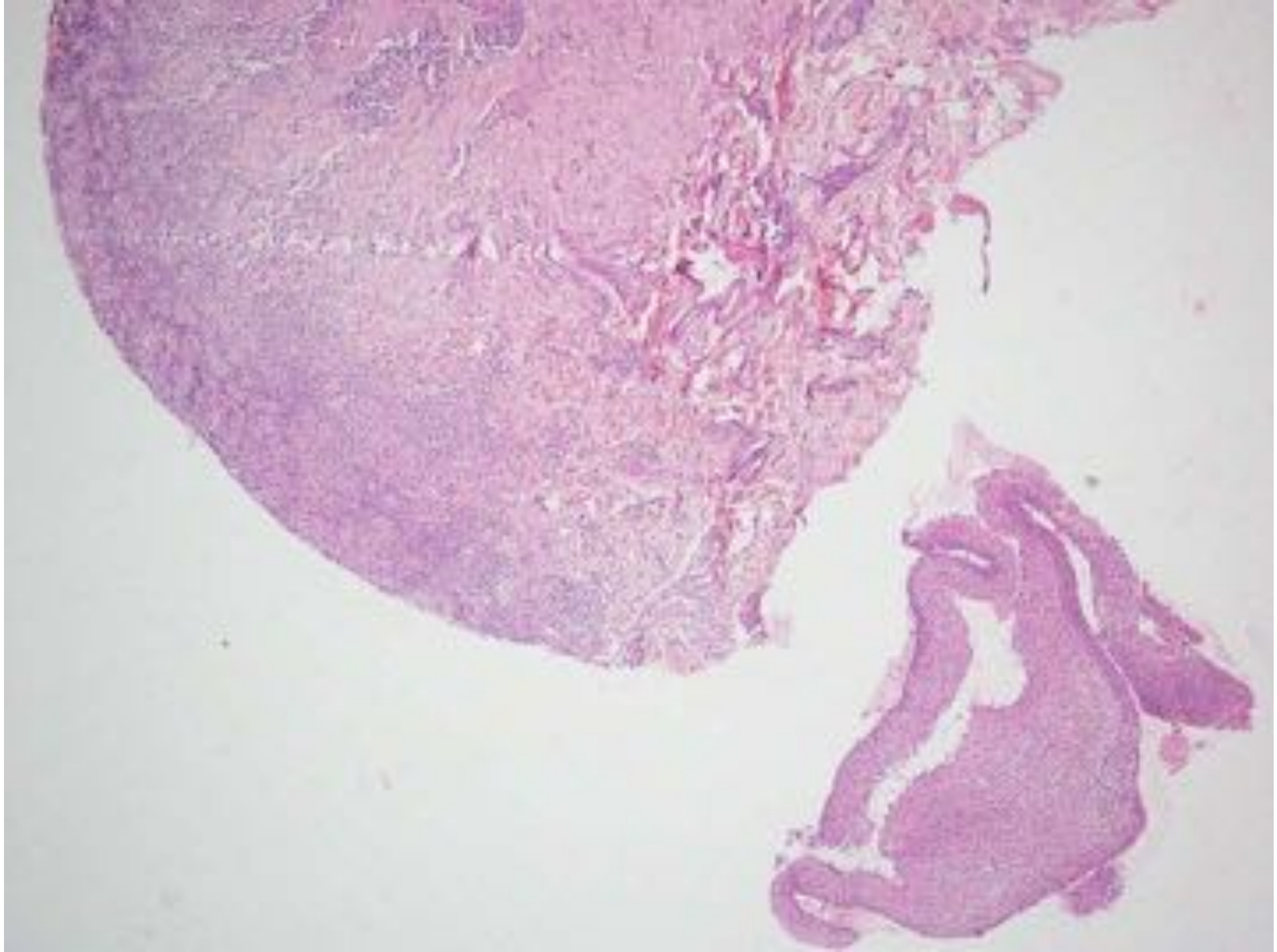


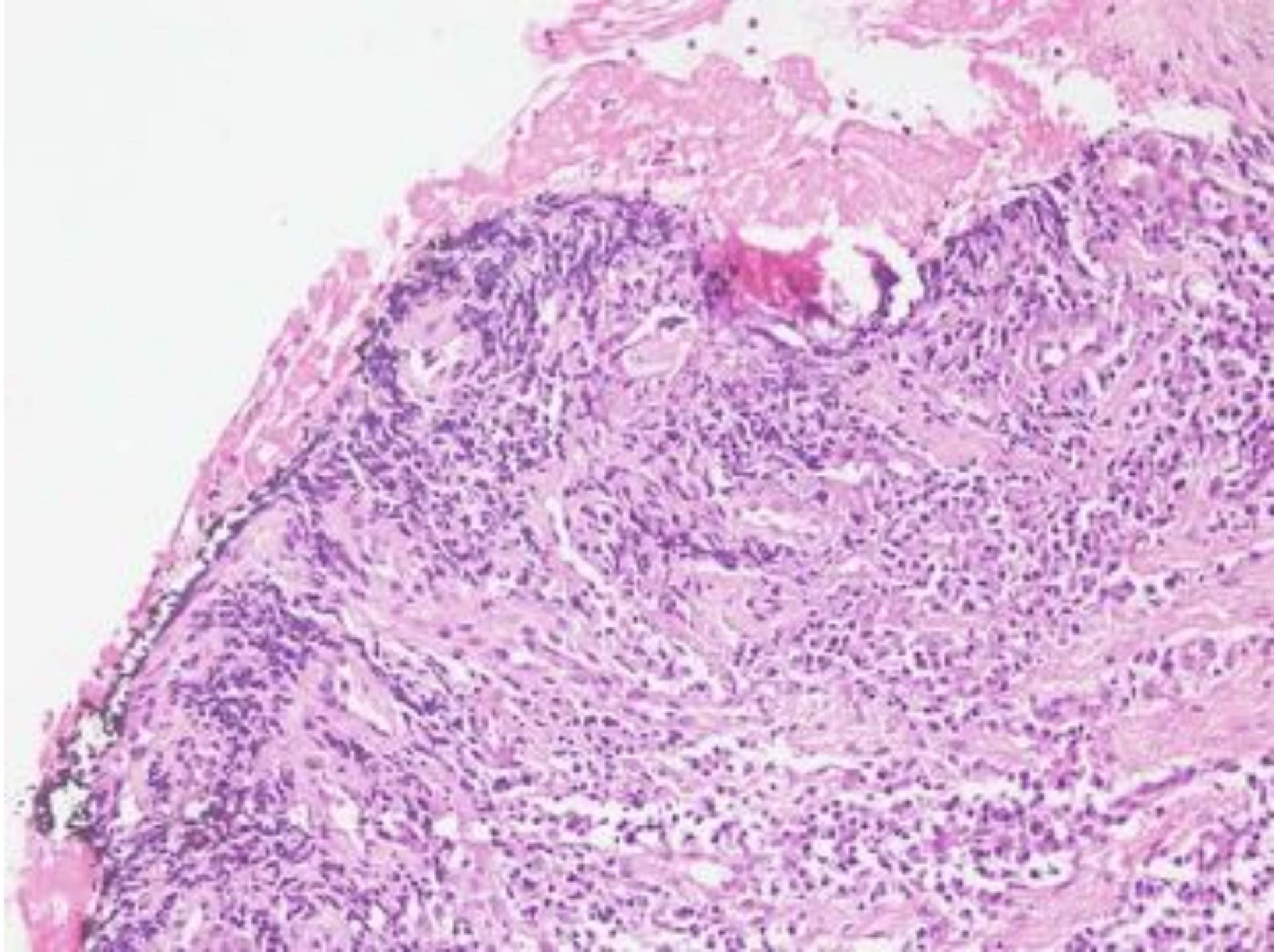


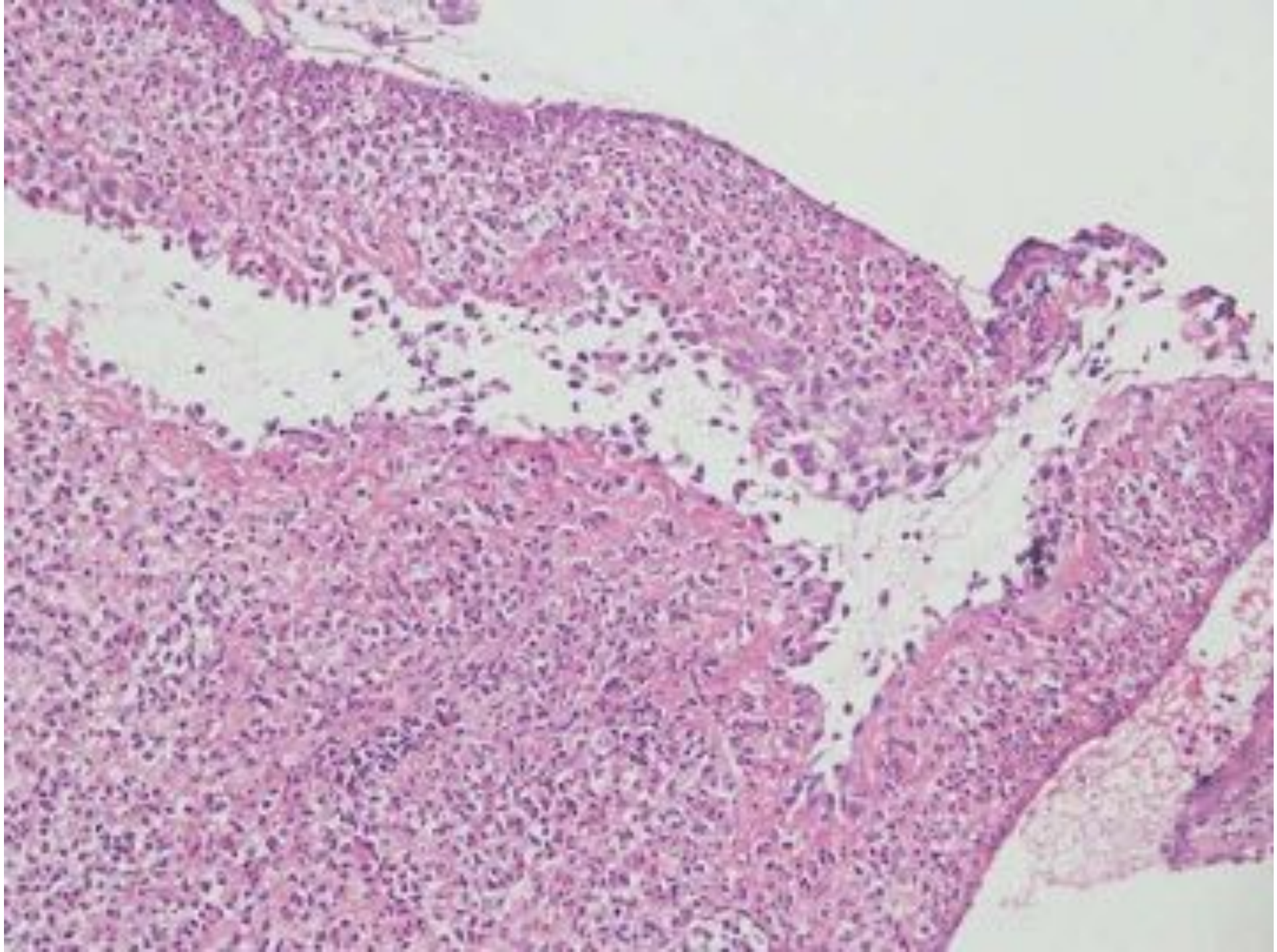


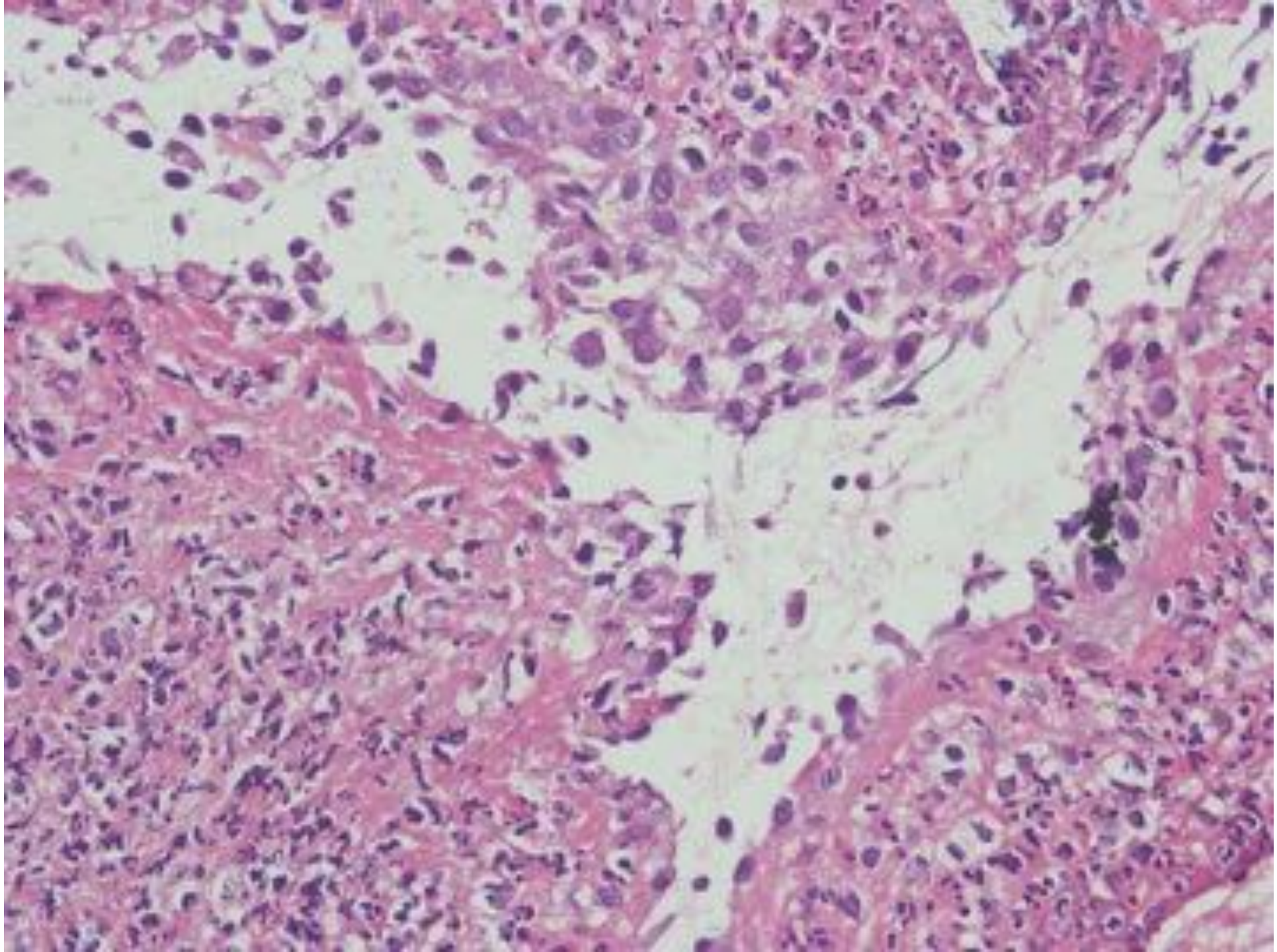










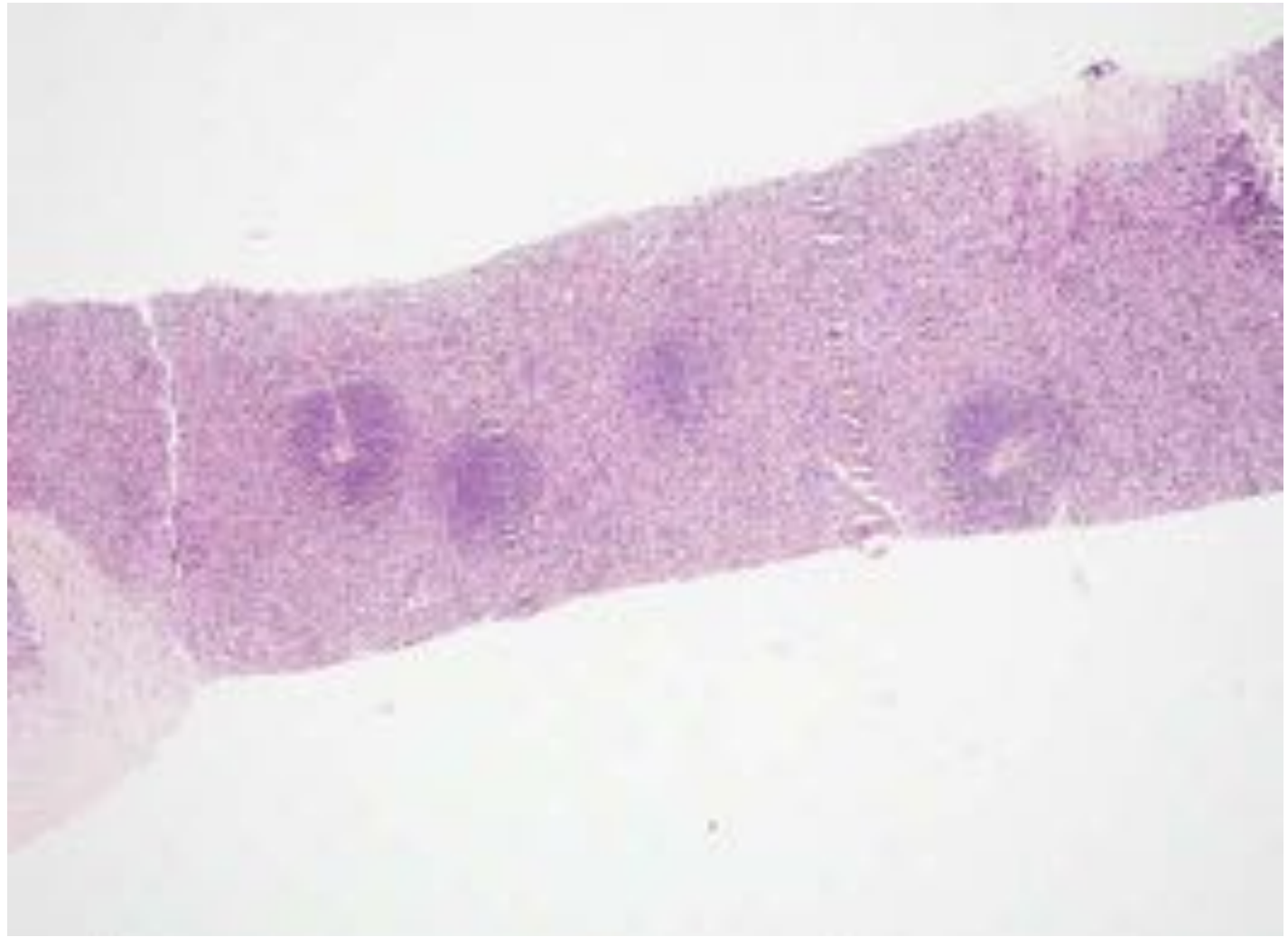


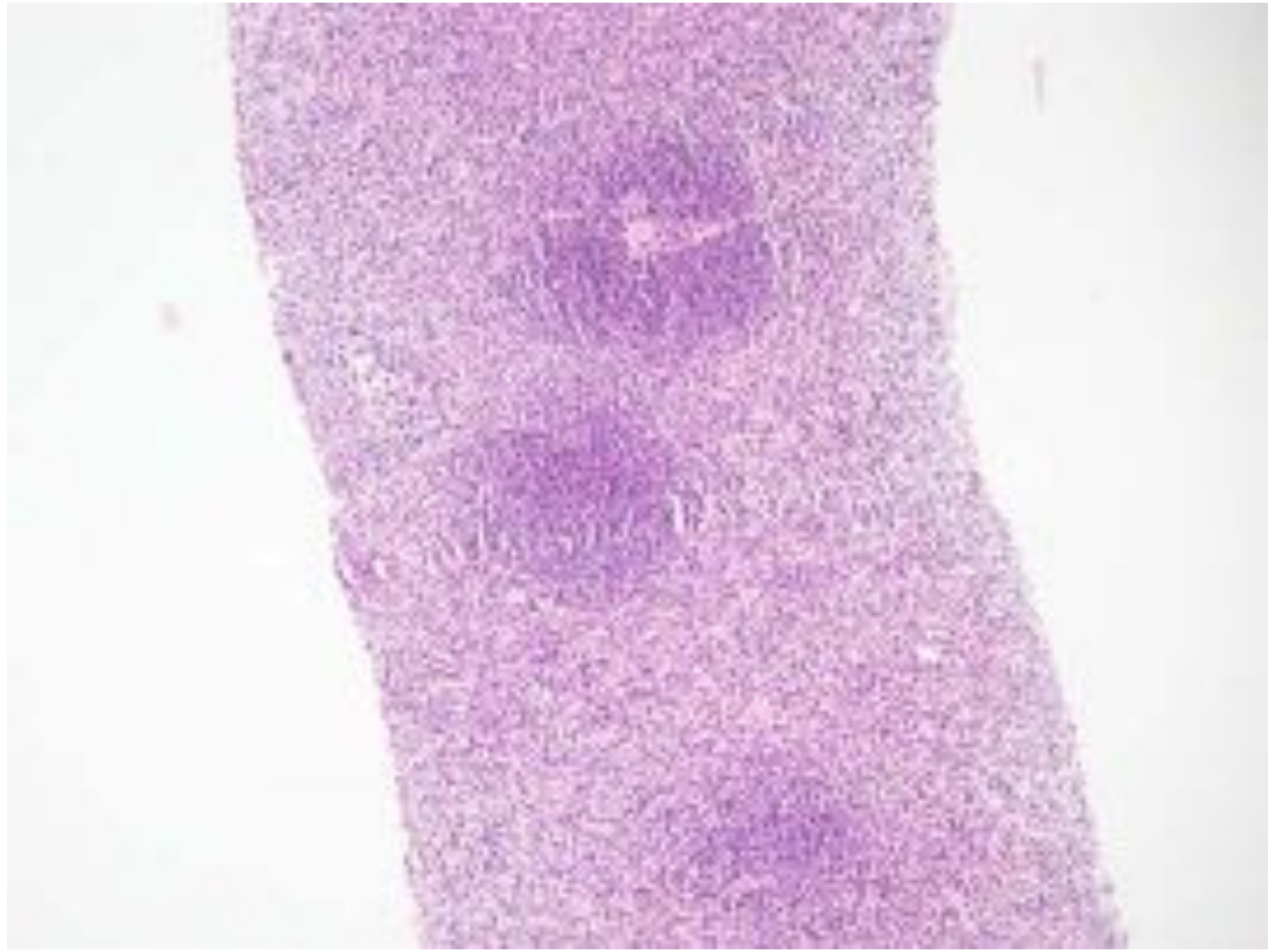
Two samples for DIF from mucosa

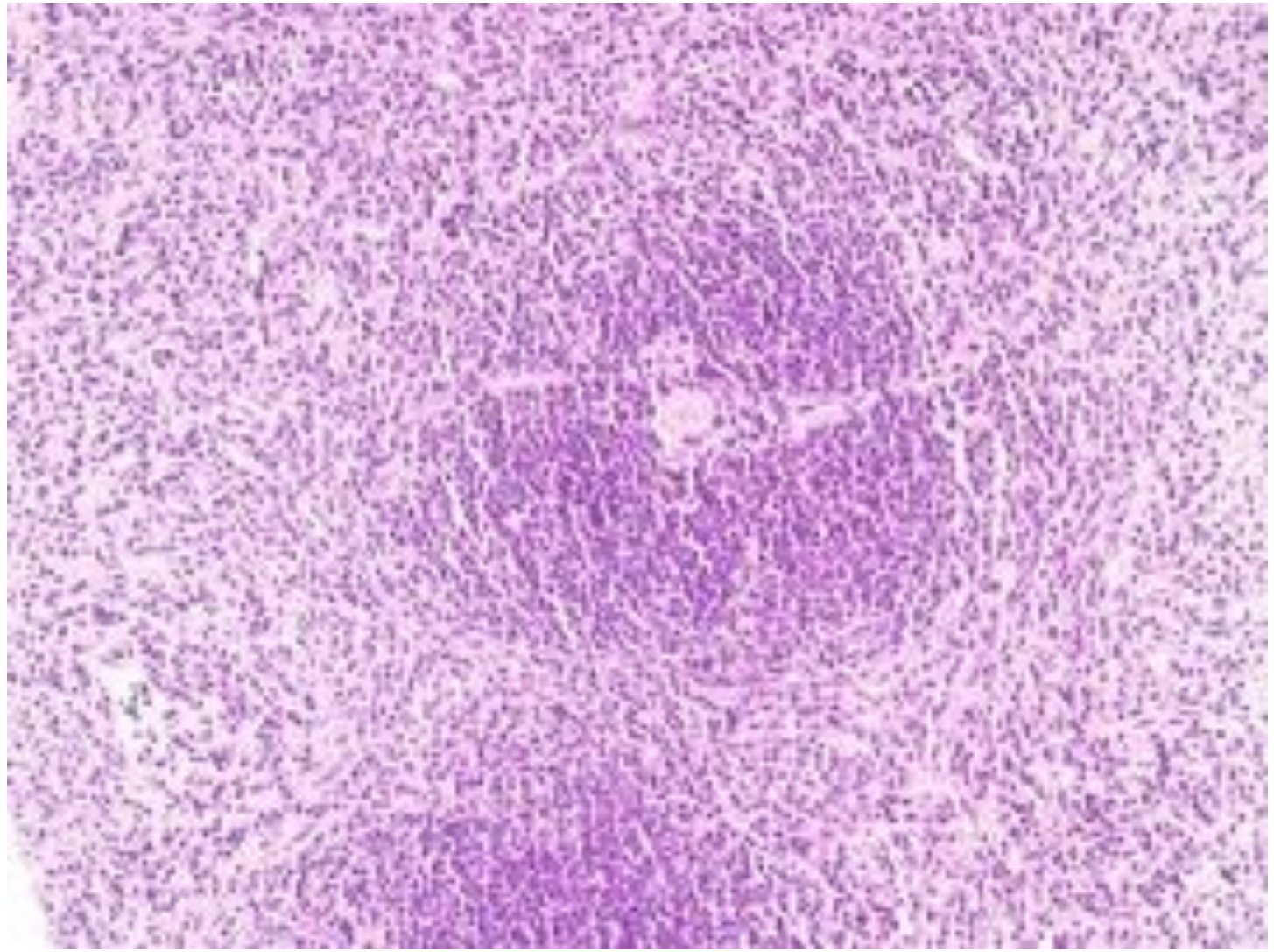
- No epithelium noted

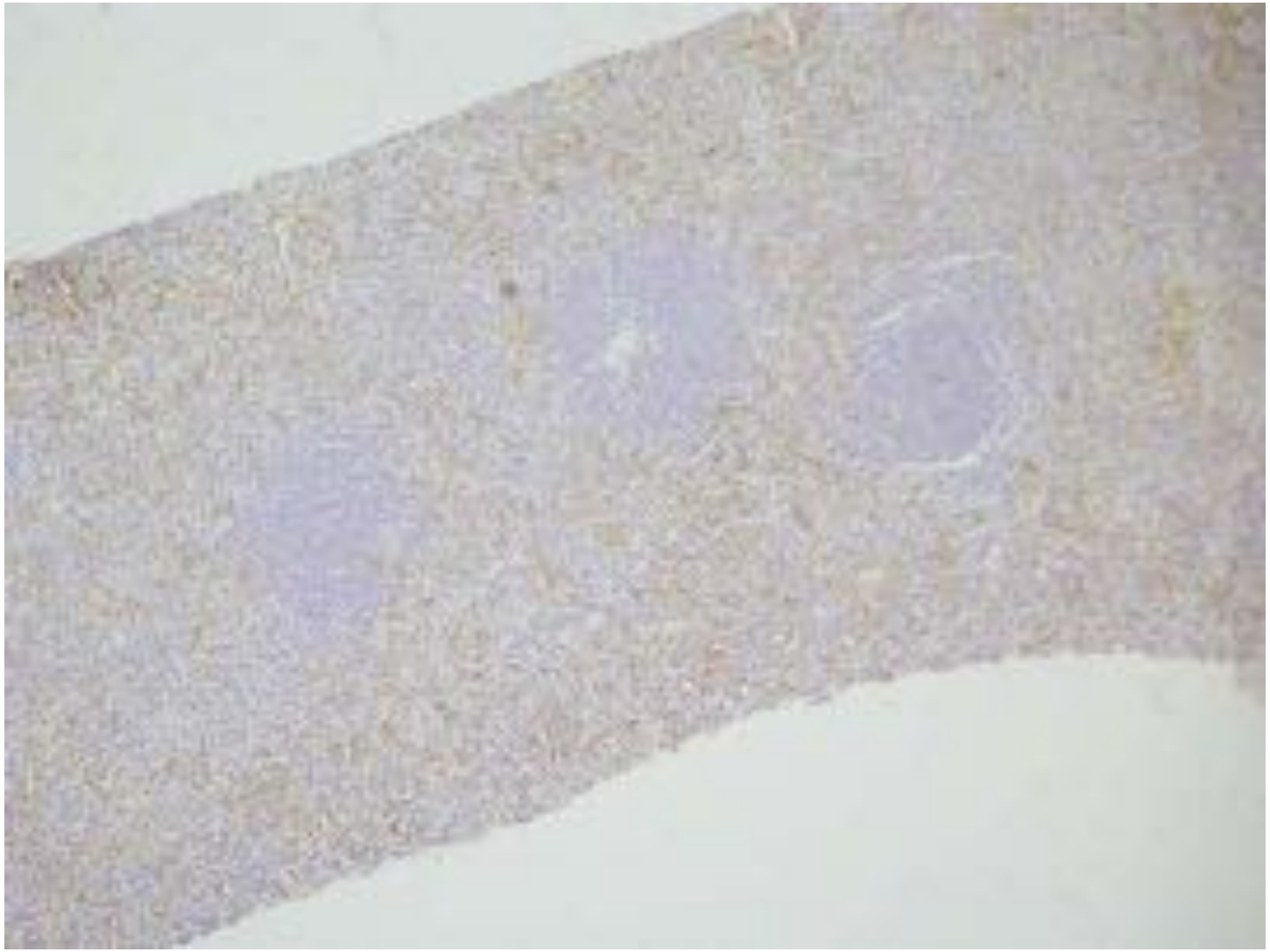
- Third sample for DIF
  - ►Ig G – 2+ TO 3+ ICS
  - ►Ig A – Negative
  - ►Ig M – Negative
  - ►C3 - Negative

Abdominal mass – core biopsy-1

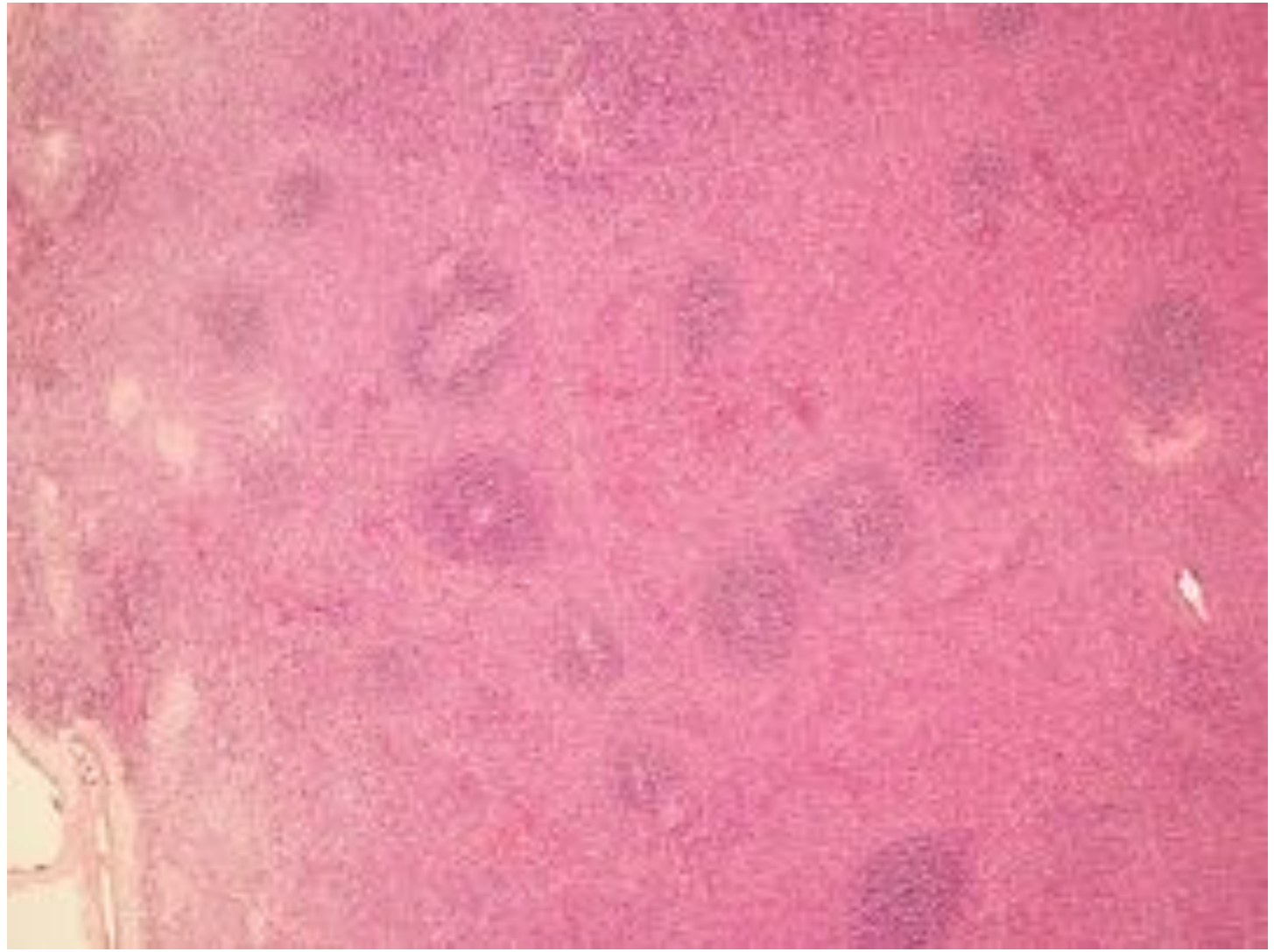


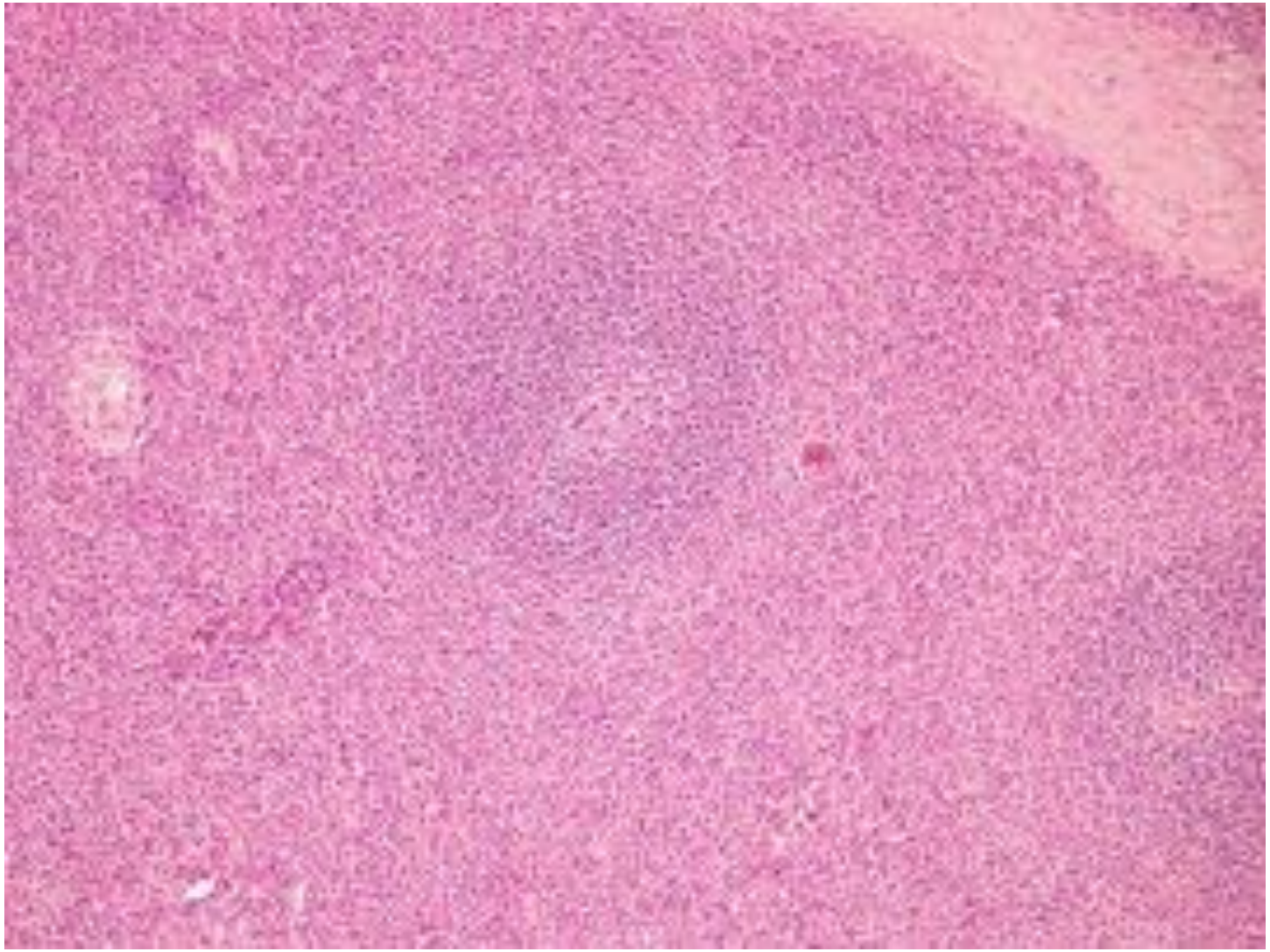


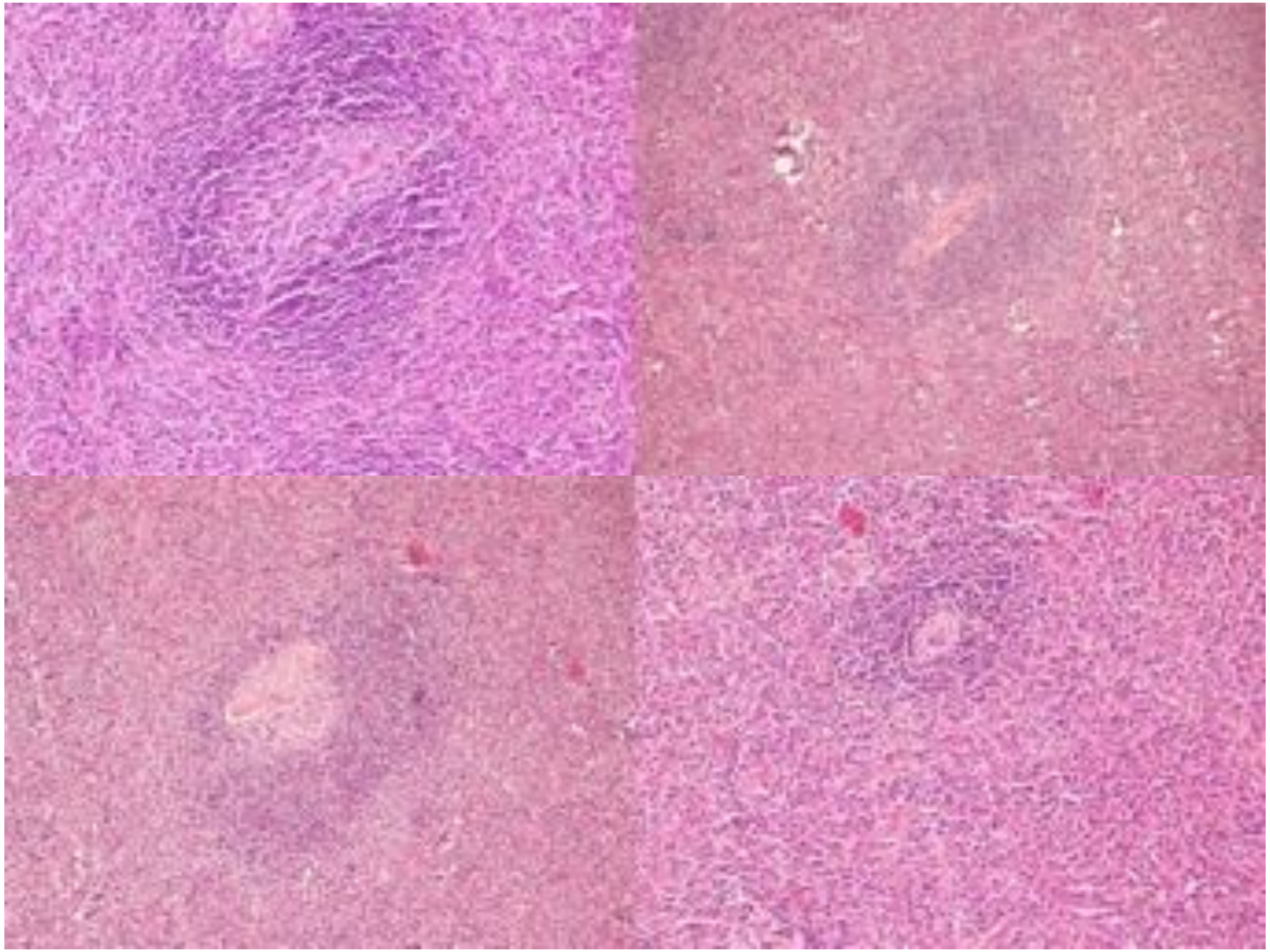


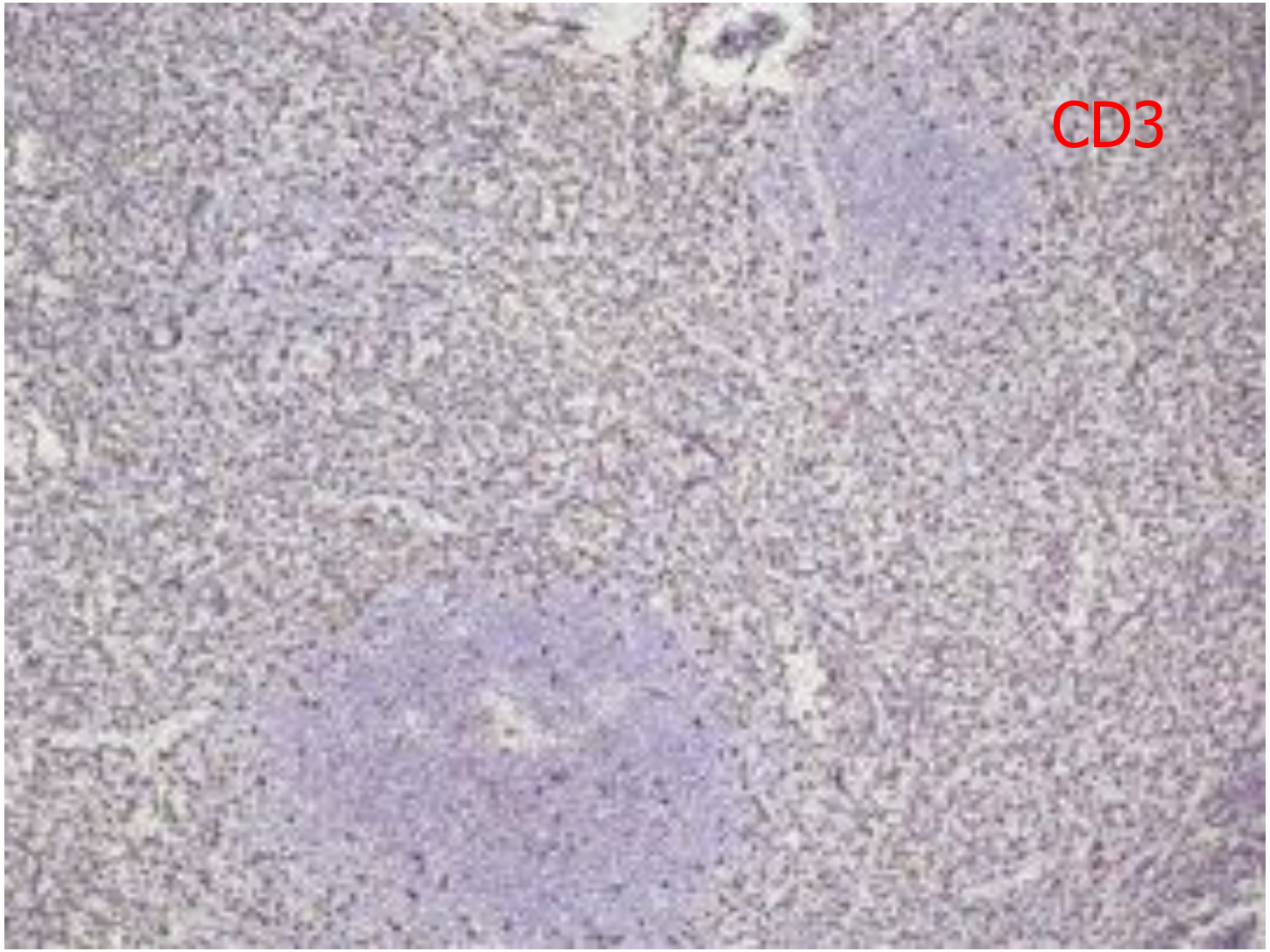


Abdominal mass –core biopsy-2








CD3

DIAGNOSIS:
Castleman’s disease, hyaline vascular type with paraneoplastic pemphigus

CASE 4

DR VINUTHA RANGAPPA

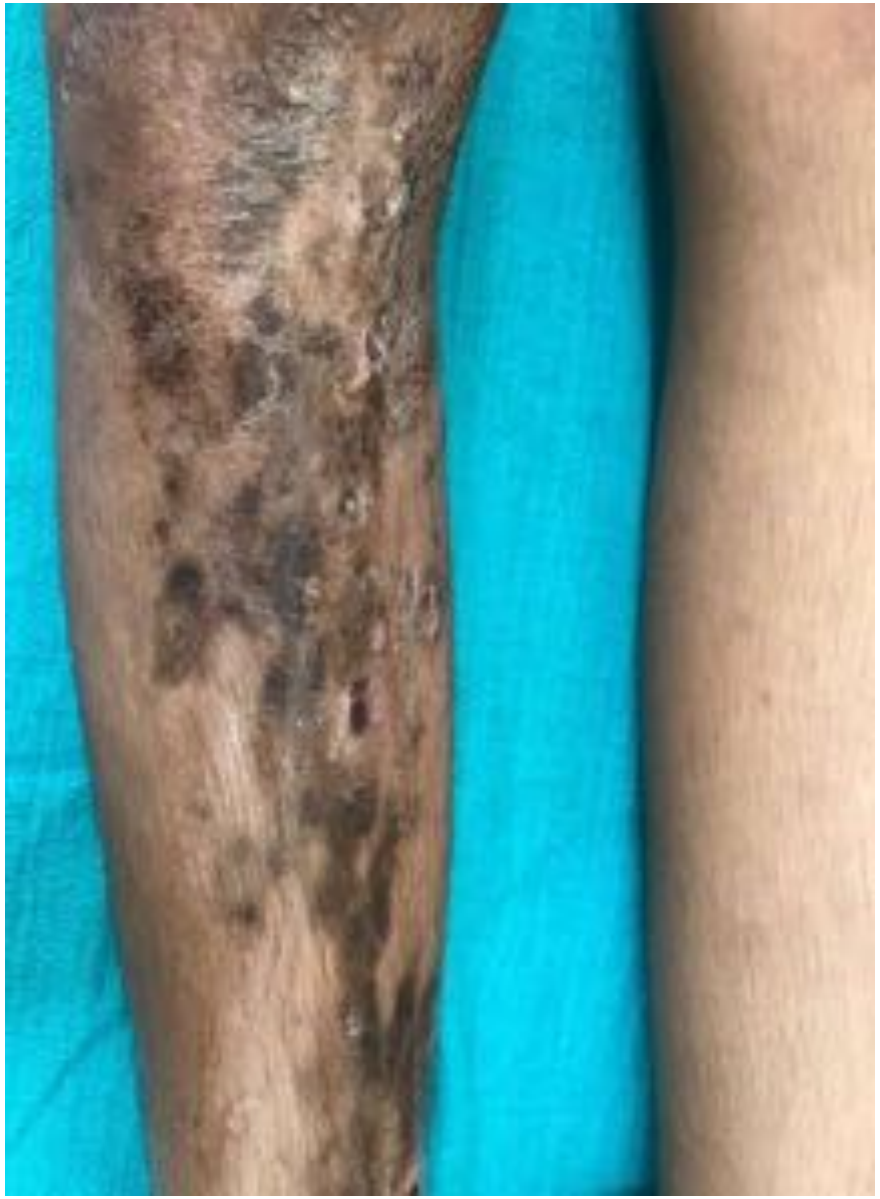
Case 4

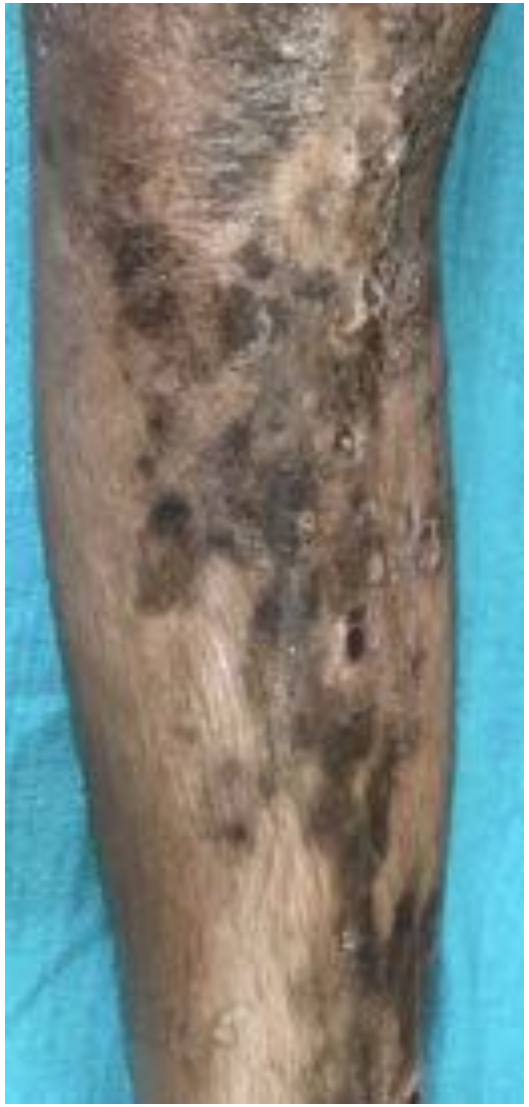
- A 14 year old girl presented with asymptomatic linear white colored lesions initially over right
- lower limb which later involved right side of trunk and right upper limb in a span of one year.
- Lesions later turned dark in color along with

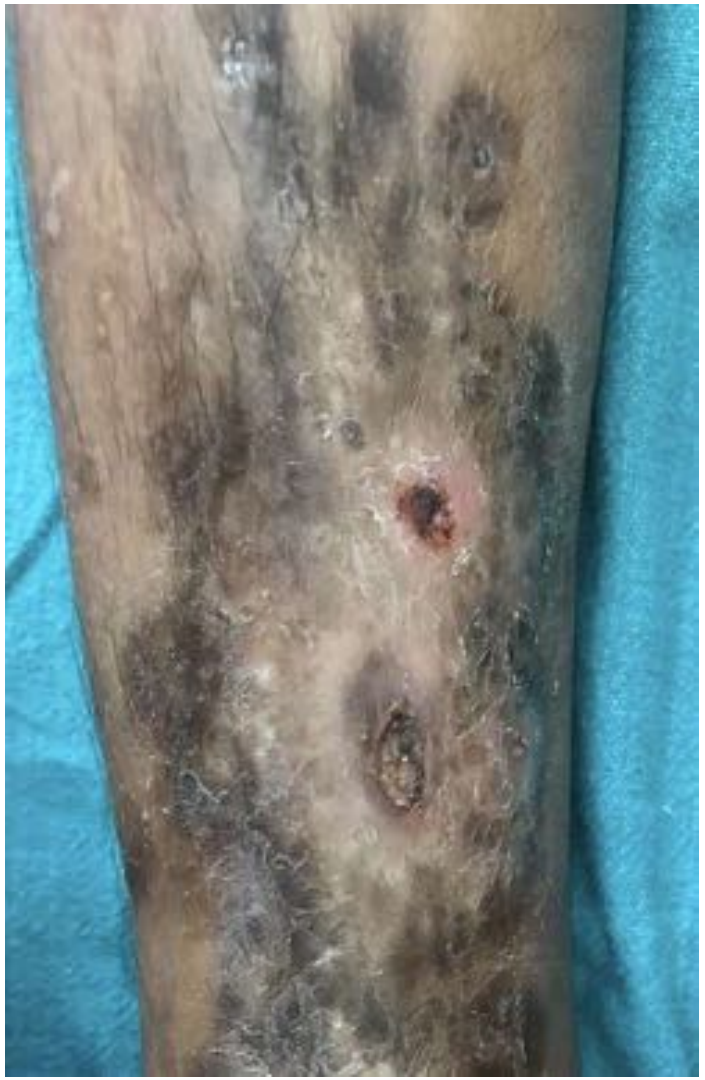
  hardening of skin. Also developed ulceration over

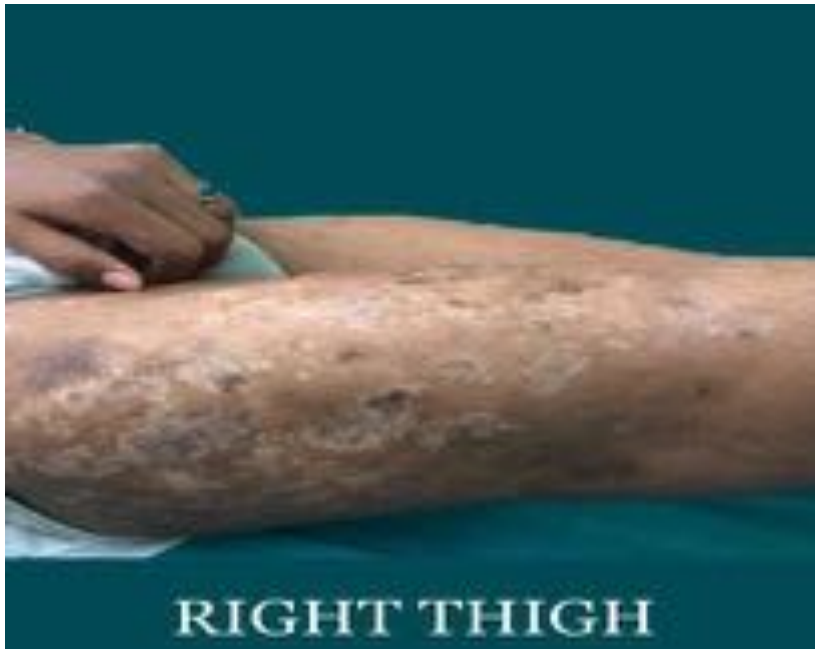
  right lower limb.

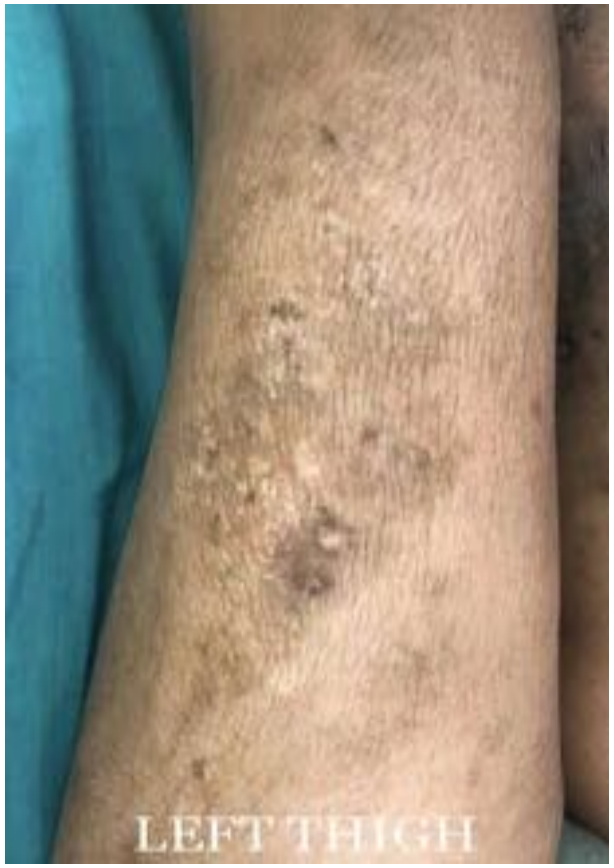
RIGHT LEG



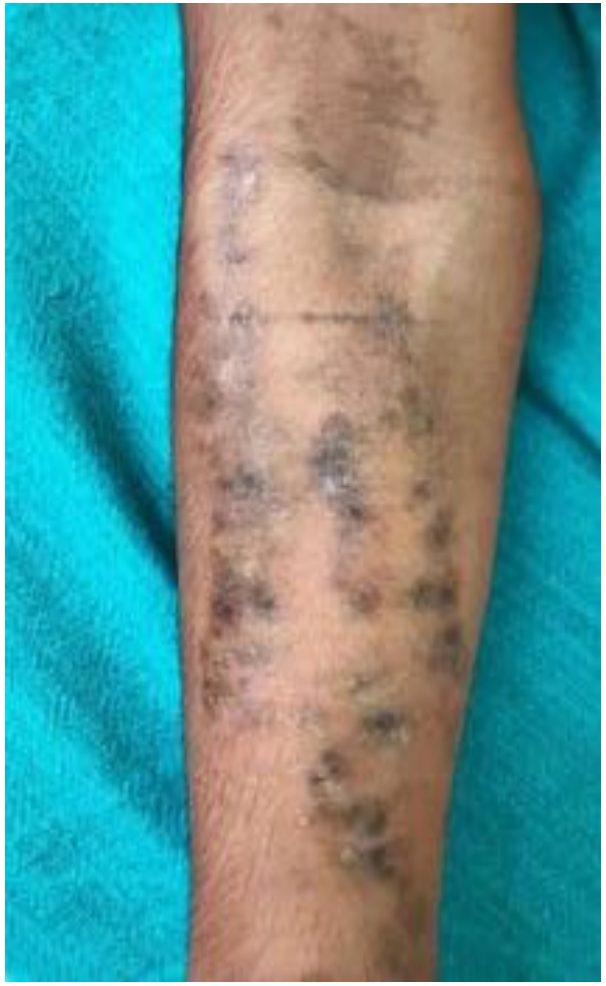


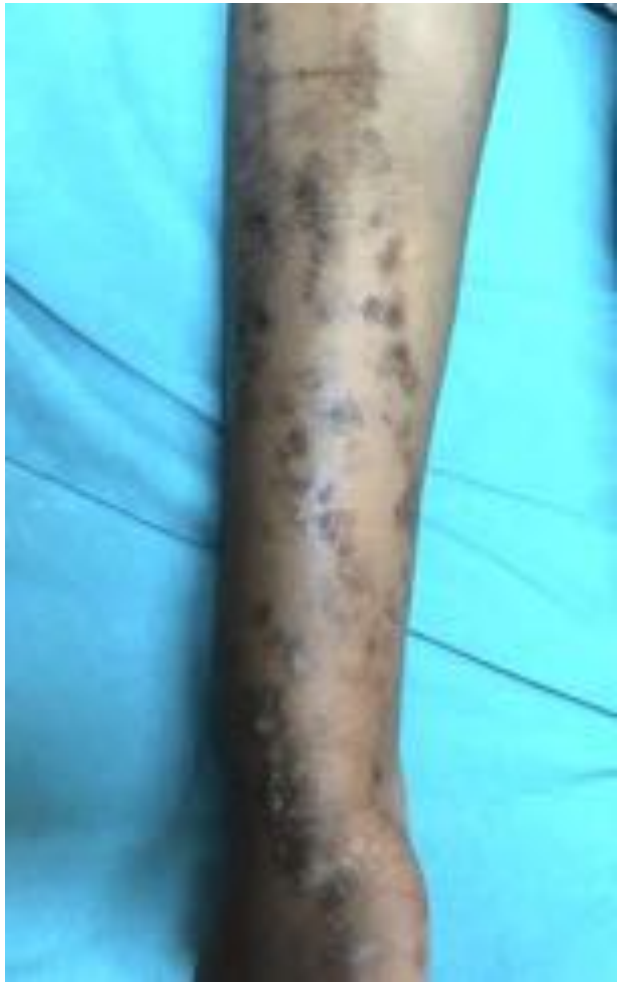


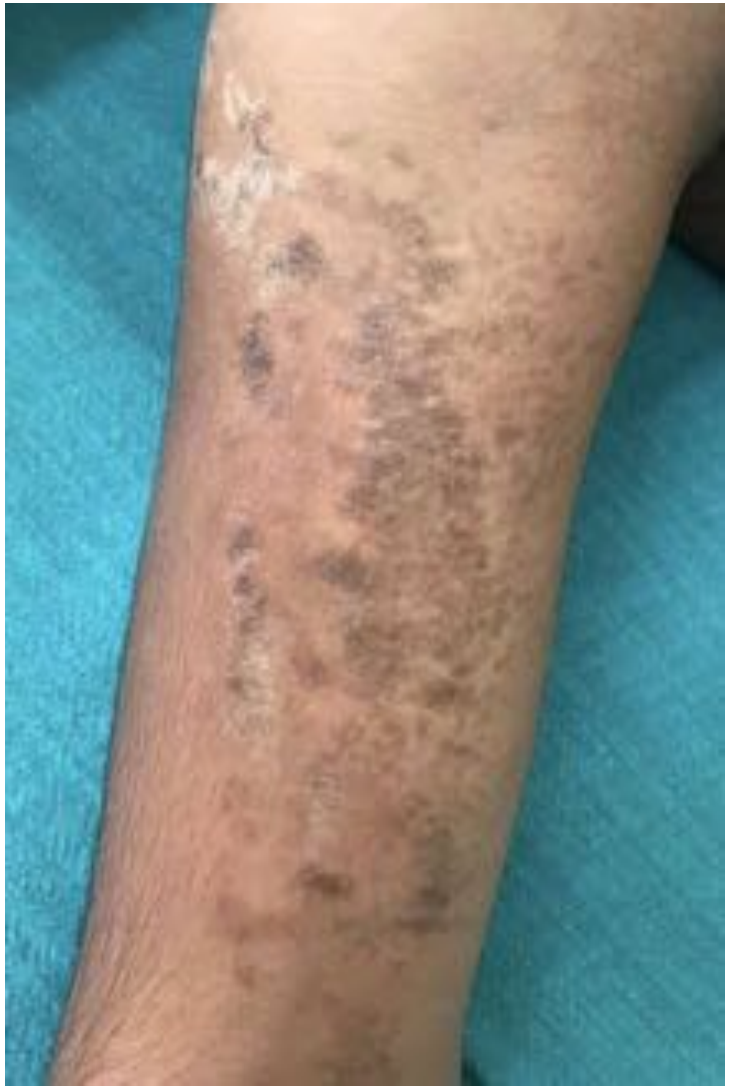

RIGHT THIGH


LEFT THIGH

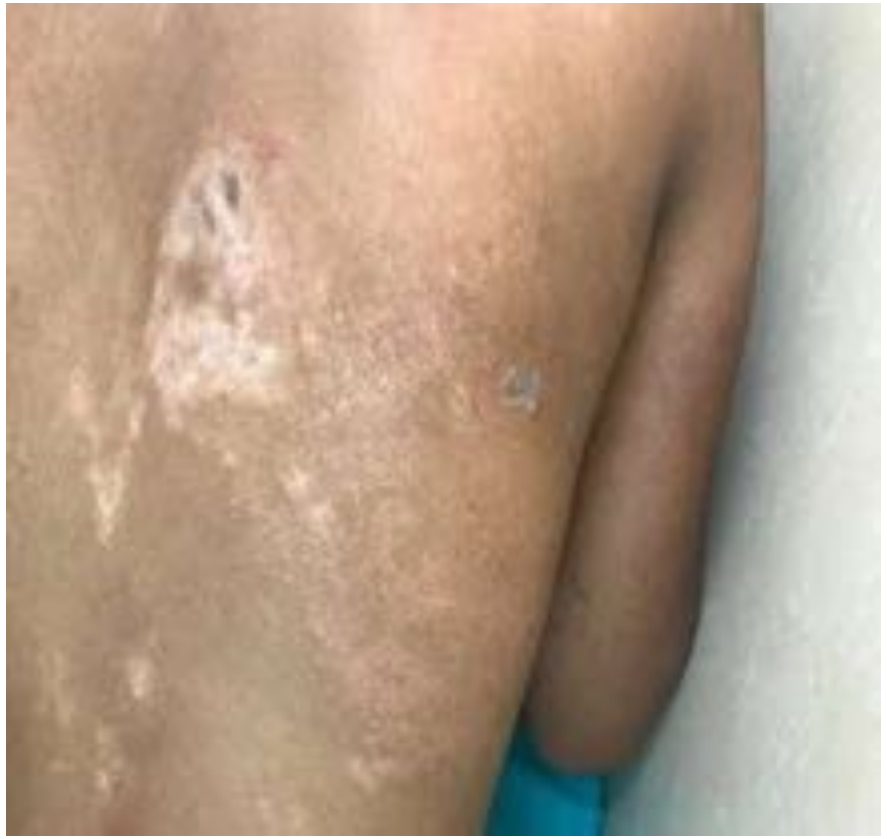
RIGHT UPPER LIMB

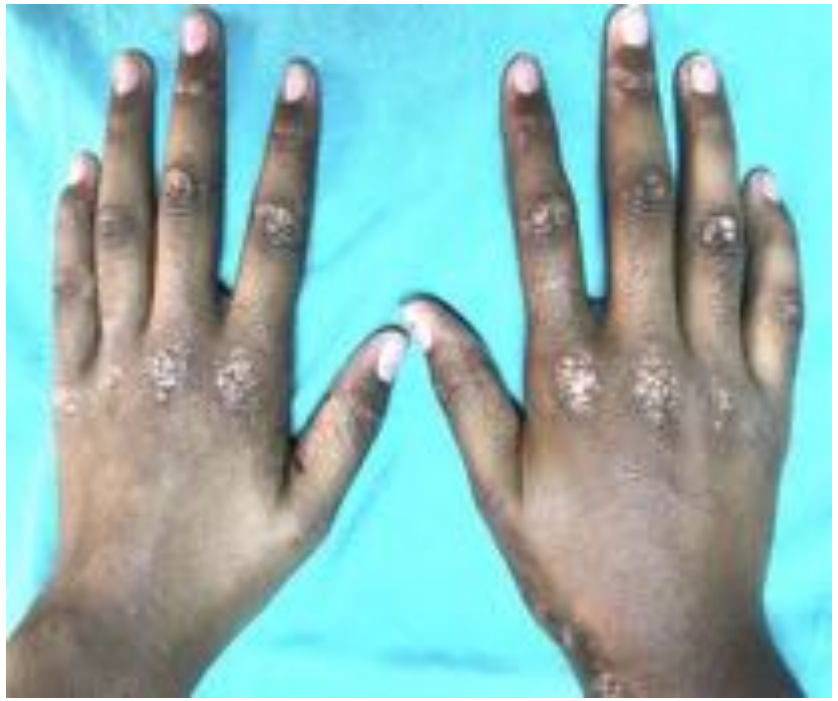


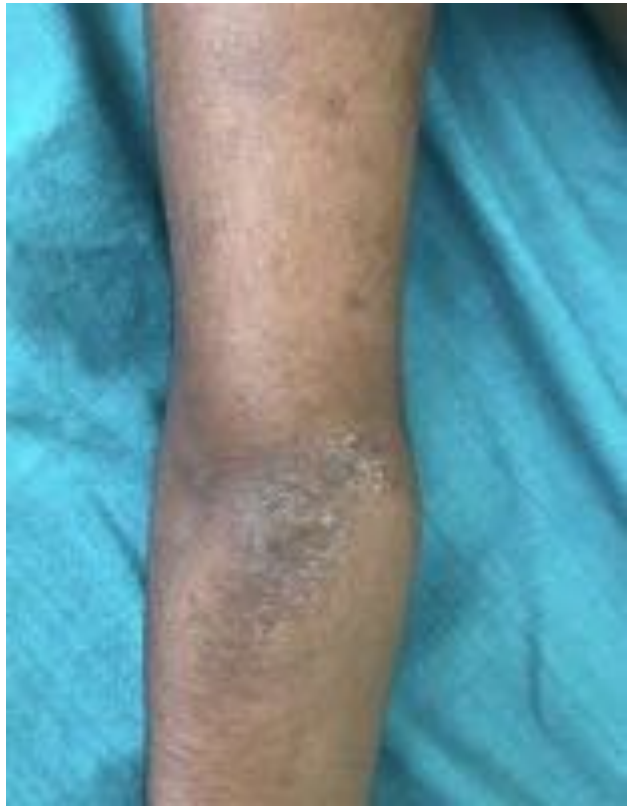




RIGHT SIDE OF TRUNK AND NECK







OTHER RELEVANT HISTORY

- There was presence of joint pain involving wrist and knee joints.
- No h/o pain over small joints or history suggestive of Raynaud's phenomenon .
- No h/o pain involving proximal muscles.
- Treatment history- She was treated with topical psoralen and sunlight exposure prior to our visit.

  After which she developed burning sensation and redness.
- There was no other significant previous medical or family history.

EXAMINATION POINTS

- lesions were mostly confined to right side of the trunk and right upper, lower limb.
- There was induration of skin over lower and upper limb lesions.
- Few hypopigmented lesions were following the lines of Blaschko.
- Tenderness was present over ulcerations.
- There was no muscle tenderness and joint examination was normal.

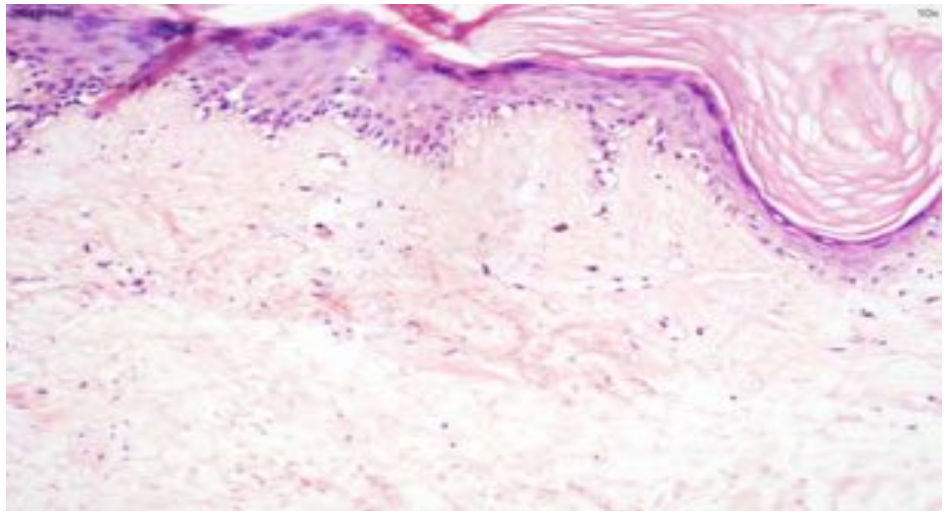
What could be the differentials?

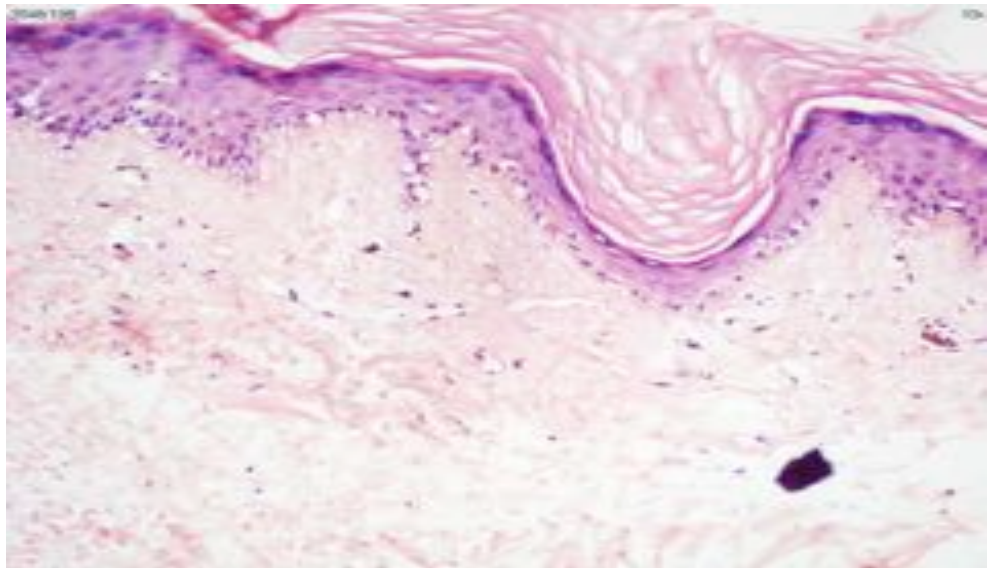
- **DIFFERENTIALS:**
- Linear extragenital Lichen sclerosus et atrophicus .
- Juvenile Dermatomyositis.
- Linear morphea.

SIGNIFICANT BASIC INVESTIGATIONS

- Hb –10.4g/dl, Total count –9,600cells/cm, Differential count –normal
- ESR –110
- Liver function test, Renal function test were within normal limits.
- ANA –Positive, homogenous type grade 1
- Muscle enzymes were normal.

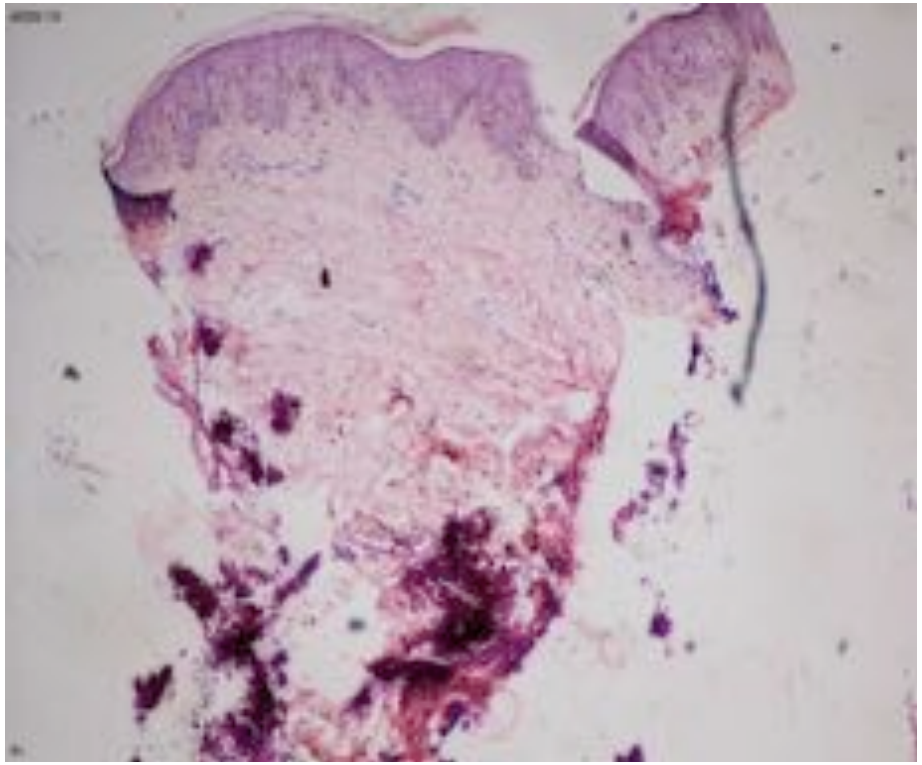
- Skin biopsy from hypopigmented lesions –epidermis shows mild irregular hyperplasia, follicular plugging with basal cell vacuolar degeneration. Dermis shows sparse superficial and mild perivascular lymphocytic infiltrate.





•Skin Biopsy from indurated plaques –epidermis with mild acanthosis. Dermis shows homogenized collagen and areas of basophilic calcium deposits.

•Calcinosis cutis



- Electromyography was normal.
- MRI of shoulder muscles was normal.
- ANA profile was negative.
- Anti-MDA 5 was positive.
- Chest X-RAY and pulmonary function test –normal.

DIAGNOSIS

- Unilateral linear Lichen sclerosus et atrophicus with Juvenile amyopathic Dermatomyositis

CASE 5

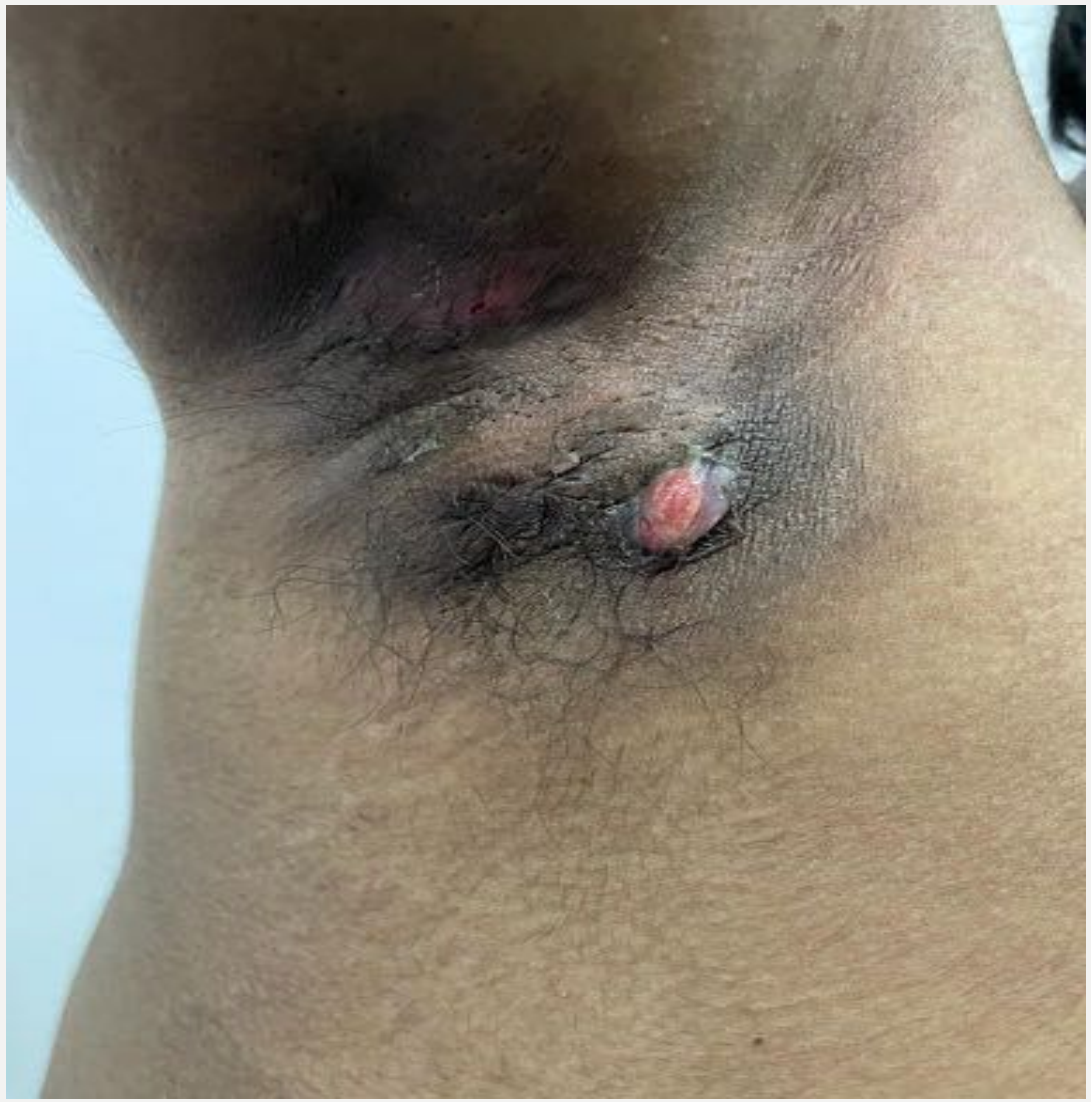
DR. NIBEDITA PATRO

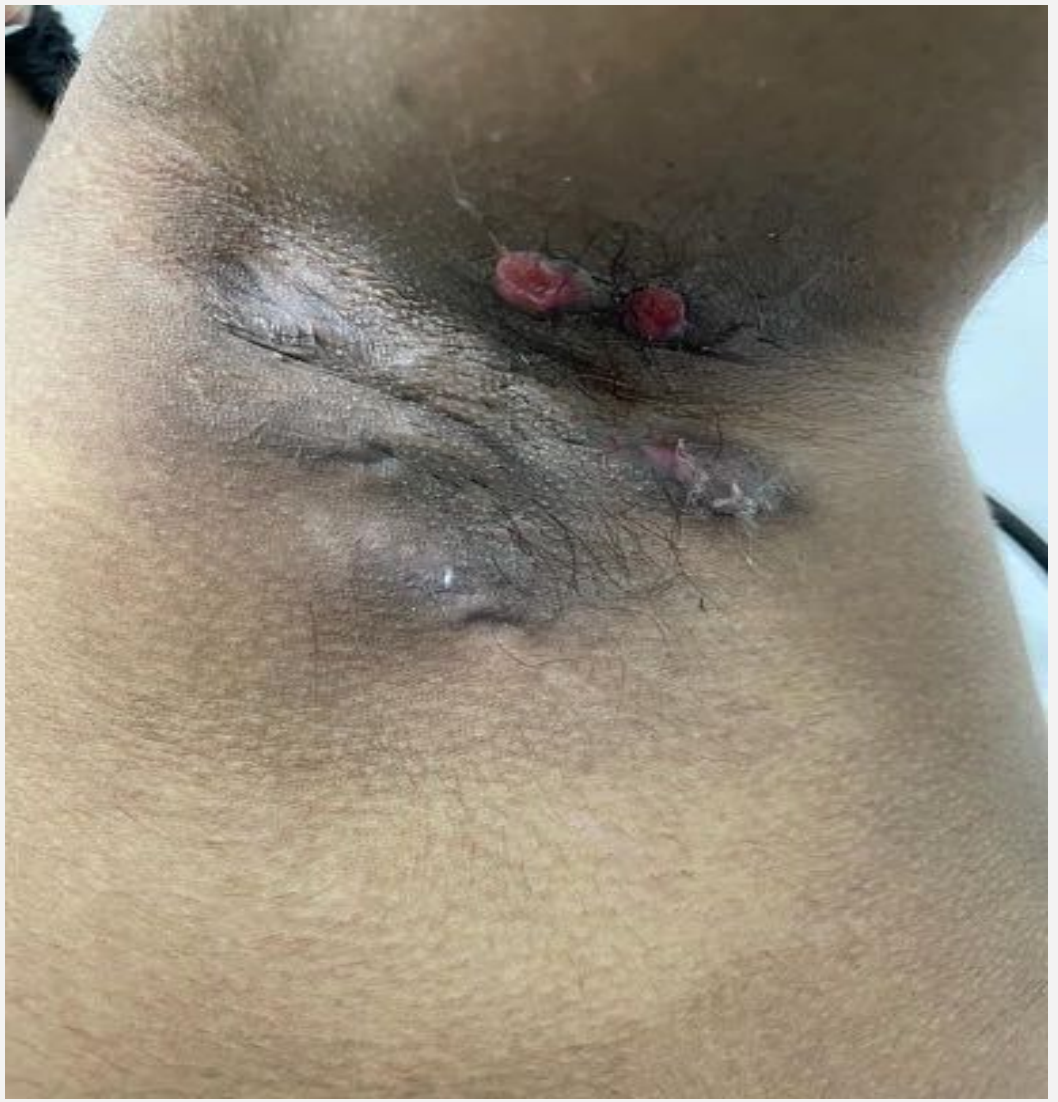
TREATING HIDRADENITIS SUPPURATIVA IN A TEENAGER
A DIFFICULT ROAD AHEAD

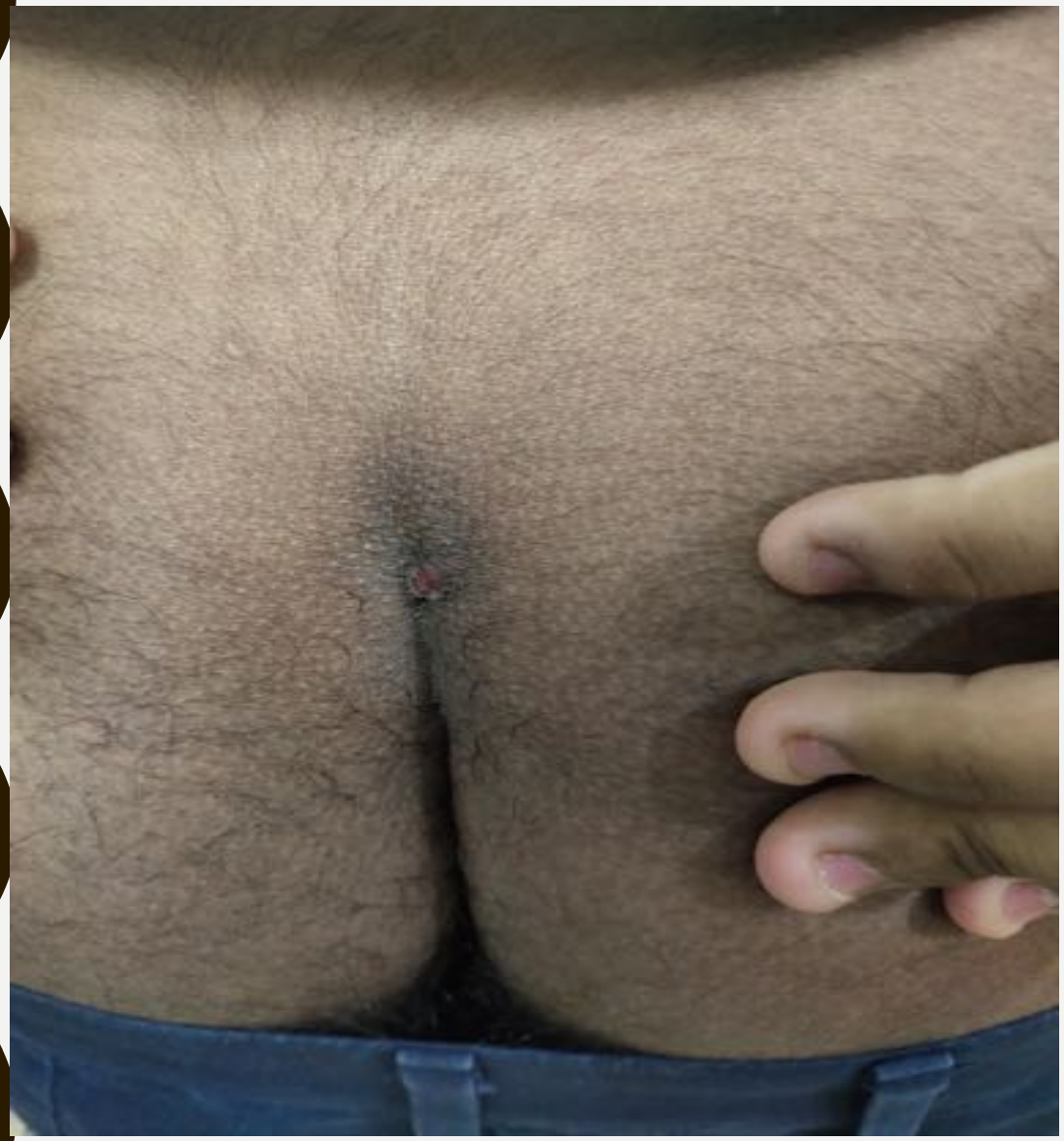
- A 16 years old boy presented with
- Recurrent tender nodules with seropurulent discharge on & off
- Sites - Bilateral axilla and intergluteal folds for 2 years duration
- Not associated with fever, cough, loss of appetite
- Healing with scar formation

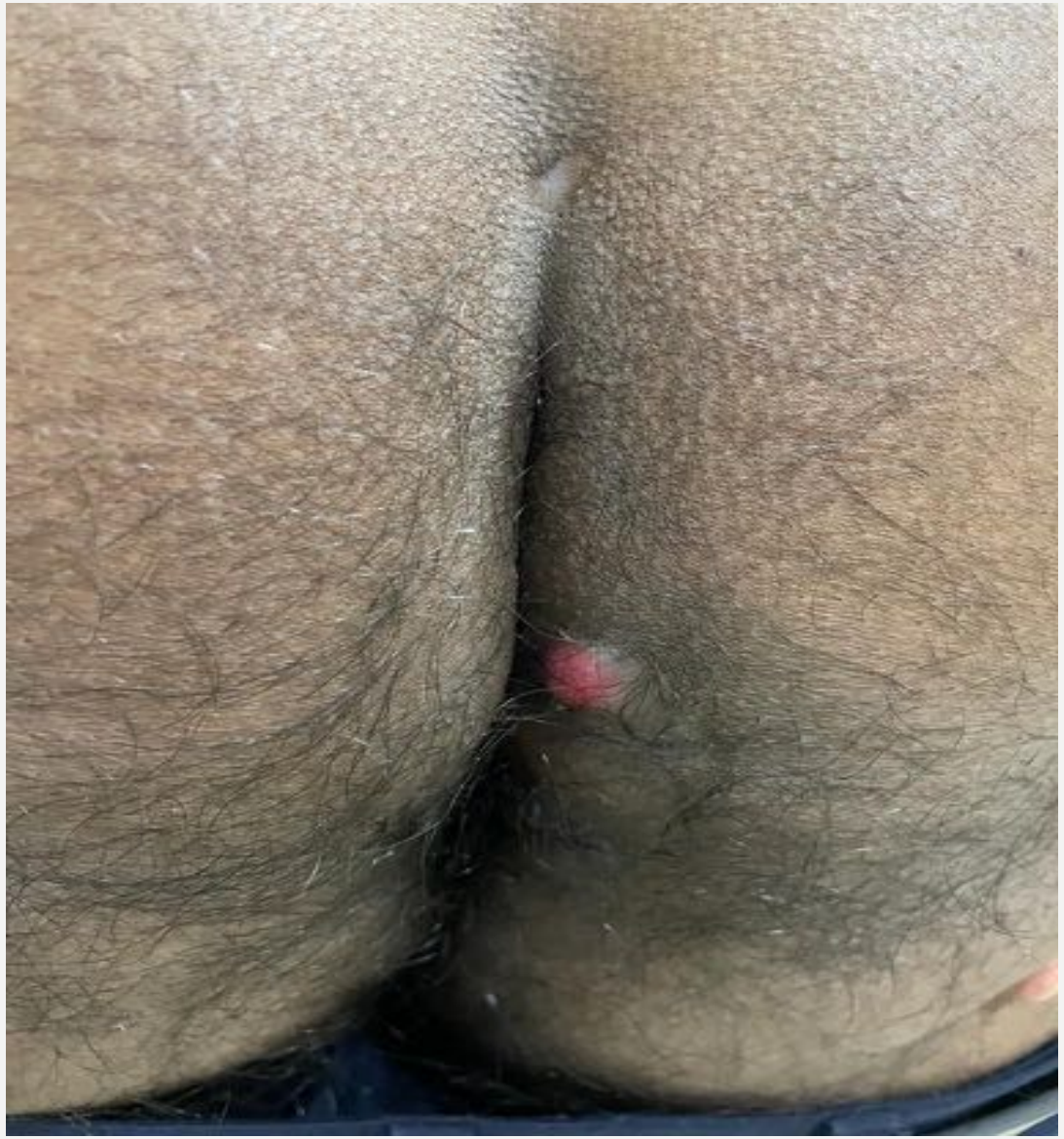
CLINICAL EXAMINATION

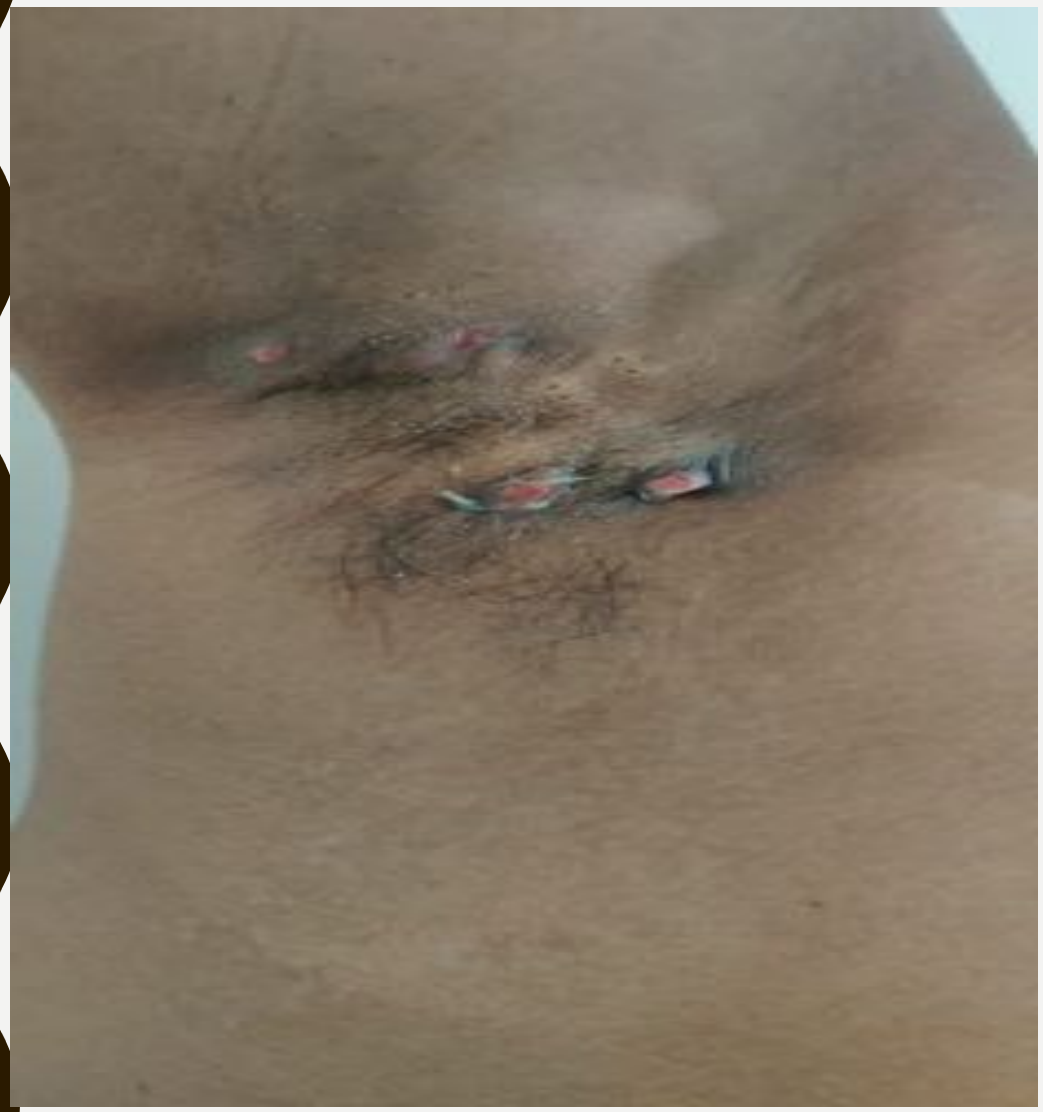
- Multiple erythematous tender nodules, few with granulation tissue present over bilateral axilla and intergluteal folds
- Multiple open macro-comedones seen on bilateral axilla
- Few areas of scarring of old healed lesions present on bilateral axilla

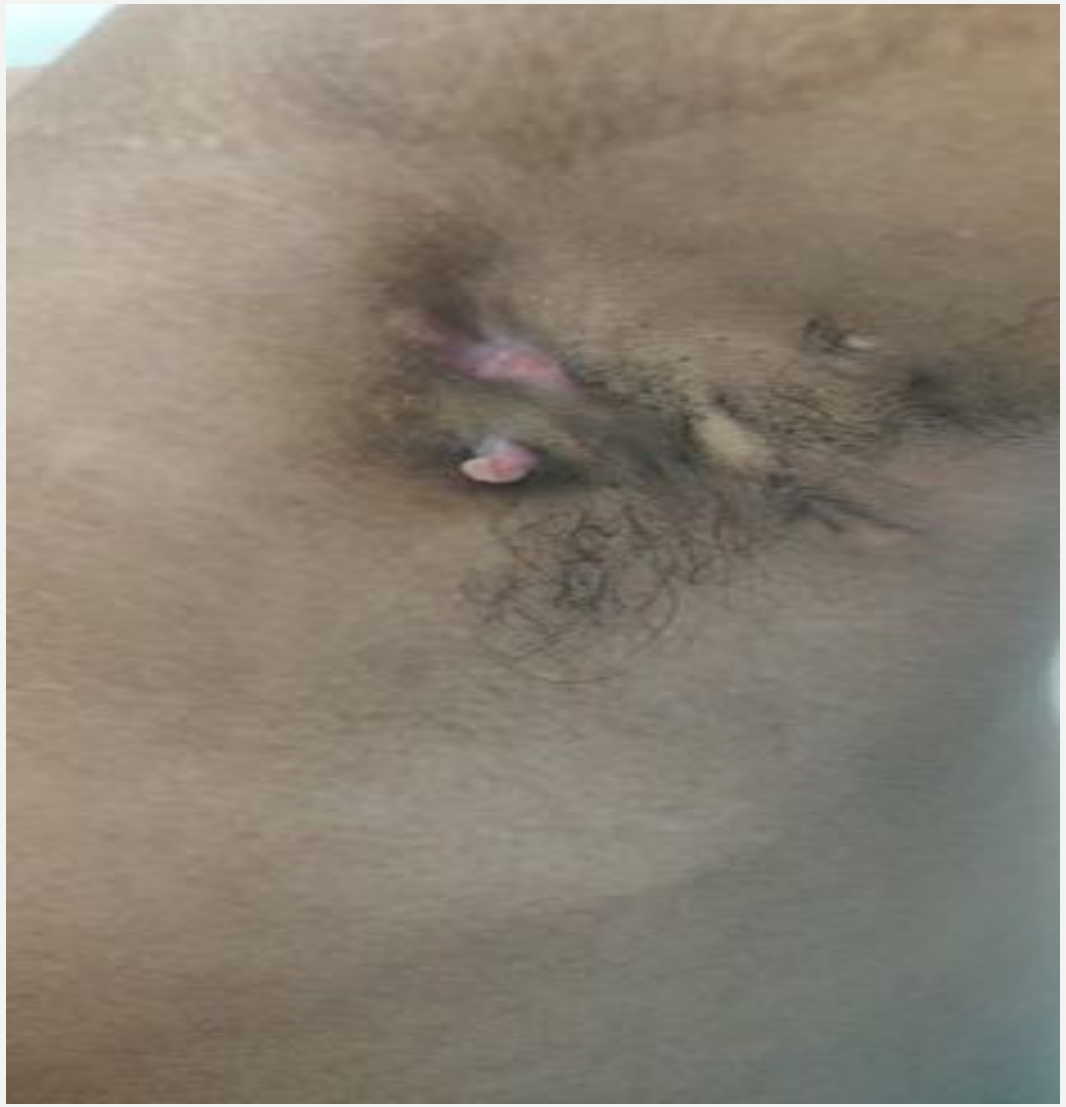












DIFFERENTIAL DIAGNOSIS

- Hidradenitis suppurativa
- Recurrent furunculosis
- Scrofuloderma
- ??

RELEVANT INVESTIGATIONS

- CBC, LFT, KFT, lipid profile, FBS, routine stool & urinalysis – normal
- Mantoux test – negative
- Pus C/S – Staph. Aureus
- Biopsy s/o Hidradenitis Suppurativa

THERAPEUTIC APPROACH

- Age dependent?
- Severity dependent?
- Financial constraint?
- Resource poor setting?
- Affection on QoL?
- Personal experience?

TREATMENT SEQUENCE FOLLOWED OVER PAST 2 YEARS

1) Doxycycline 100mg BD + topical clindamycin (1 month)
   - 1st line Rx
   - Mostly inflamed lesions initially

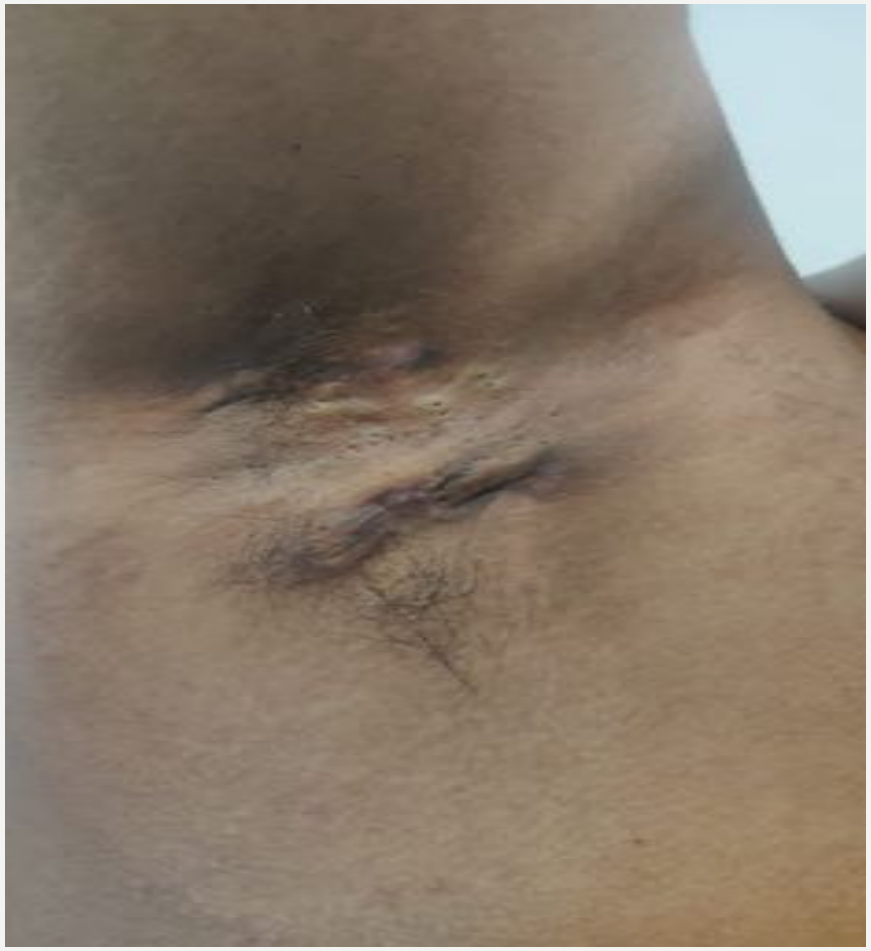
2) Isotretinoin 20mg / 30mg (2 months)
   - Decreased inflammation after doxycycline therapy
   - Multiple comedones
   - Teenager

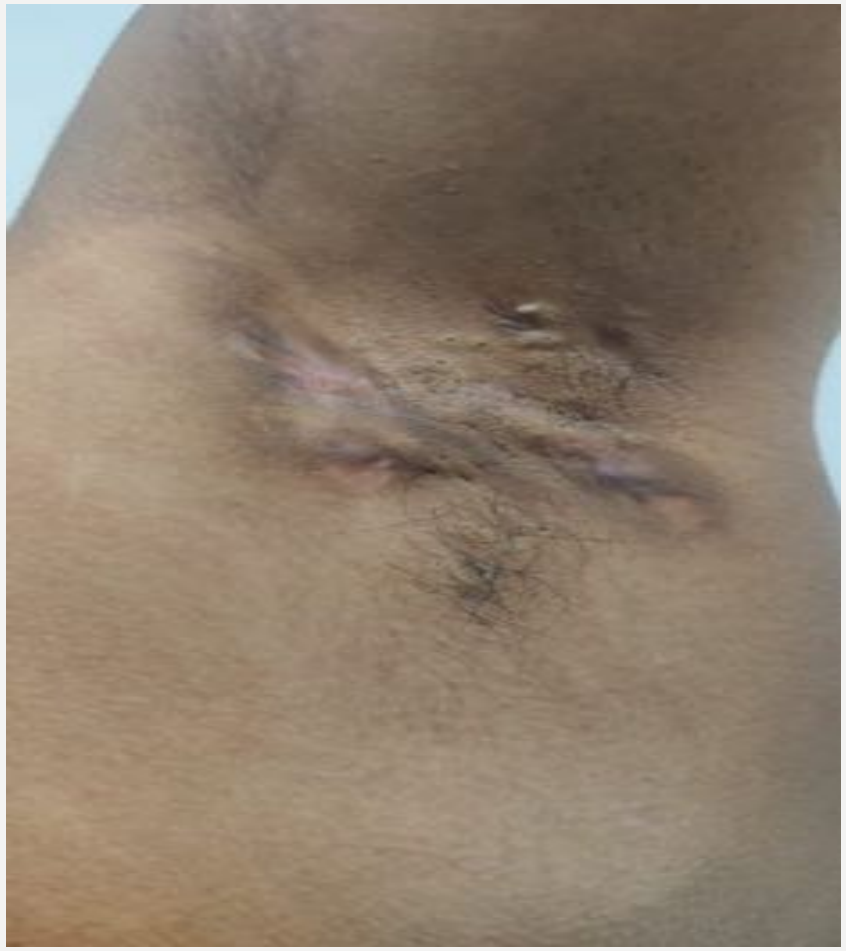
3) Doxycycline 100mg BD / Minocycline 65mg OD (2 months)
   - Recurrent painful nodules

4) Adalimumab 160mg / 80mg / 40mg (Initiation phase)
   - Recurrent painful nodules not responding to tetracyclines

5) Adalimumab 40mg weekly (3 months) (Maintenance phase)

REMISSION OF LESIONS AFTER INITIAL ADALIMUMAB THERAPY





6) Adalimumab 40mg every 2 weeks

- Due to financial constraint
- Complete remission for 3 months

7) Adalimumab + Doxycycline 100mg thrice weekly

- 1 – 2 new painful nodules/week

8) Adalimumab + Apremilast 30mg BD

- Additional therapy tried to decrease dose/frequency of adalimumab

9) Adalimumab + Acitretin 25mg OD

- 1 – 2 new painful nodules/week
- Non response to additional doxycycline & apremilast

10) Intralesional steroid on requirement basis given during whole treatment period

- ILS Triamcinolone given on & off for few acute painful lesions

LABORATORY PARAMETERS FOLLOWED

- Normal Hemogram, renal, hepatic parameters & lipid profile – **initial 1 year**

- Mild Transaminitis (SGOT – 43U/L, SGPT – 82U/L) – **14 months post Rx**

15 months post Rx -

- Leukopenia (TLC – 5,800/cmm)
- DC - Neutropenia & Lymphocytosis (N_{32} L_{61} E_{02} M_{05} B_{00})
- Transaminitis (SGOT – 98U/L, SGPT – 154U/L)
- Adalimumab & acitretin on hold
- Multiple new lesions developing

- **Further course of treatment???**

CASE 6

DR RAHUL MAHAJAN

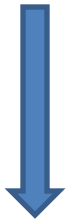
Multiple ulcers since early childhood

CASE PRESENTATION

- 15 years old boy from Gujarat
- Symptomatic since 12 years

History of present illness

- Developed first painful papulo-pustular lesion on right thigh → ulcer within 1 week → healed with scarring after 1½ year with multiple courses of antibiotics



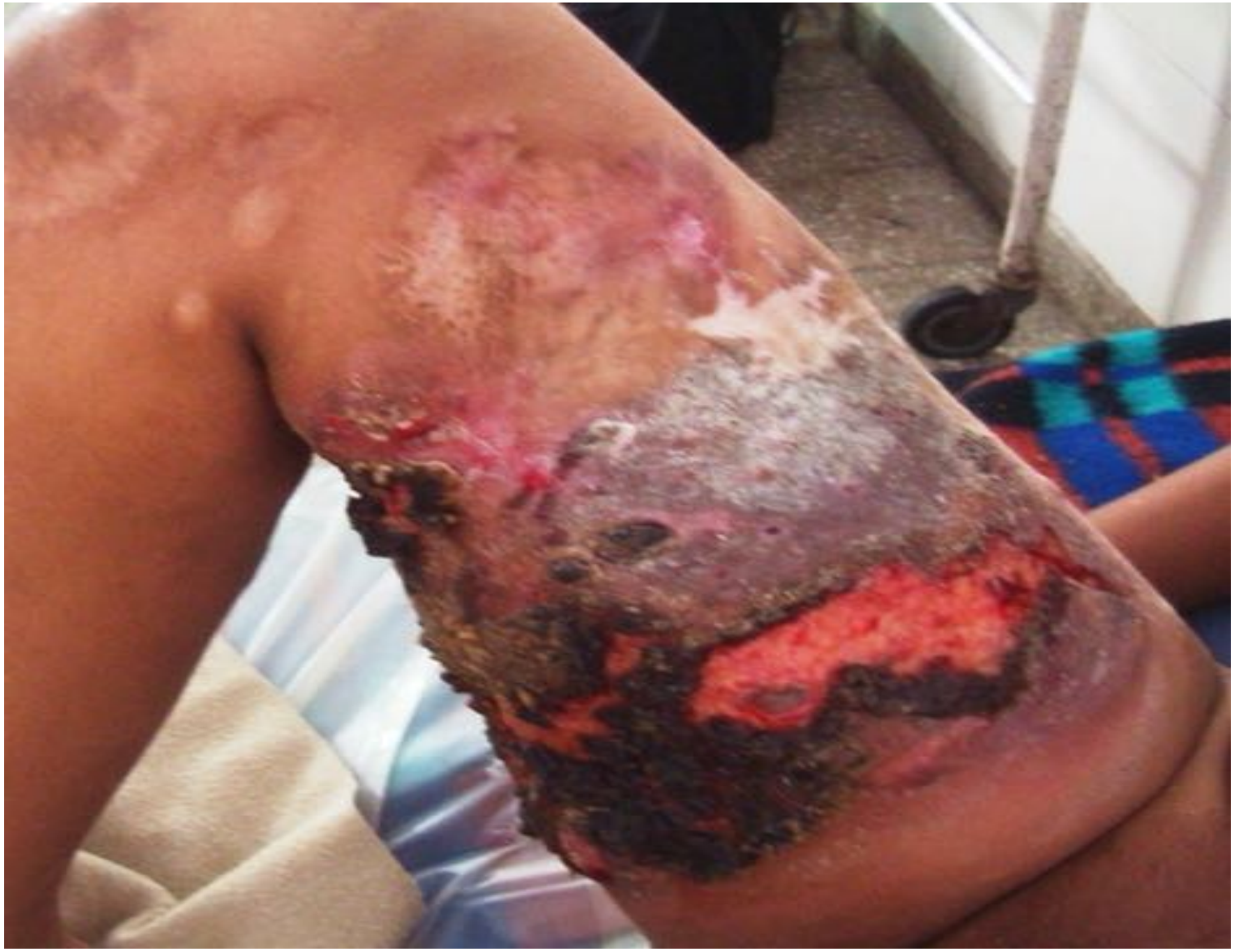
- Recurrent painful papulo-pustular lesions developed on both thighs and legs every 3-4 months

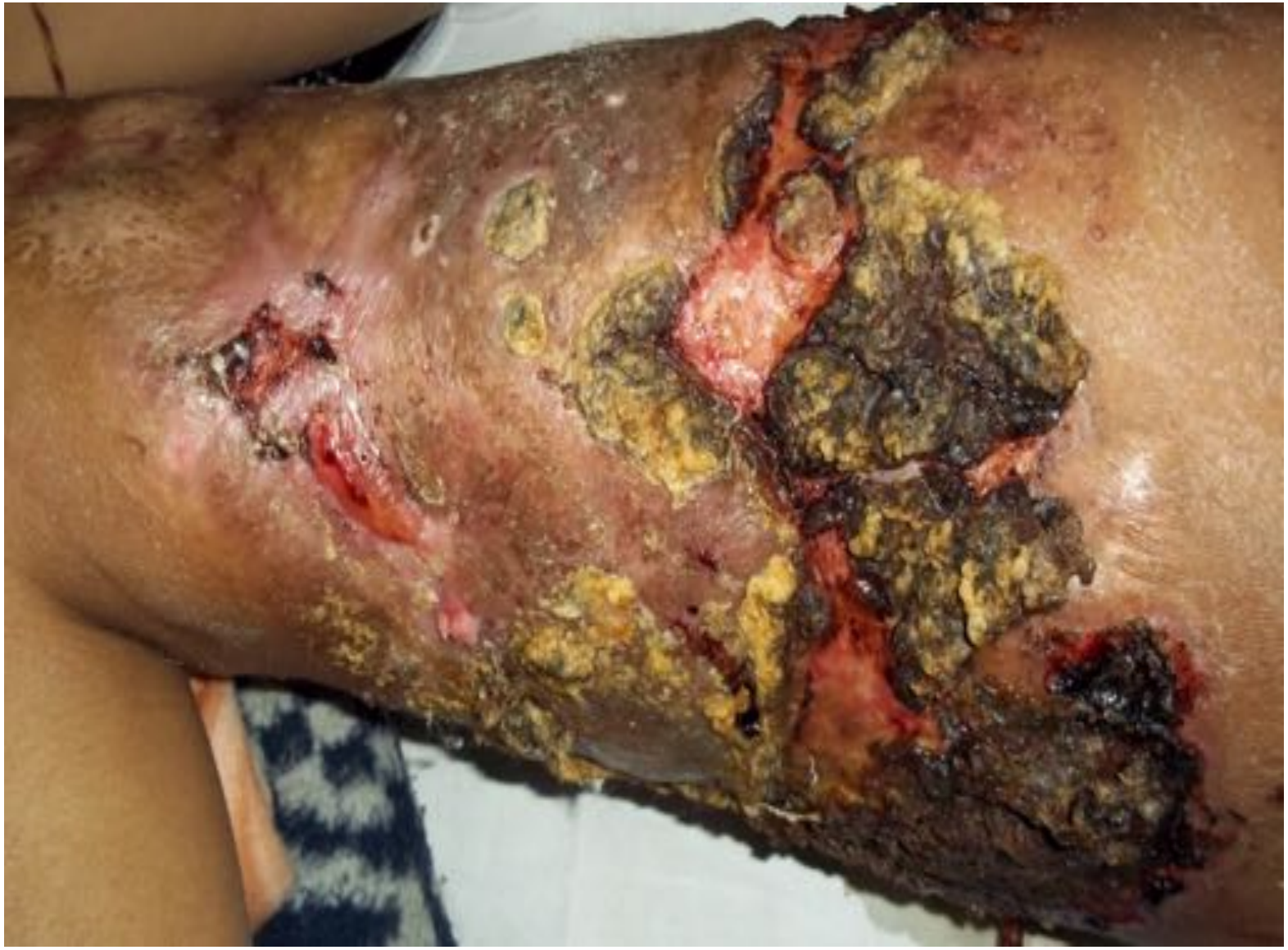
- About 3 years ago, minor trauma to knee → tender erythematous swelling → I&D done → non-healing ulcer

- Skin grafting done with donor graft taken from anterior area of the thigh

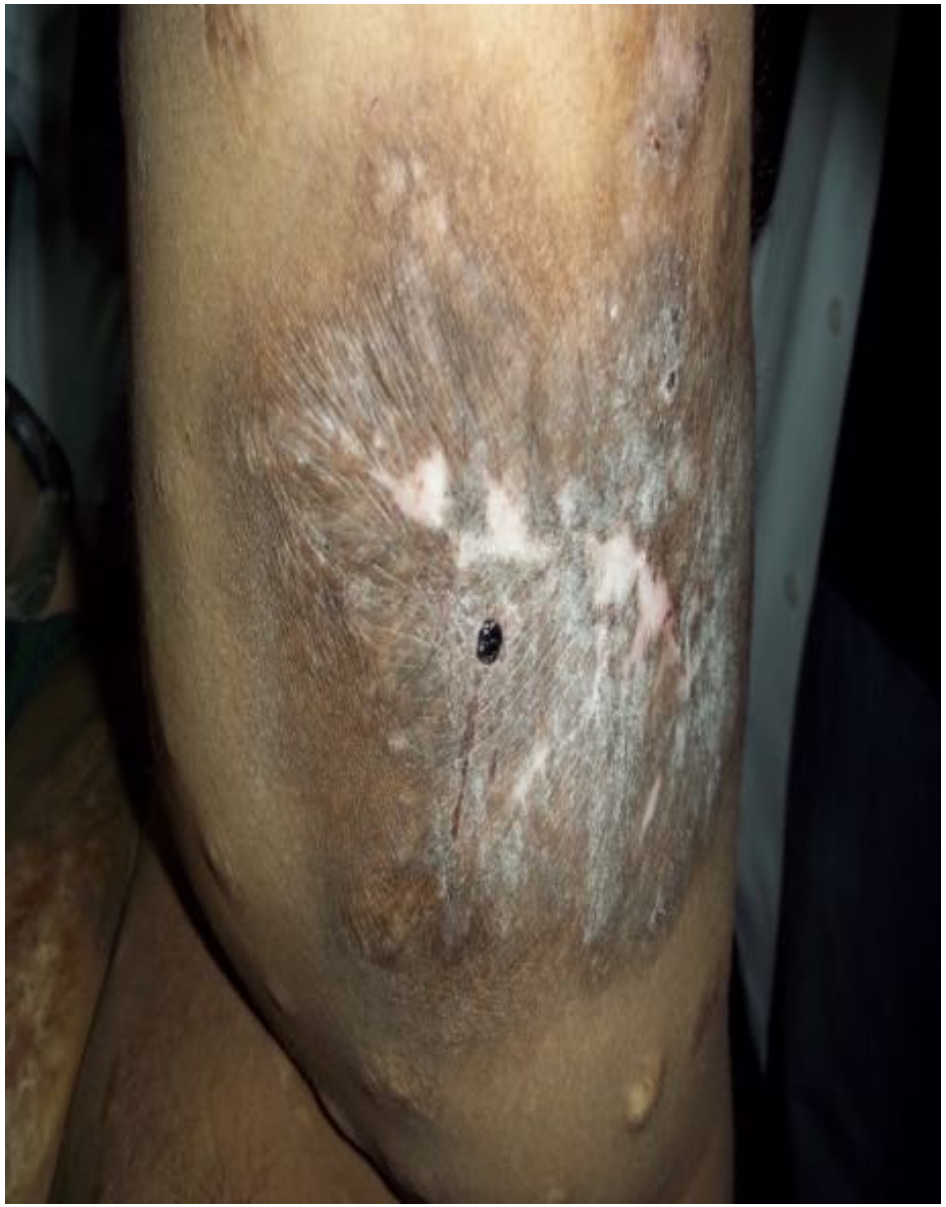
- Led to the non- healing ulcer at the donor site

Chief Complaints

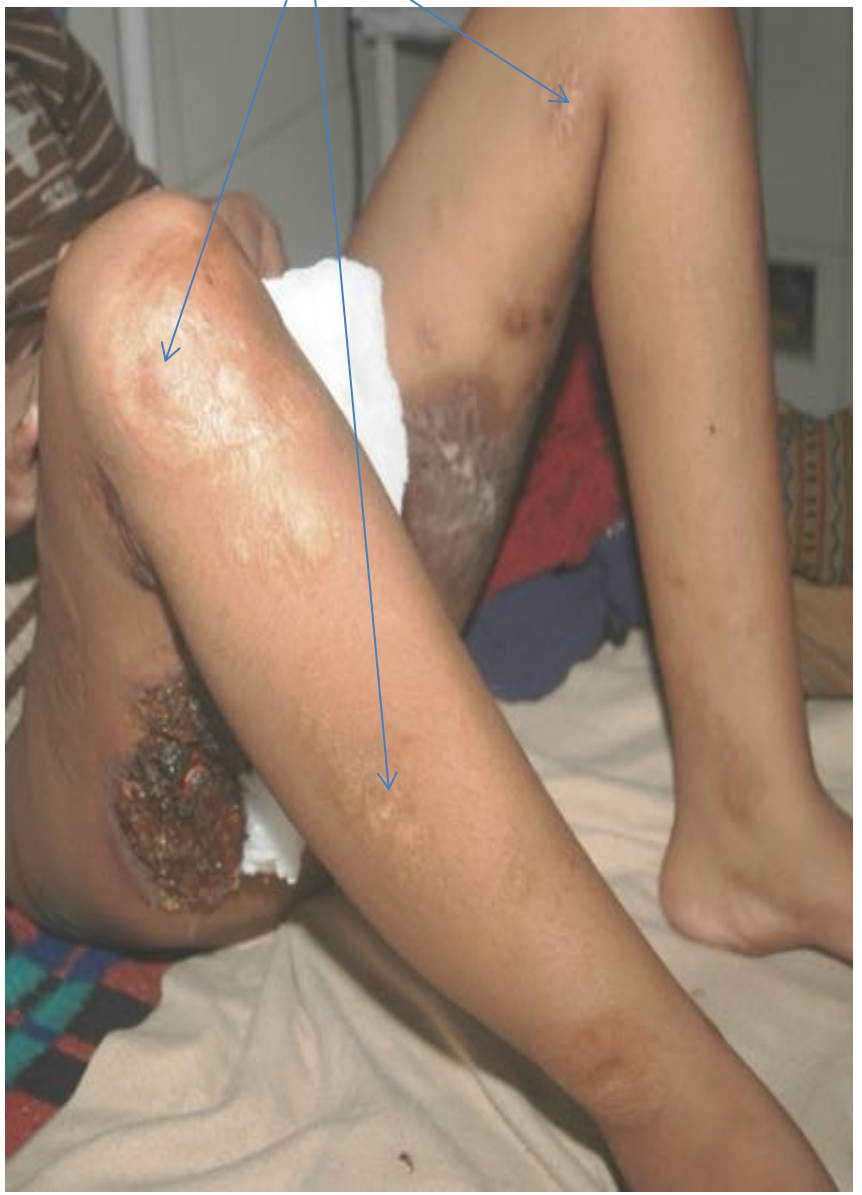




At places hypertrophic scars



Multiple cribriform scars



Past History:

- h/o recurrent episodes of chest infection
- No significant h/o GI complaints

Treatment History:

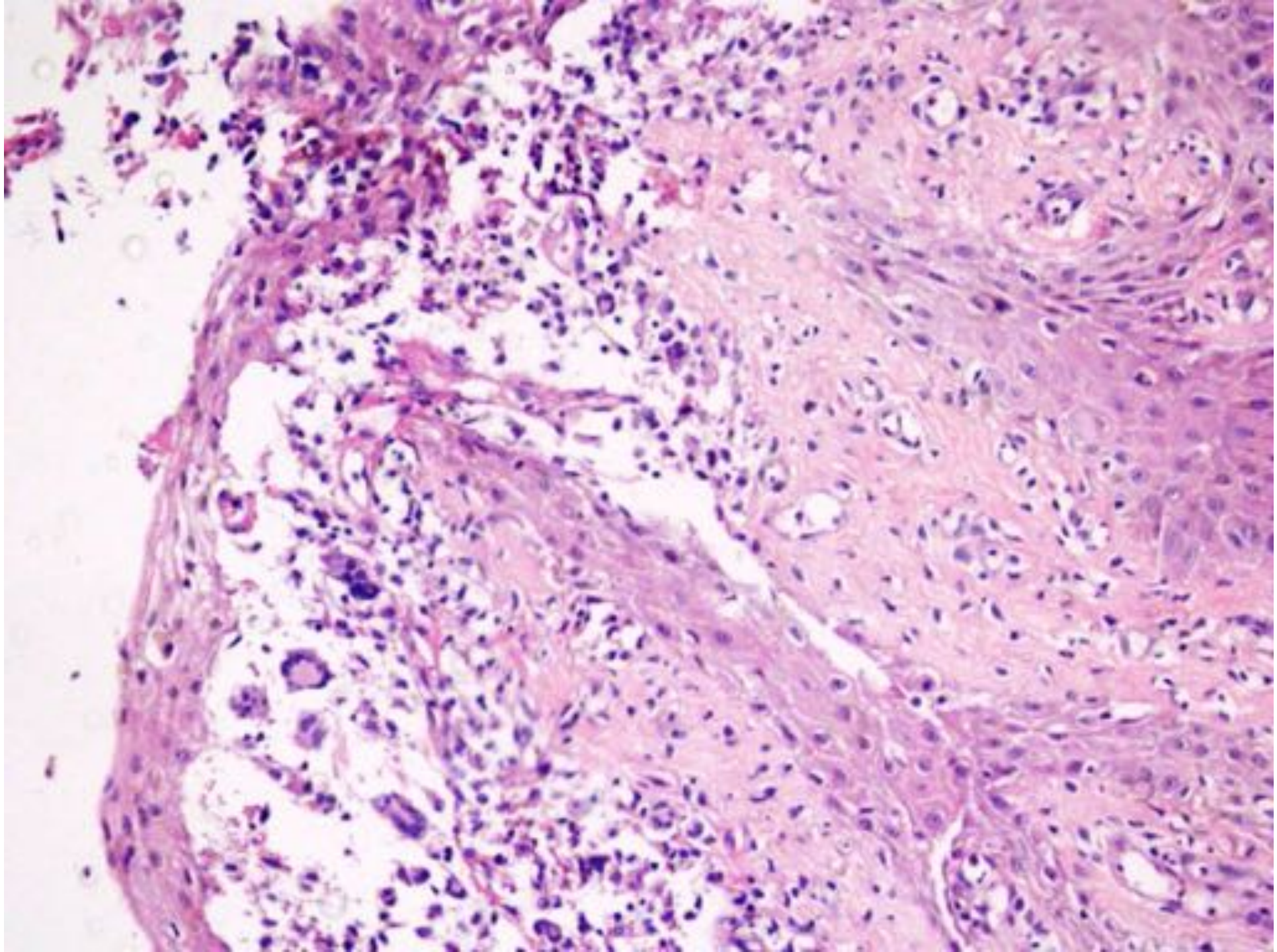
- I/v corticosteriods & antibiotics
- Multiple courses of traditional treatments

Family History: No consanguinity, No such family history

Investigations

	10/1/11	12/1/2011	17/1/2011	25/1/2011
Hb (g/dl)	**8.1**	**9.2**	**8.9**	**8.9**
TLC/UL	**30,000**	**55,000**	**49,600**	**41,080**
DLC (N/L/M/E)	--	**92**/4/3/1	**98**/2	**92**/6/1/1
Platelets/UL	--	**654×10^3**	**591×10^3**	**790×10^3**
Na^+/ K^+/ Cl^-	146/4.8/104	132/5.48/98	----	153/4.81/109
Urea/Crt (mg/dl)	22/0.2	24/0.56	----	33/0.49
LFT's	NORMAL	----	----	----

- **Wound swab culture:** Staph aureus
- **ANA/ ANCA:** Negative
- **Tuberculin test:** negative
- **Chest X-ray:** Normal
- **USG:** mild hepatomegaly
- **Nitroblue tetrazolium reduction test:** Normal
- Immunoglobulin levels - Normal



Clinical diagnosis

- **Pyoderma Gangrenosum**
  - **Not very inflammatory margin**
  - **Too young an age**
  - **Relatively less neutrophils on histopathology**

- **Some other dermatoses masquerading as PG**
  - **? primary immunodeficiency disease**
  - **? Wegeners granulomatosis**

Planned for further immunodeficiency work-up

Treatment

- Prednisolone 80 mg OD
- Antibiotics - Amoxy-clavulunic acid 625 mg TID and Ciproflaxacin 500 mg BD
- Aceclofenac and paracetamol BD
- Calcium supplements BD
- Daily dressing

- Ulcer decreased in size and patient was discharged on

➢ Tab. Prednisolone 60 mg OD

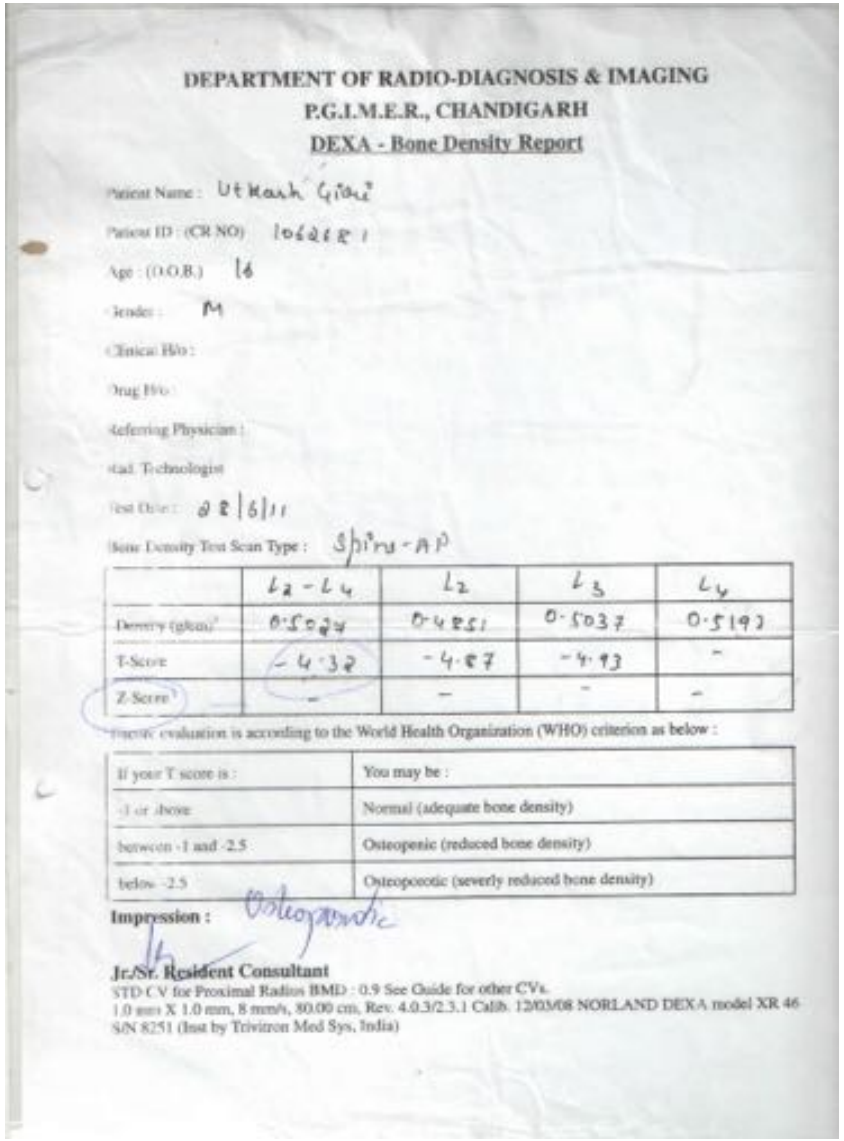
➢ Tab. Co-trimoxazole OD prophylaxis

Follow-up

- Attempt to taper and stop corticosteroid led to appearance of new pustules on the thighs

- Started on **Azathioprine 50 mg OD** along with **co-trimoxazole prophylaxis**

- Vitamin D3 60,000 IU once weekly



DEPARTMENT OF RADIO-DIAGNOSIS & IMAGING
P.G.I.M.E.R., CHANDIGARH
DEXA - Bone Density Report

Patient Name : Utkash Girdi
Patient ID : (CR NO) 106218
Age : (D.O.B.) 14
Gender : M
Clinical Hx :
Drug Hx :
Referring Physician :
Cal. Technologist
Test Date : 22/6/11
Bone Density Test Scan Type : Spine-AP

	L2 - L4	L2	L3	L4
Density (g/cm²)	0.5024	0.4851	0.5037	0.5193
T-Score	-4.32	-4.87	-4.93	-
Z-Score	-	-	-	-

Diagnostic evaluation is according to the World Health Organization (WHO) criterion as below :

If your T score is :	You may be :
-1 or Above	Normal (adequate bone density)
between -1 and -2.5	Osteopenic (reduced bone density)
below -2.5	Osteoporotic (severly reduced bone density)

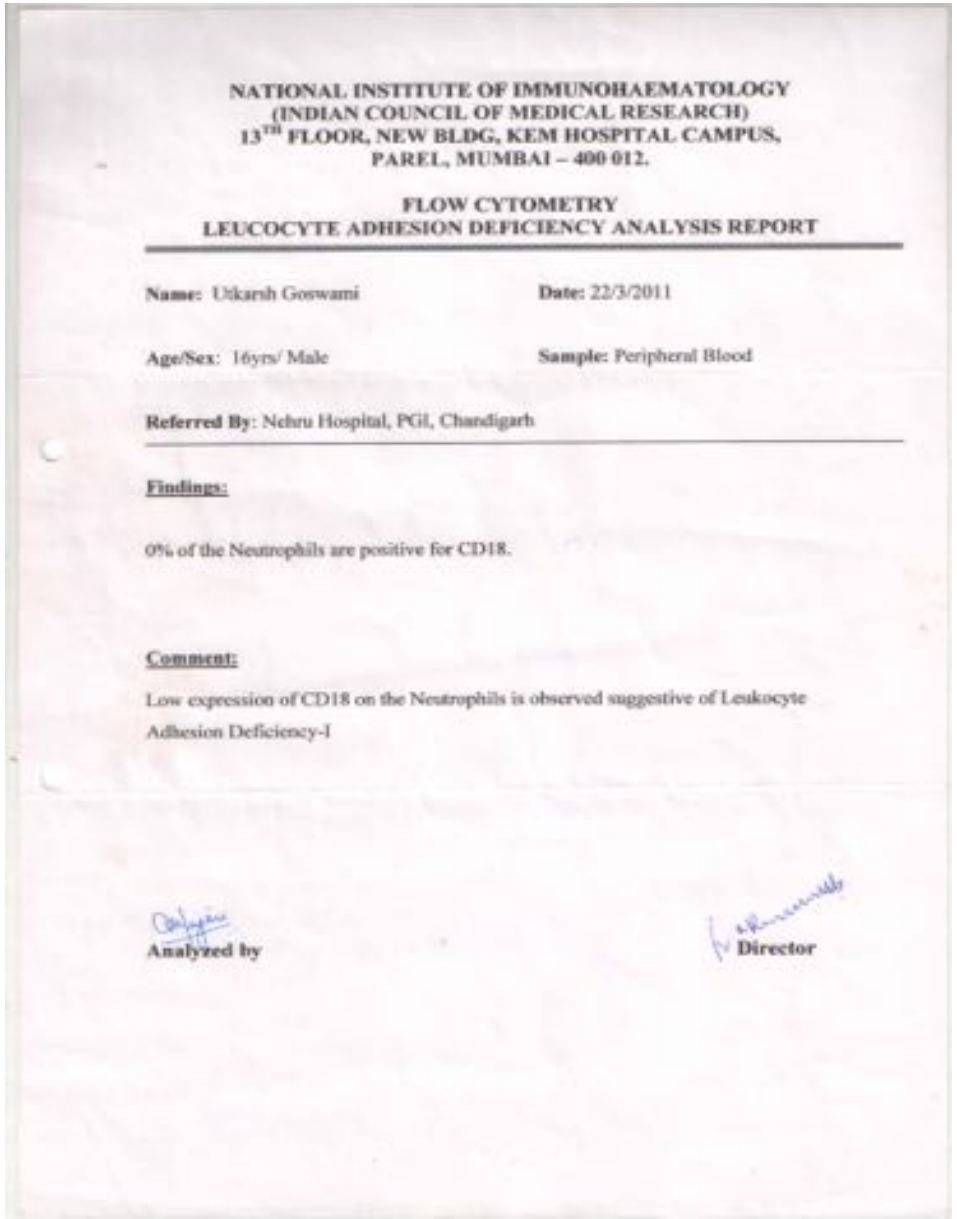
Impression : Osteoporotic

Jr./Sr. Resident Consultant

STD CV for Proximal Radius BMD : 0.9 See Guide for other CVs.
1.0 mm X 1.0 mm, 8 mm/s, 80.00 cm, Rev. 4.0.3/2.3.1 Calib. 12/03/08 NORLAND DEXA model XR 46
S/N 8251 (Inst by Trivitron Med Sys, India)

Flow Cytometry report

- **0% of neutrophils positive for CD18**
- **Suggestive of LAD-I**



NATIONAL INSTITUTE OF IMMUNOHAEMATOLOGY
(INDIAN COUNCIL OF MEDICAL RESEARCH)
13TH FLOOR, NEW BLDG, KEM HOSPITAL CAMPUS,
PAREL, MUMBAI – 400 012.

FLOW CYTOMETRY
LEUCOCYTE ADHESION DEFICIENCY ANALYSIS REPORT

Name: Utkarsh Goswami **Date:** 22/3/2011

Age/Sex: 16yrs/ Male **Sample:** Peripheral Blood

Referred By: Nehru Hospital, PGI, Chandigarh

Findings:

0% of the Neutrophils are positive for CD18.

Comment:

Low expression of CD18 on the Neutrophils is observed suggestive of Leukocyte Adhesion Deficiency-I

Analyzed by **Director**

Courtsey: National Institute of Immunohaematology

KEM hospital, Mumbai.

Final diagnosis

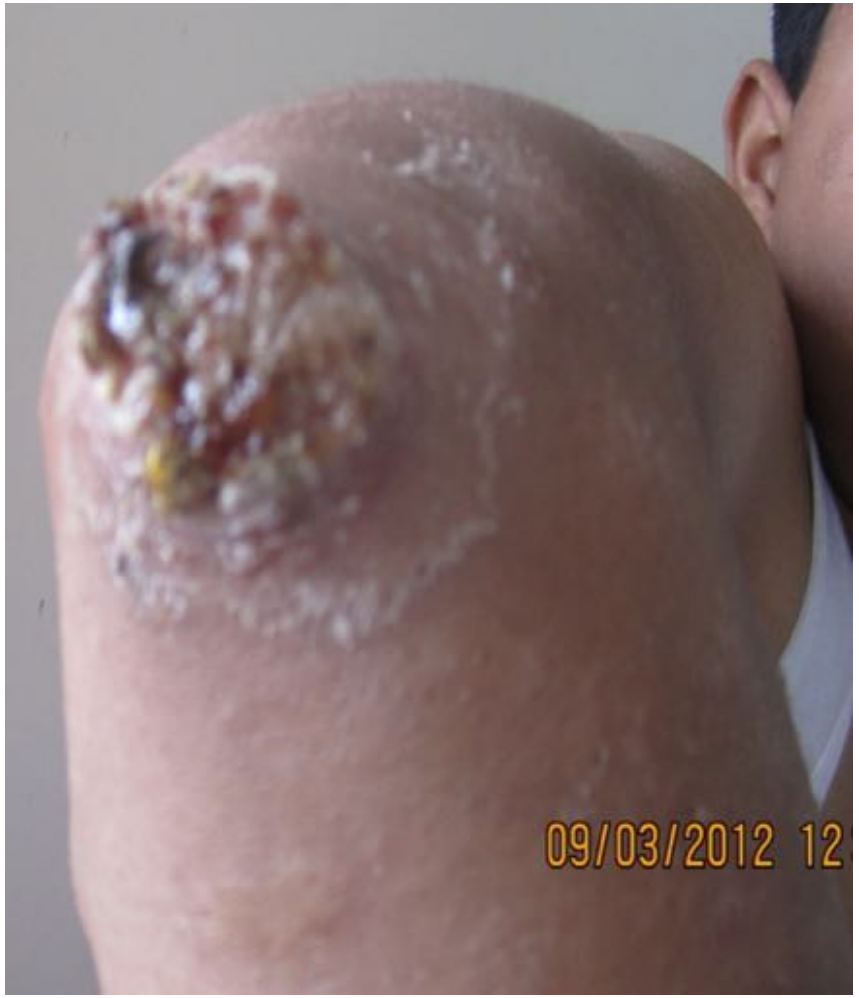
Leukocyte adhesion defect type 1

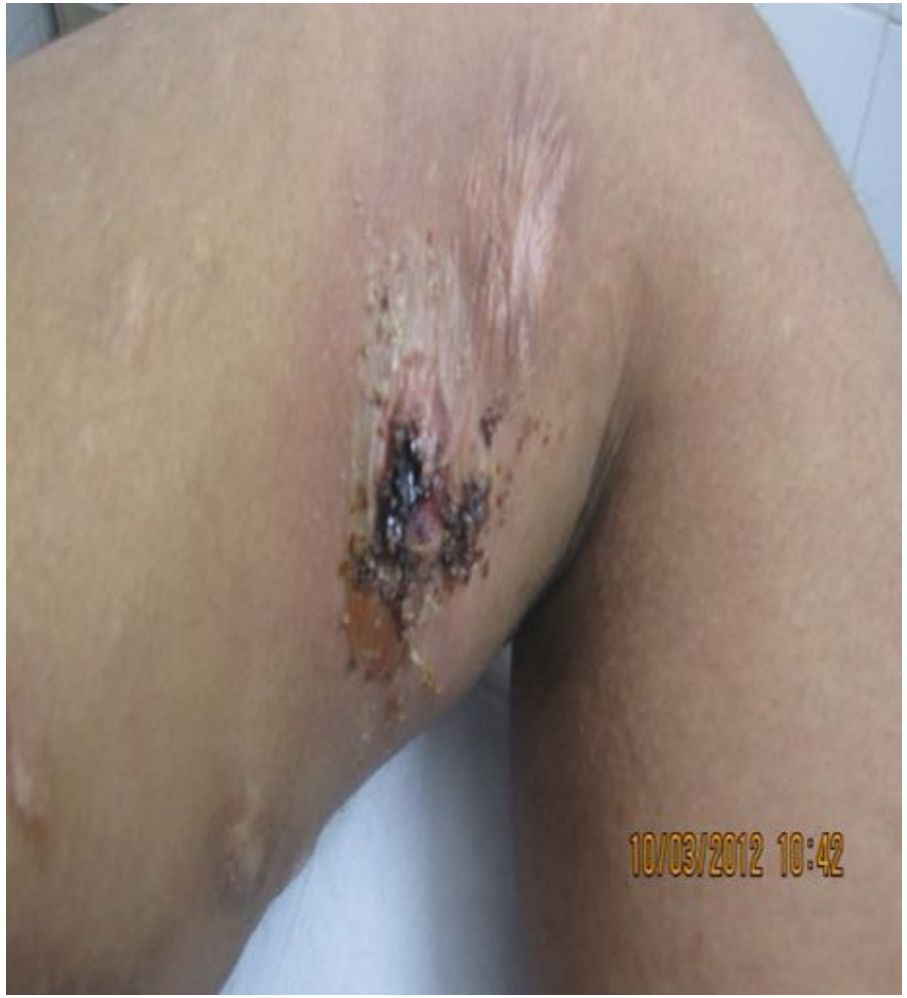
Another hurdle - Genetic Mutation Study

- **Study of gene sequencing of Exons segments , did not show any mutation**

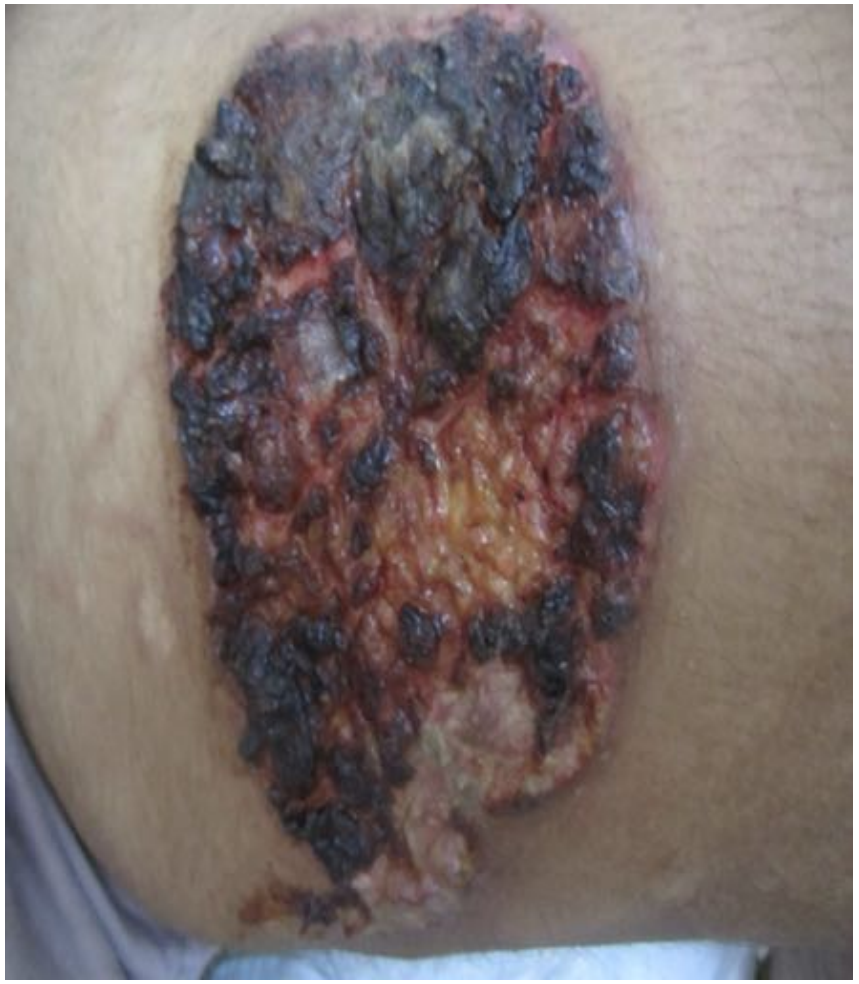
Courtsey: Dr. Amos Etzioni

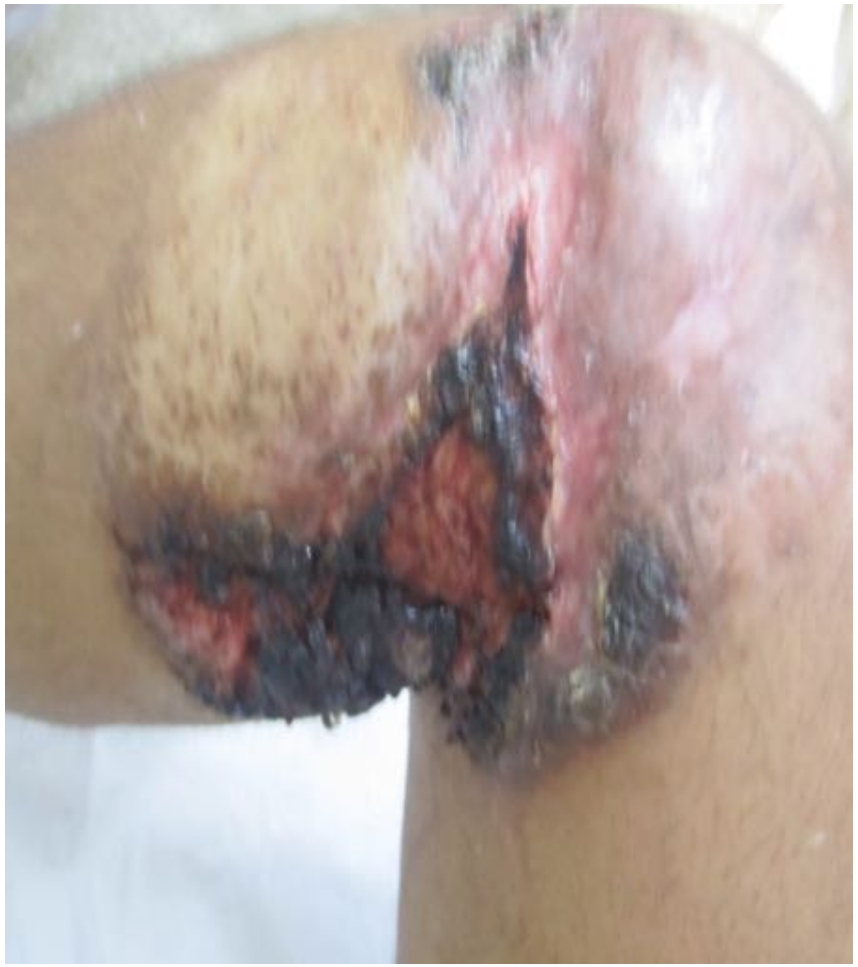
After one year





Still no end to misery





Management issues?

- Diagnosis in absence of genetic mutation?
- Role of immunosuppression ?
- Role of prophylactic antibiotics?
- Duration of antimicrobial therapy?

- The patient primarily had PG like lesions as the primary complaint (which were responding partially to immunosupression), and little of infections, he was kept on cotrimoxazole prophylaxis along with oral steroids/azathioprine.
-

- Finally, due to his continuing morbidity, his mother took a strong call opting for Haematopoietic Stem Cell Tranplantation with his sibling as the matched donor.
- After 40 days, 99.9% of cells were positive for CD 18.

CASE 7

DR RAHUL MAHAJAN

Vascular stain in a new-born

Case history

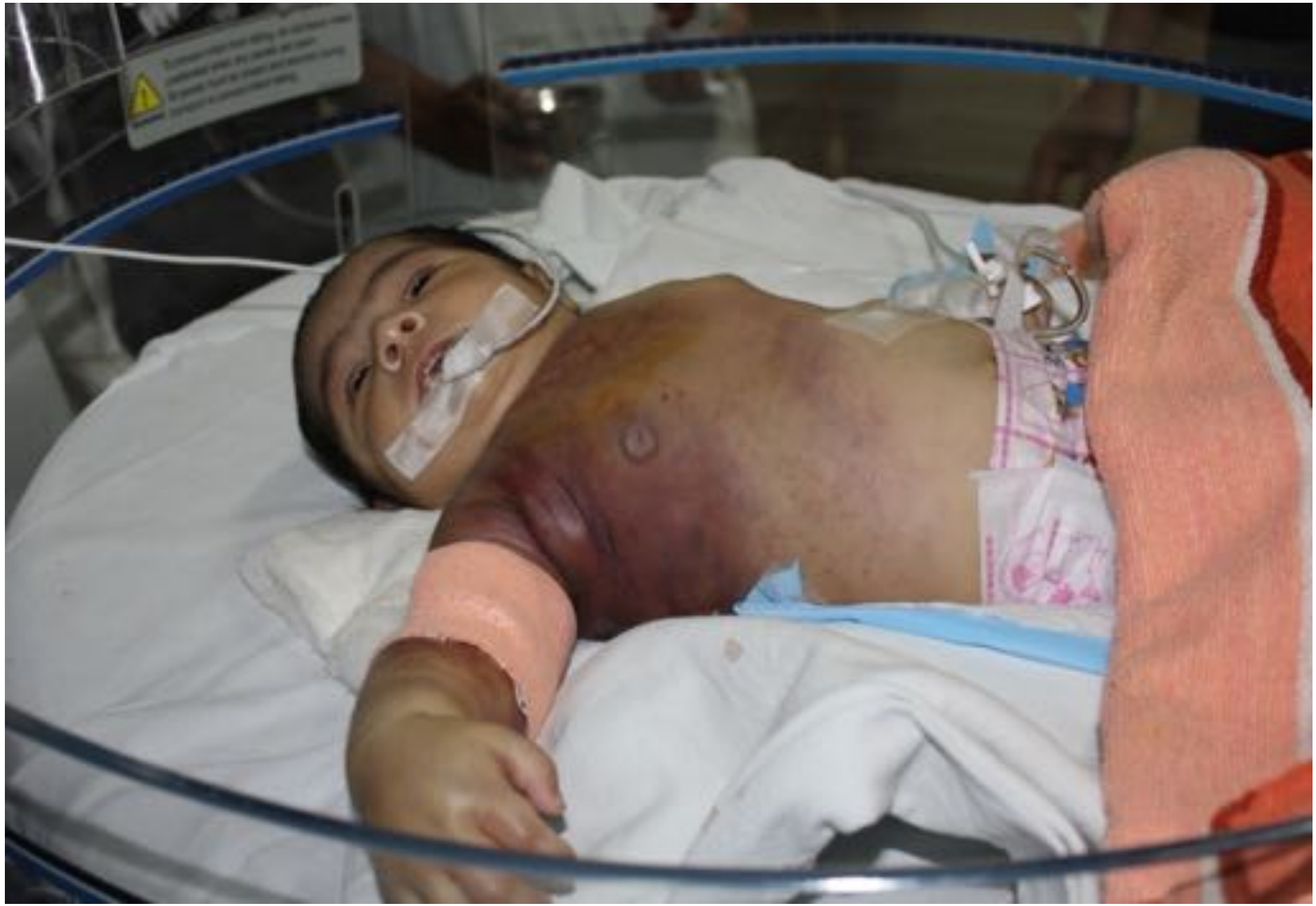
- Full term female baby referred to emergency department on day 25 of life with a 1.5 cms bluish lesion on the right side of the chest wall

- Day 4 of birth, fever (38.3°Celcius) with refusal to feed
- Treated for suspected sepsis with intravenous antibiotics for 5 days with improvement… discharged

- On day 15 of life with an increase in the size of the chest wall lesion associated with local signs of inflammation but no localized tenderness/bleeding

- Rest of the systemic examination was unremarkable.
- She was feeding well otherwise.

- This time, no response to intravenous antibiotics



Clinical possibilities

- Vascular tumour/malformation with probable Kassabach-Meritt phenomenon

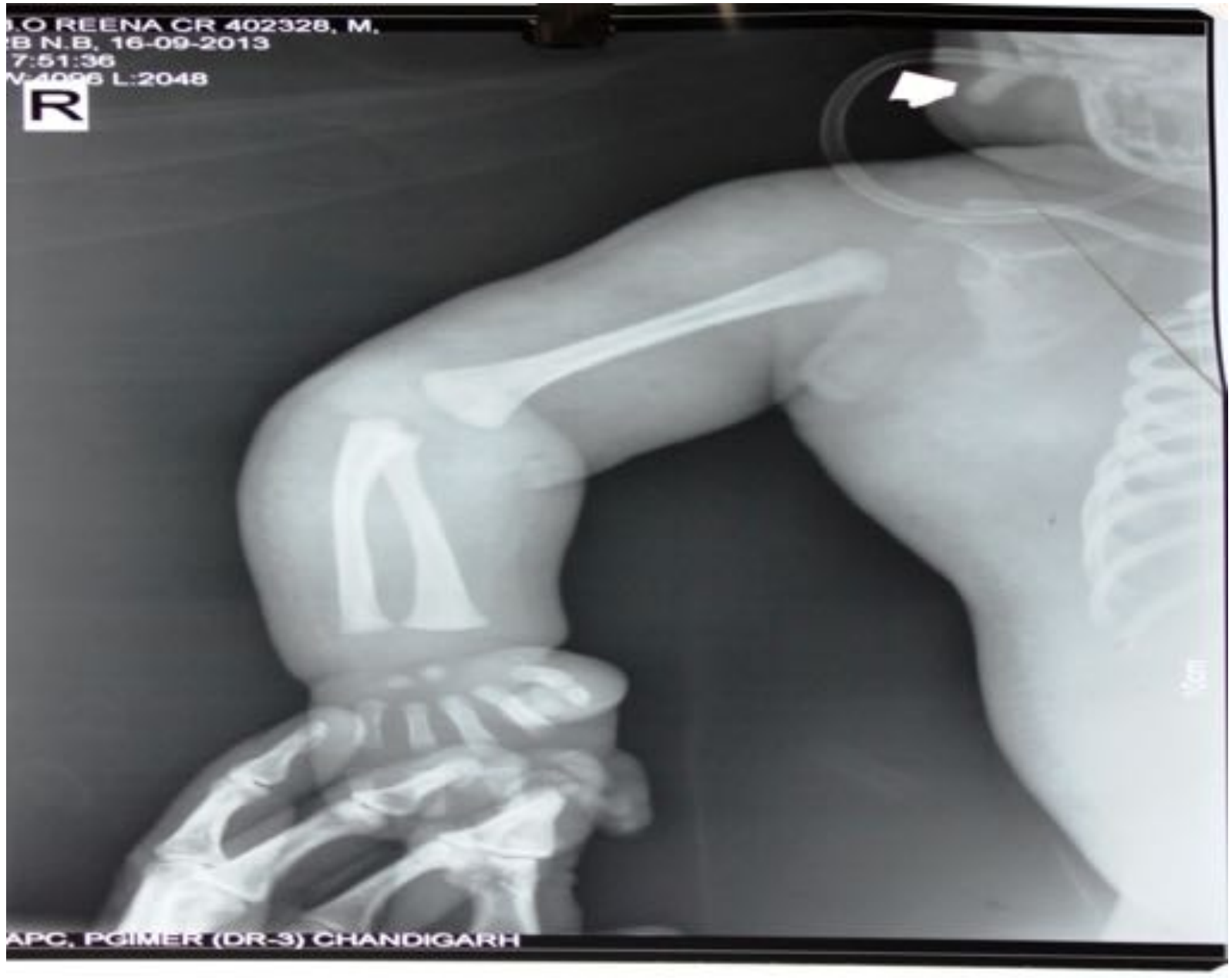
- ?soft tissue sarcoma

Investigations

- Low haemoglobin (5 g/dL), Leucocytosis (TLC - 17,000 per cmm), with neutrophilia (57%), and thrombocytopenia (6,000 per cmm)

- Coagulation profile
  - Prothrombin index (PTI) of 76% wit
  - marginally elevated partial thromboplastin time (aPTT) (42 seconds) and
  - hypofibrinogenemia (0.86 g/L).

- Doppler ultrasound study suggestive of
  - massive subcutaneous edema in the right upper limb, extending up to the chest wall, with an underlying pleural effusion and collapse of the ipsilateral lung, without any evidence of localized thrombosis.
  - Not sure about hemangioma or vascular malformation of any internal organ including liver.


O REENA CR 402328, M,
B N.B, 16-09-2013
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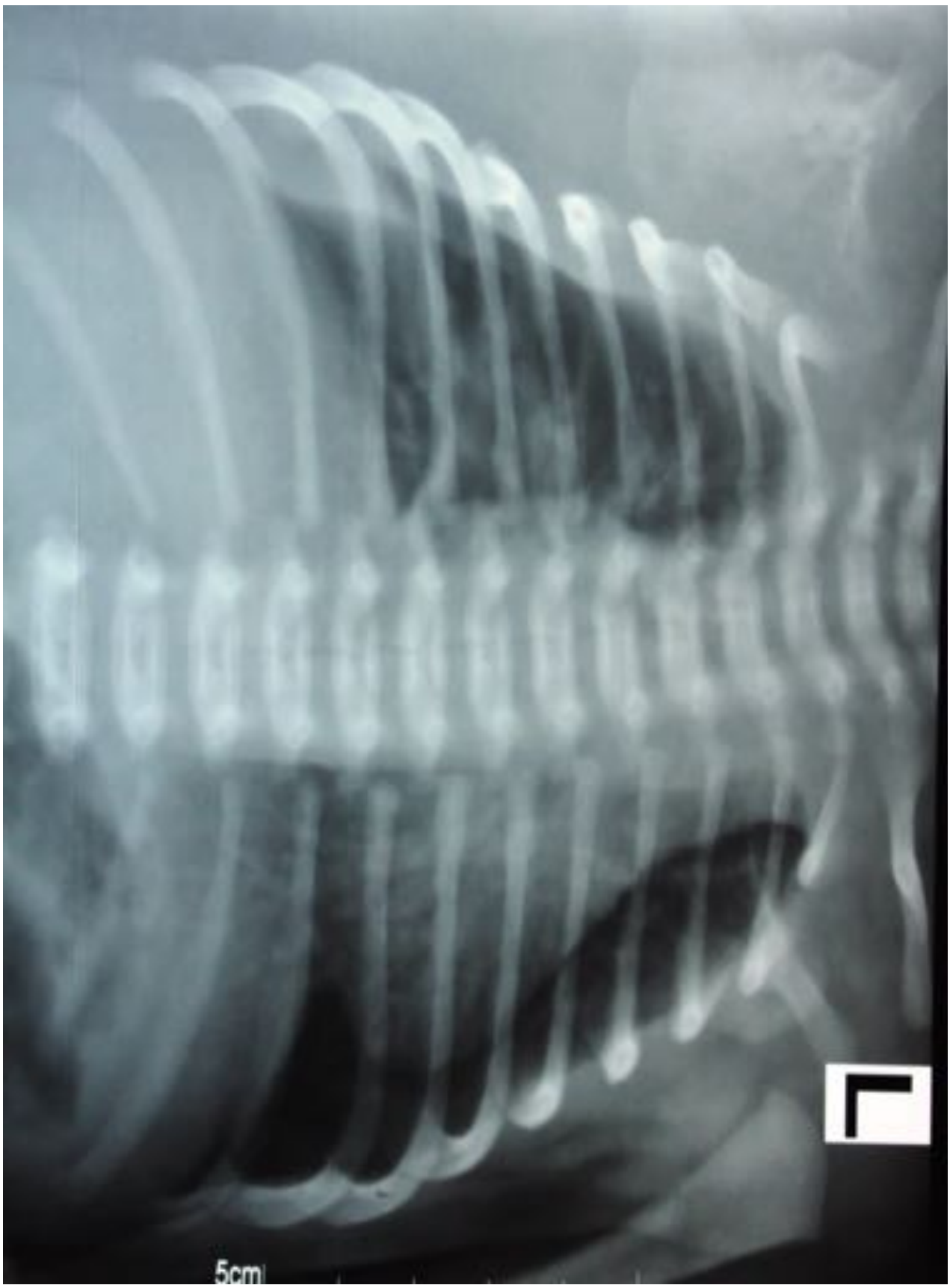
R
APC, PGIMER (DR-3) CHANDIGARH


5cm

Initial treatment

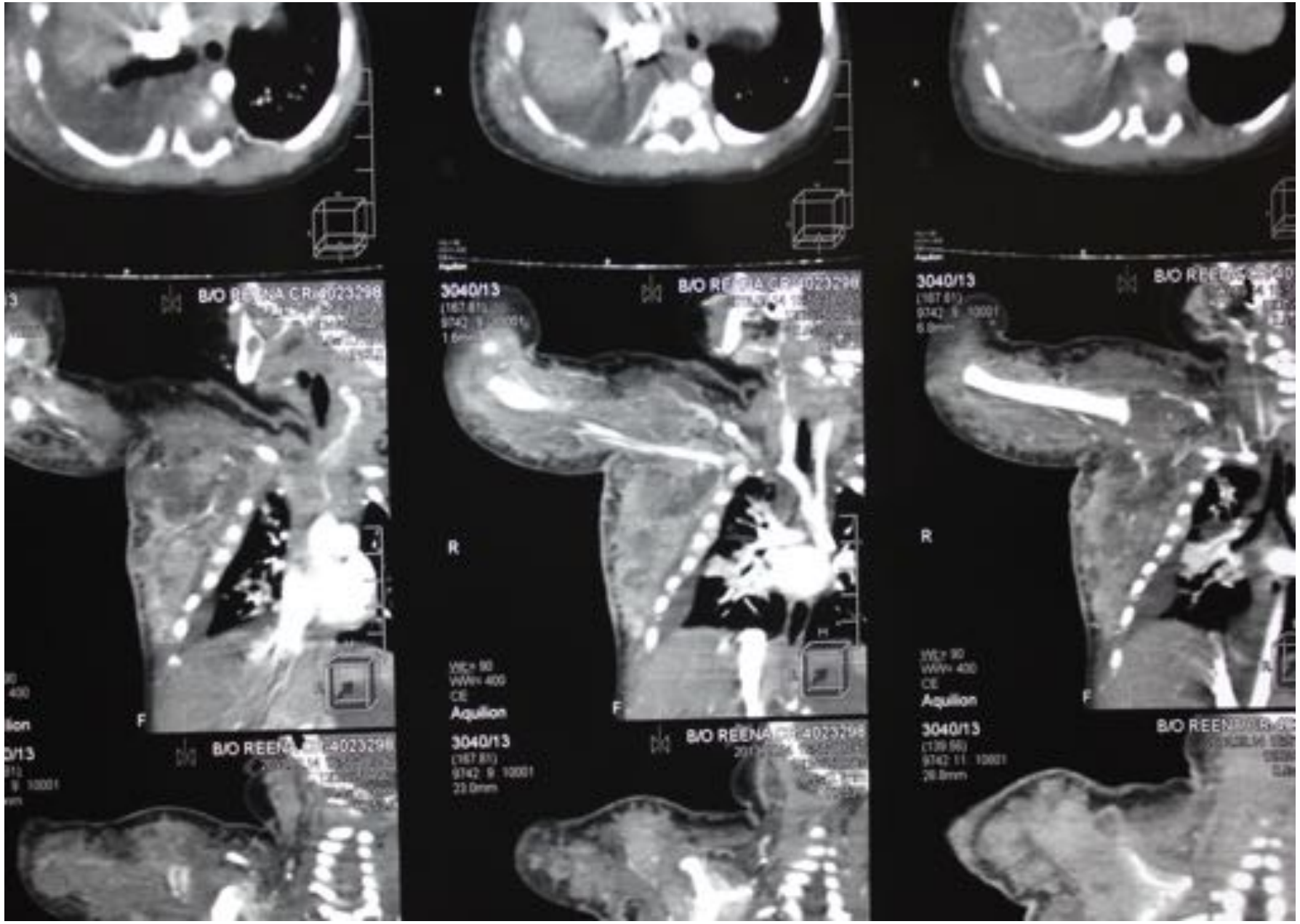
- Intravenous antibiotics (vancomycin and meropenem) on day 1 of admission.
- Breast feeds continued.
- Platelet concentrates (10 ml/kg) were also transfused.
- Day 2 of hospital stay, baby developed respiratory distress →CPAP were started.
- In view of deterioration in the clinical condition, intravenous antifungal therapy (Amphotericin B) was also initiated.

Further investigations

- However, distress continued to increase.
- Subsequent, C-reactive protein also increased to 27.6 mg/dL.
- Persistent thrombocytopenia
- Blood culture was sterile.

Treatment

- Oral prednisolone 1.5 mg/kg/day started
- Respiratory distress continued to worsen
- Arterial blood gas - hypoxemia & hypercarbia
- Baby also received Vincristine (0.05 mg/kg/week)
- Multiple blood products transfused throughout the course of hospital stay (5 times packed RBCs, 7 times platelet concentrate, 2 times each fresh frozen plasma, and cryoprecipitate).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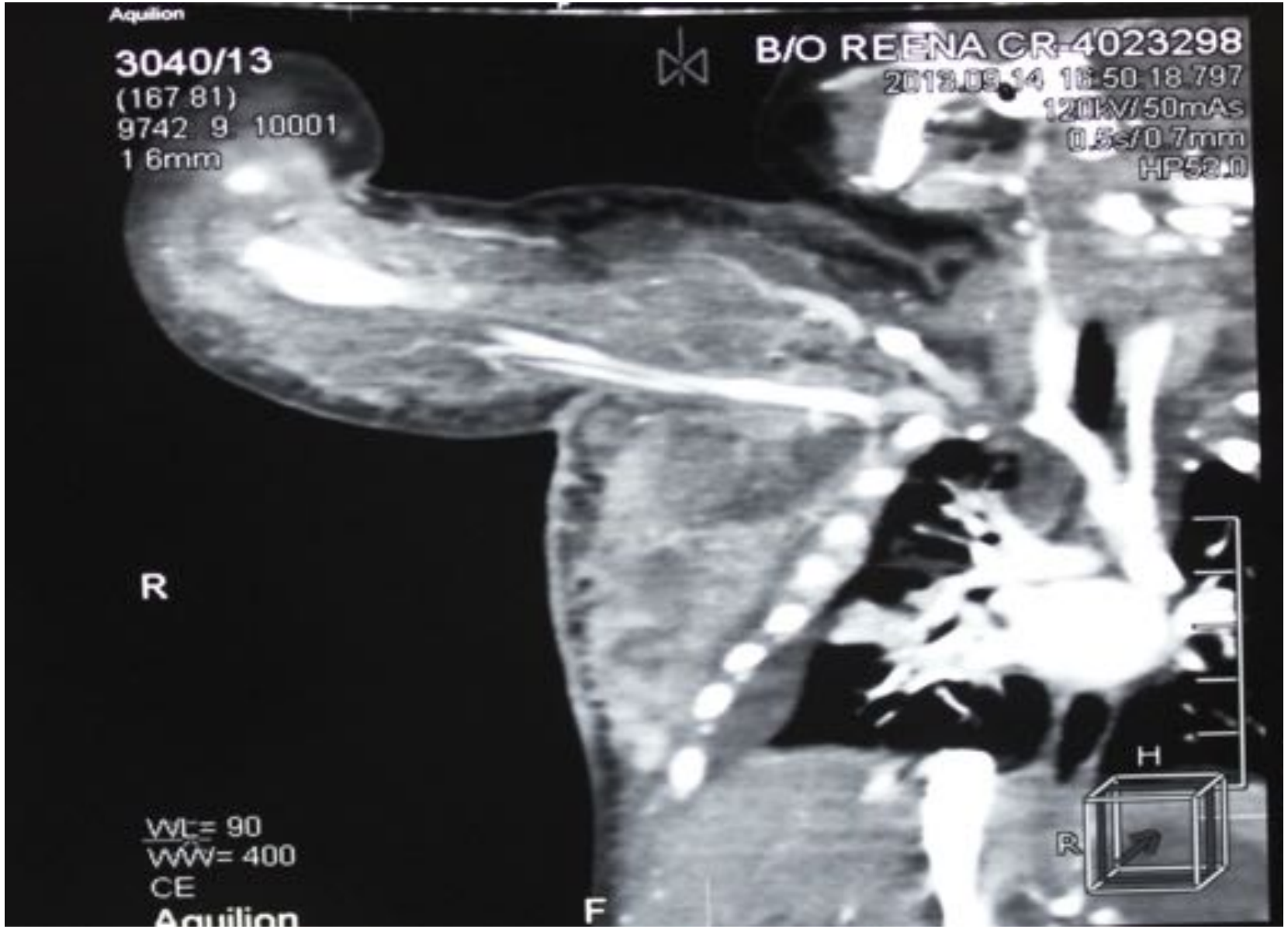
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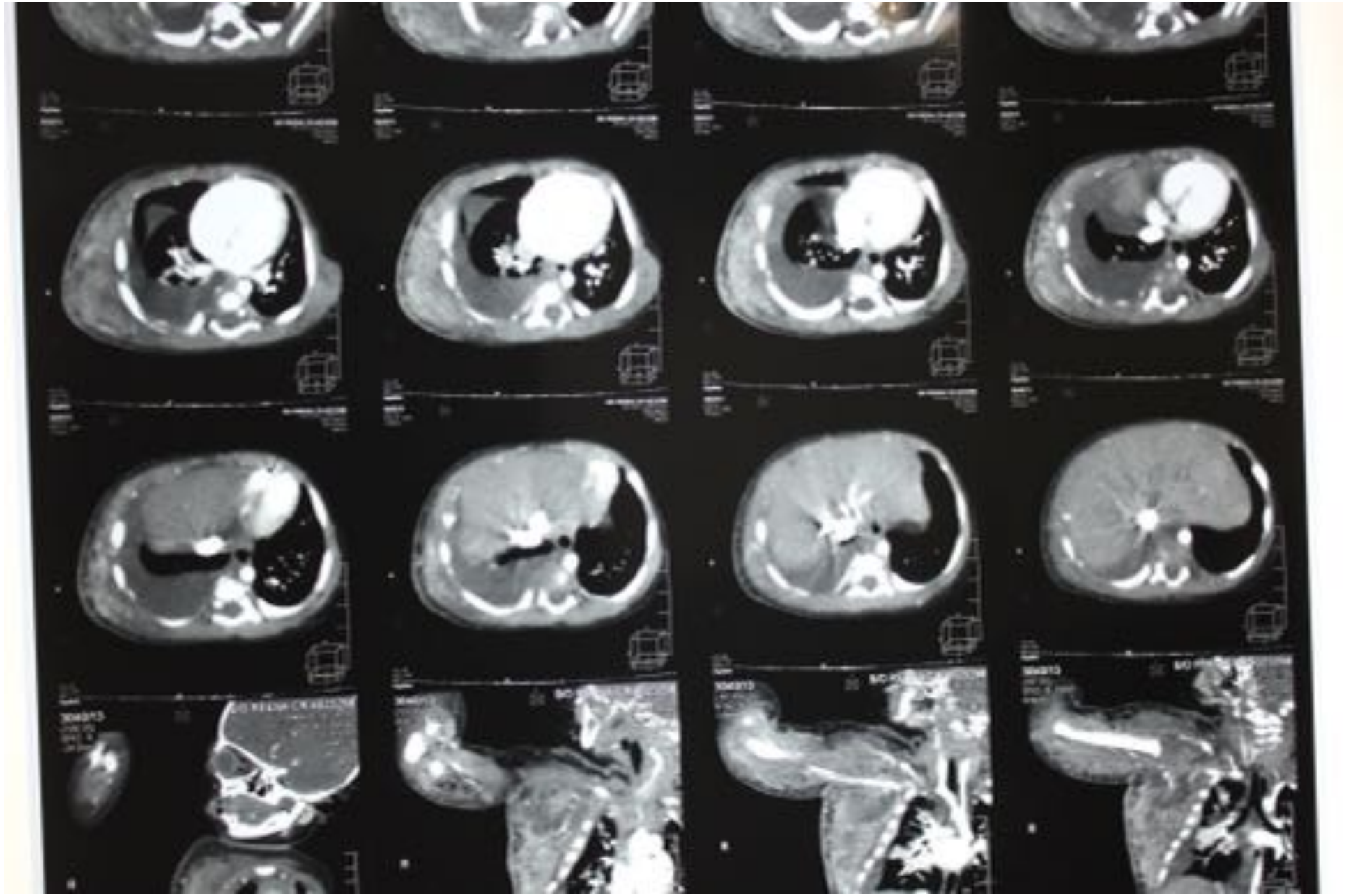

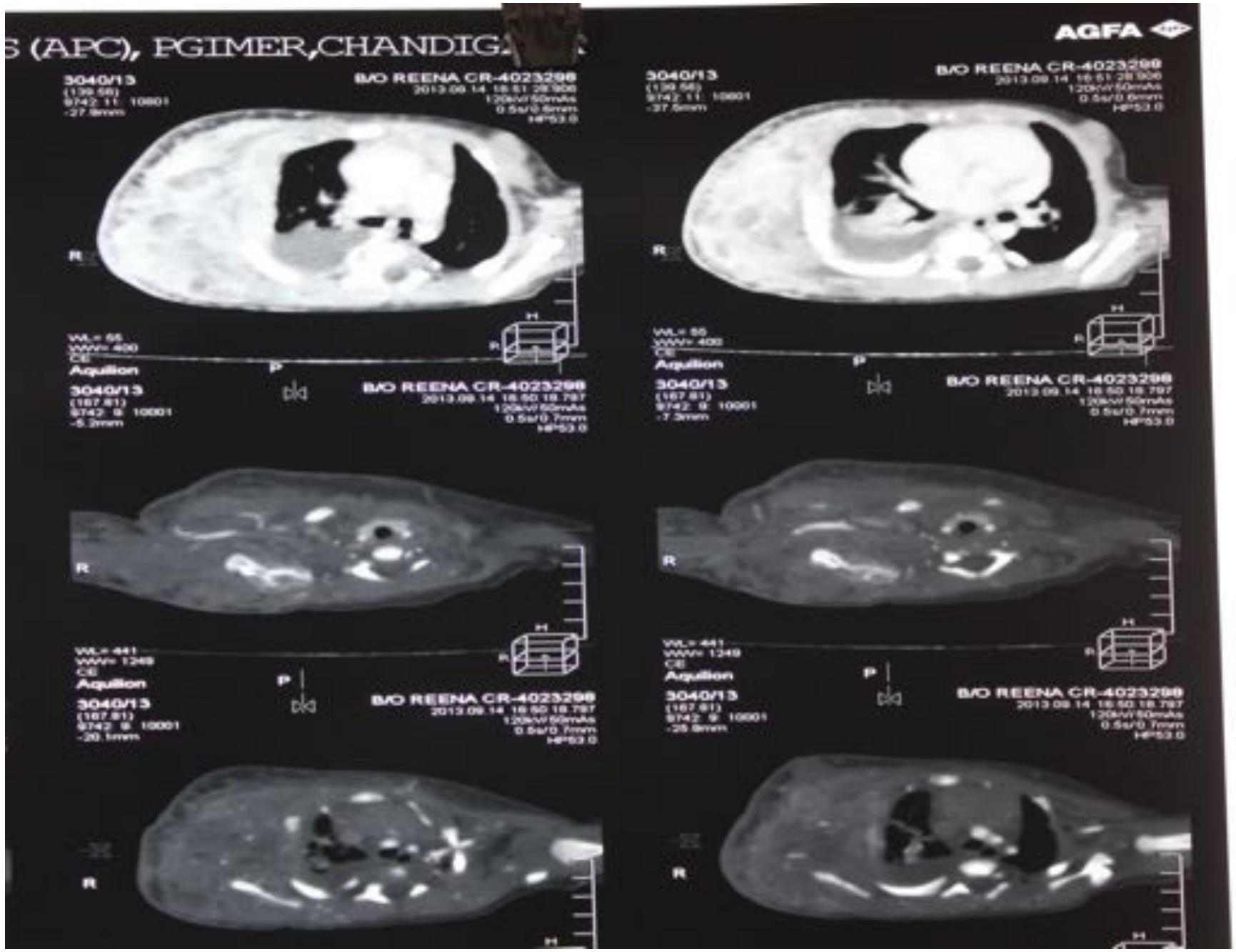
(APC), PGIMER,CHANDIG
AGFA
3040/13
B/O REENA CR-4023298
WL= 55
WW= 400
CE
Aquilion
3040/13
B/O REENA CR-4023298
WL= 441
WW= 1249
CE
Aquilion
3040/13
B/O REENA CR-4023298
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B/O REENA CR-4023298

No response

- Continued respiratory distress
- Mechanical ventilation with the P-SIMV mode with initial PIP of 18 mm Hg, PEEP 5 mm Hg, FiO2 of 60% and the ventilator rate of 45 breaths per minute
- intercostal chest drain was inserted on day 35 of life (? collection on the right side causing a mass effect), after which around 65 mL of blood was drained and slowly replaced with packed cells – bleeding difficult to control

Still no response

- Transient improvement in ventilation following the insertion of the chest tube f/b distress
- Switched over to high frequency oscillatory ventilation (HFOV) due to worsening respiratory distress and persistent hypercarbia.
- CRP 23 mg/dL, TLC -7400 per cmm with 72% neurtophils, platelet count - 37,000 per cmm

- Baby however, continued to worsen with progressive respiratory failure inspite of invasive ventilation (hypoxemic and hypercarbic respiratory failure)

- On day 38 of life (day 13 of hospital stay), baby succumbed to respiratory failure.

Final diagnosis

Kaposiform hemangioendothelioma with Kasabach–Merritt phenomenon

Discussion

THE JOURNAL OF PEDIATRICS • www.jpeds.com WORKSHOP/SYMPOSIUM SUMMARY

Consensus-Derived Practice Standards Plan for Complicated Kaposiform Hemangioendothelioma

Beth A. Drolet, MD[1], Cameron C. Trenor, III, MD[2], Leonardo R. Brandão, MD[3], Yvonne E. Chiu, MD[1], Robert H. Chun, MD[4], Roshni Dasgupta, MD[5], Maria C. Garzon, MD[6], Adrienne M. Hammill, MD, PhD[7], Craig M. Johnson, DO[8], Brook Tlougan, MD[6], Francine Blei, MD[9], Michèle David, MD[10], Ravindhra Elluru, MD, PhD[11], Ilona J. Frieden, MD[12], Sheila F. Friedlander, MD[13], Ionela Iacobas, MD[14], John N. Jensen, MD[15], David M. King, MD[16], Margaret T. Lee, MD[17], Stephen Nelson, MD[18], Manish Patel, DO[19], Elena Pope, MD[20], Julie Powell, MD[21], Marcia Seefeldt, RN[22], Dawn H. Siegel, MD[1], Michael Kelly, MD, PhD[23], and Denise M. Adams, MD[7]

- The radiology showed typical findings of reticular stranding of subcutaneous tissue.
- Despite the institution of appropriate therapy in the form of steroids/vincristine/antibiotics/fluids etc, the infant quickly deteriorated after the second day of admission, and thereafter despite another 10 days of ICU care, could not be saved.
-

- **Learning Points**
- Kaposiform hemangioendothelioma/tufted angioma with Kasabach-Merritt syndrome is a **TRUE DERMATOLOGIC EMERGENCY** and needs urgent recognition and treatment.

- Any delay can be fatal

CASE 8

DR RAHUL MAHAJAN

Demographic details

- Master K ; 14 years
- DOB- 14.07.2007
- Firozpur Punjab

- Chronic eczema since 1 year age
- Non-healing leg ulcers for last 5 years

History

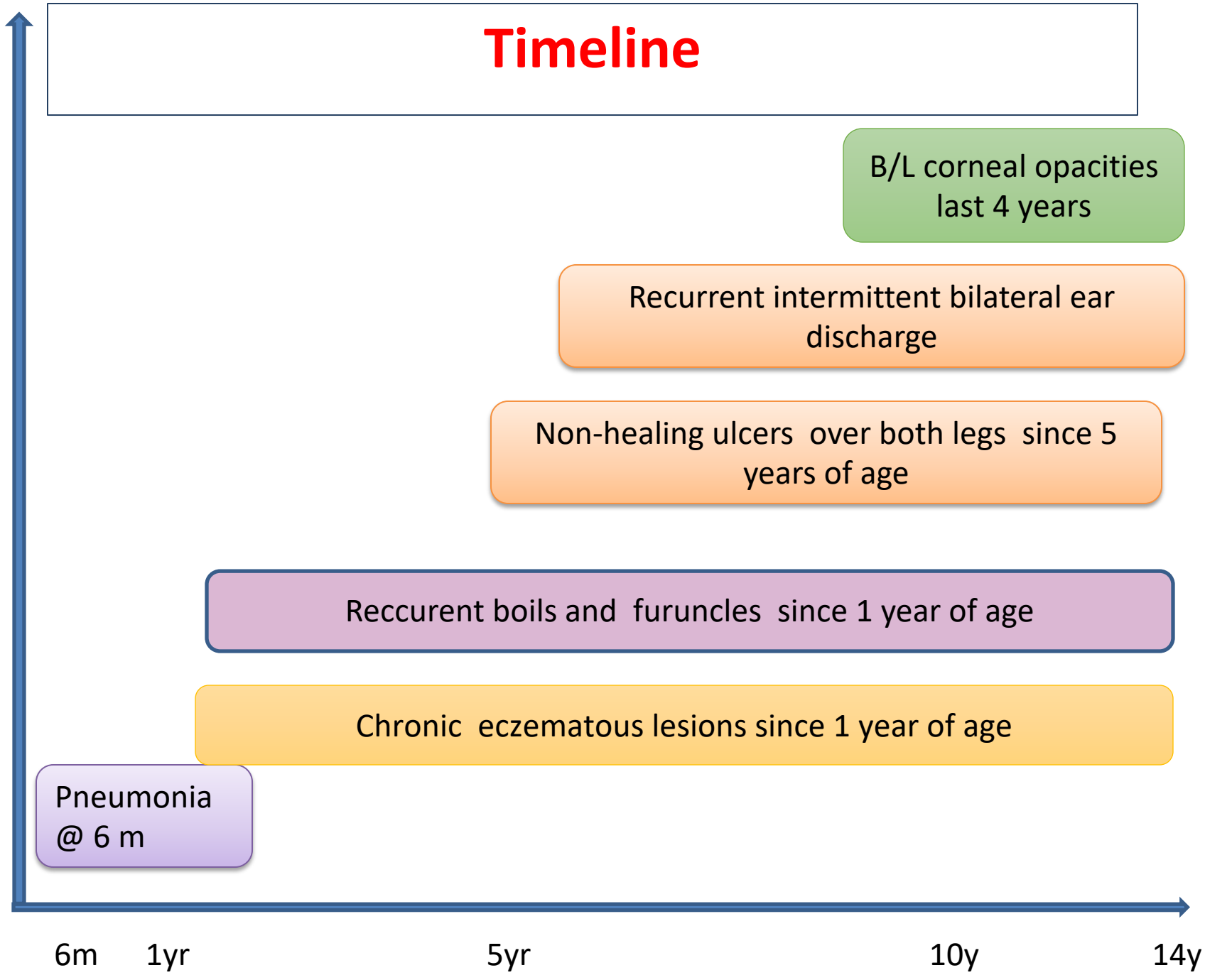
- Family history- third born child to a non-consanguineous marriage

  No similar history in the family
- Developmental history-

  Motor and language-normal

  Cognitive delay present (similar to 5 years old)
- Immunisation history- completed till date

Timeline


B/L corneal opacities last 4 years
Recurrent intermittent bilateral ear discharge
Non-healing ulcers over both legs since 5 years of age
Reccurent boils and furuncles since 1 year of age
Chronic eczematous lesions since 1 year of age
Pneumonia @ 6 m
6m
1yr
5yr
10y
14y

Examination

- Facies- cushingoid facies ,loss of scalp hair, scaly lesions over face ,short pointed nose, deformed left pinna,
- Eye- loss of eye-lashes, ectropion ,muddy sclera, corneal opacities

Clinical image

- Ear – no discharge at present
- Mild-pallor
- No edema,clubbing
- PR- 84/min
- RR- 24/min
- Temp.- 98.8 degree Fahrenheit
- BP – 112/76 mm of Hg (50-90th percentile)
- Right leg- 5*5 cm non-healing ulcer near ankle joint with foul smelling discharge
- Left leg- 9*5 cm non-healing ulcer around the ankle joint with foul smelling discharge

Examination

- Skin- generalised itchy eczematous skin with scaling
- Liver – 2cm below RCM , span -12 cm , firm
- Spleen- 2 cm below LCM ,firm
- Respiratory system – normal
- CVS – normal
- CNS- normal except cognitive delay

Investigations

INVESTIGATIONS	07.10.21
Hb (g/dl)	9.9
TLC (/cmm)	15,480
DLC	$P_{65}L_{25}M_{05}E_{05}$
Platelet (/cmm)	2,80,000
ESR	45 mm in 1st hr
CRP	180 mg/dl

INVESTIGATIONS	07.10.21
Total Protein(g/dl)	6.0
Albumin (g/dl)	2.1
SGOT(IU/L)	107
SGPT(IU/L)	28
Na/K/Cl (meq/l)	134/4.0/101
Urea(mg%)	10
Creatinine(mg%)	0.32

Dermatolgical examinations	10.10.21
Tzank smear	No organism
Skin Biopsy	Ulcer with granulation tissue
Pus culture	Methicillin resistant S.aureus

Immunological investigations

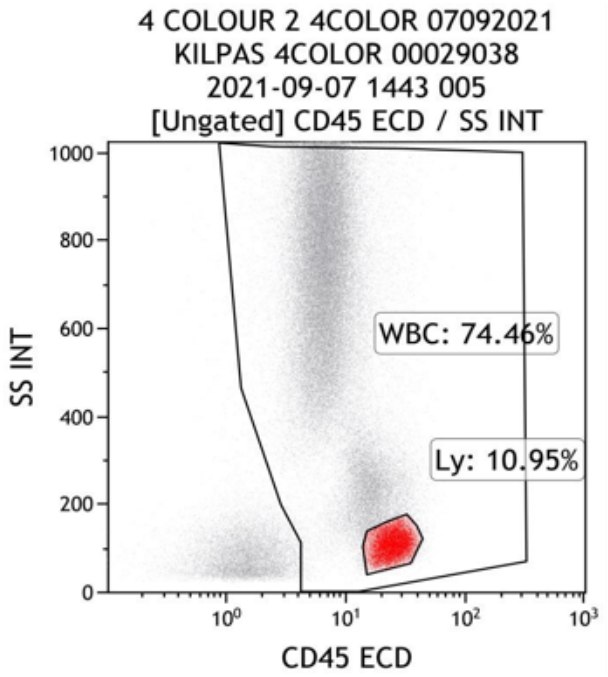
- HIV – non reactive

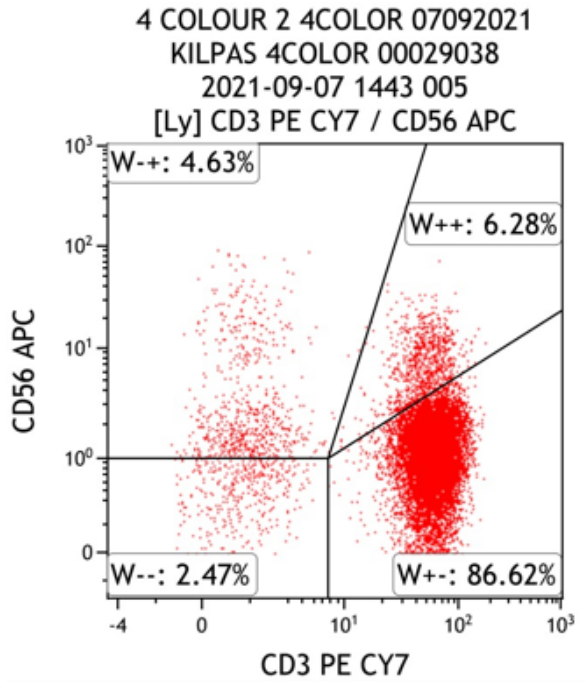
- IgG-2280(993-1992mg/dl)
- IgA- 409(70.9-284mg/dl)
- IgM-27 (54-255mg/dl)
- IgE- 16300 (<100 Ku/L)

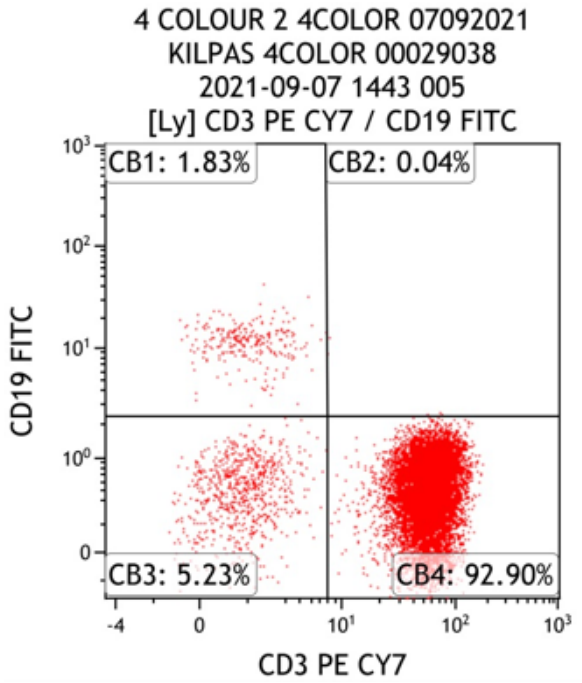
- IgG1-1550(377-1189mg/dl)
- IgG2-409(124-264mg/dl)
- IgG3-102 (34-118mg/dl)
- IgG4- 314 (7-57 mg/dl)

- NBT- normal
- C3- 158 (75-173 mg/dl)
- C4- 41 (14-40mg/dl)

Lymphocyte subset

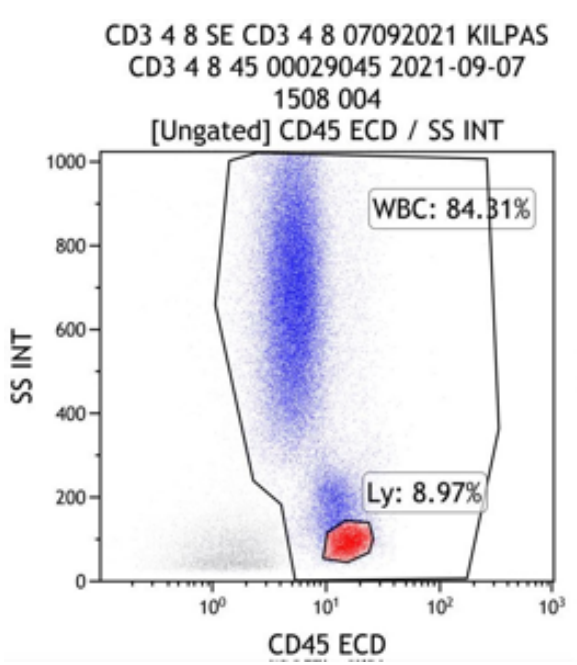


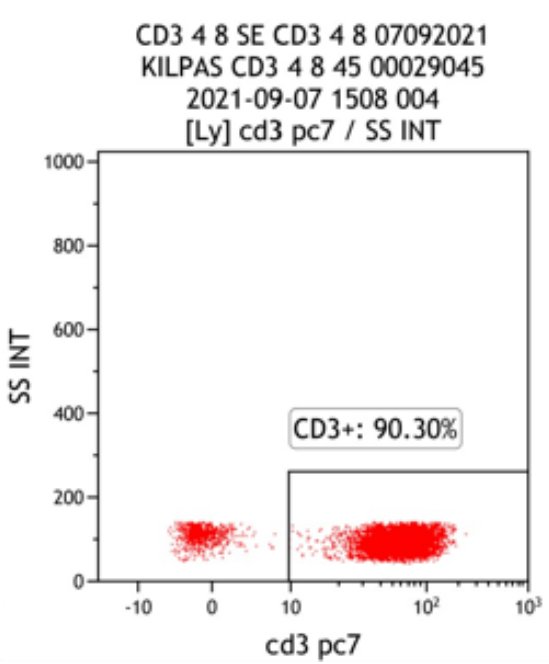


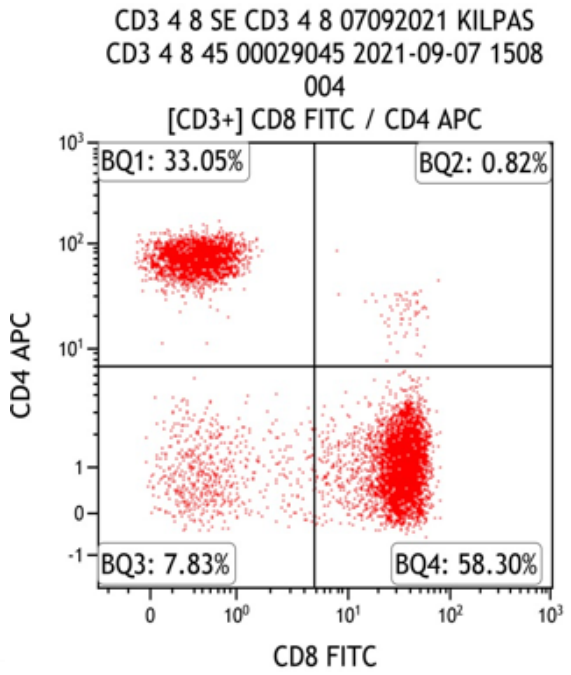


% of Populations	Normal	Patient
%CD3 + T cells	56-84	92.90
%CD19 + B cells	6-23	1.83
%CD56+ NK cells	3-22	4.63

T cell subset

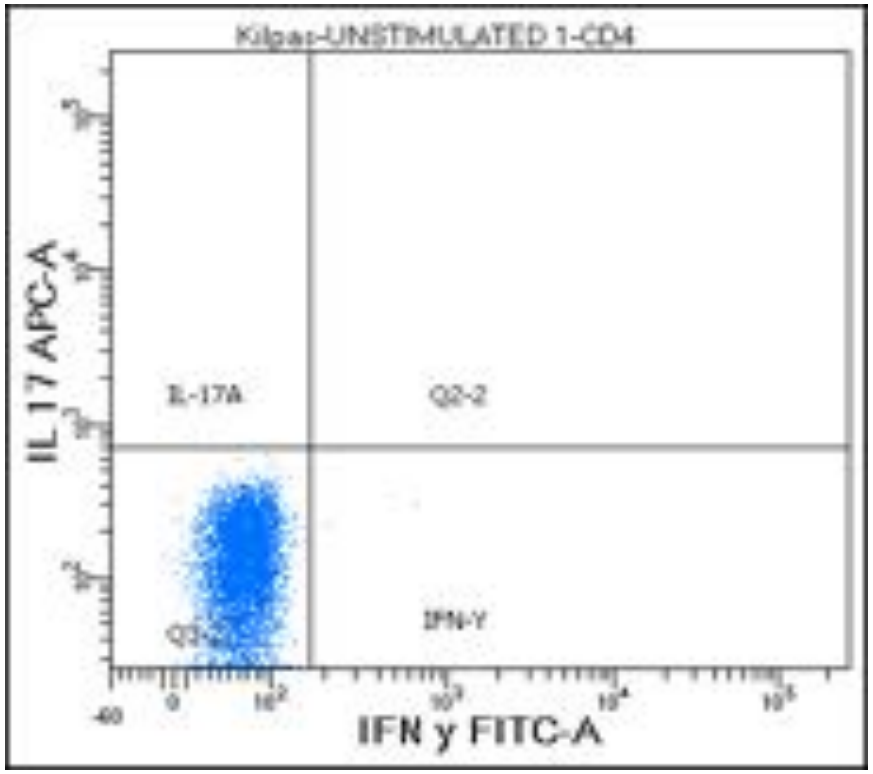


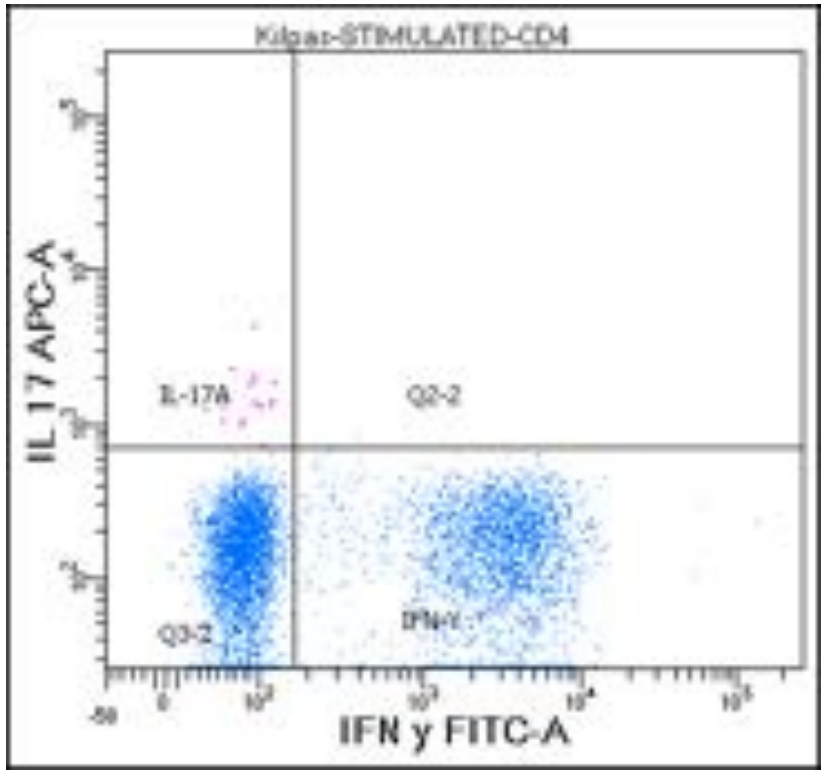




% of Populations	CONTROL	Patient
%CD4 + T cells	60.82	33.5
%CD8 + T cells	33.7	58.30
CD4 : CD8	1.8:1	0.57:1

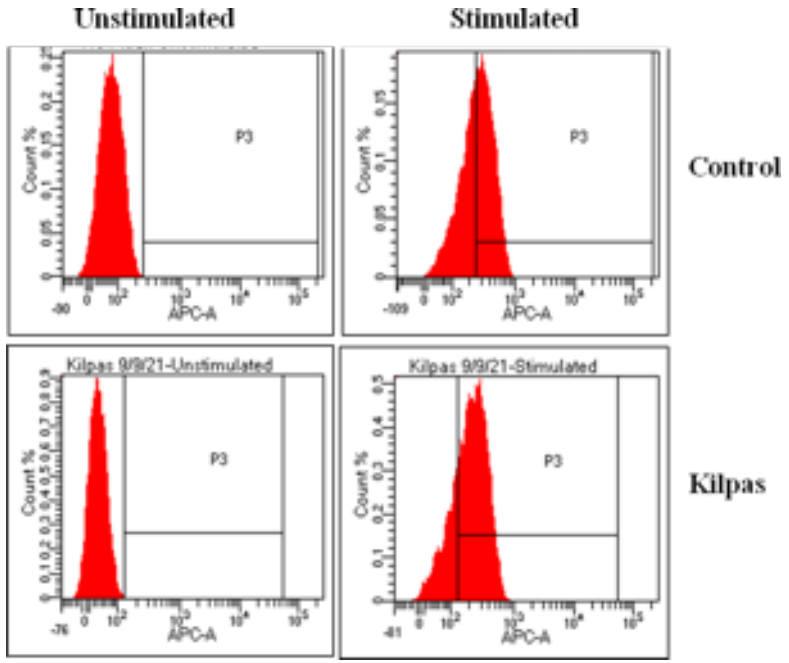
Th 17 assay





S. No	% of Populations	CONTROL	Patient
1.	CD4+ (T_H lymphocytes)	45.13% of Lymphocytes	57% of Lymphocytes
2.	CD4+ IFN-γ+ (Th1 Cells)	40.78% Of CD4+ Cells	42.9% Of CD4+ Cells
3.	CD4+ IL17A+ (Th17 Cells)	1.1% Of CD4+ Cells	0.3% Of CD4+ Cells

STAT 3 phosphorylation



	CONTROL	PATIENT
UN-STIMULATED	0.1%	0.1%
STIMULATED	44.4%	66.7%
MFI- (UN.ST)	63	53
MFI- (ST.)	243	269

Interpretation:

Normal STAT3 phosphorylation after stimulation with IL-6 suggest that protein is fully functional

Database

- Cognitive impairment
- Chronic eczema
- Non-healing leg ulcer
- Recurent ear discharge
- Abnormal facies
- Hyper Ig E
- Decreased B cell
- Altered CD4:CD8
- Reduced Th-17

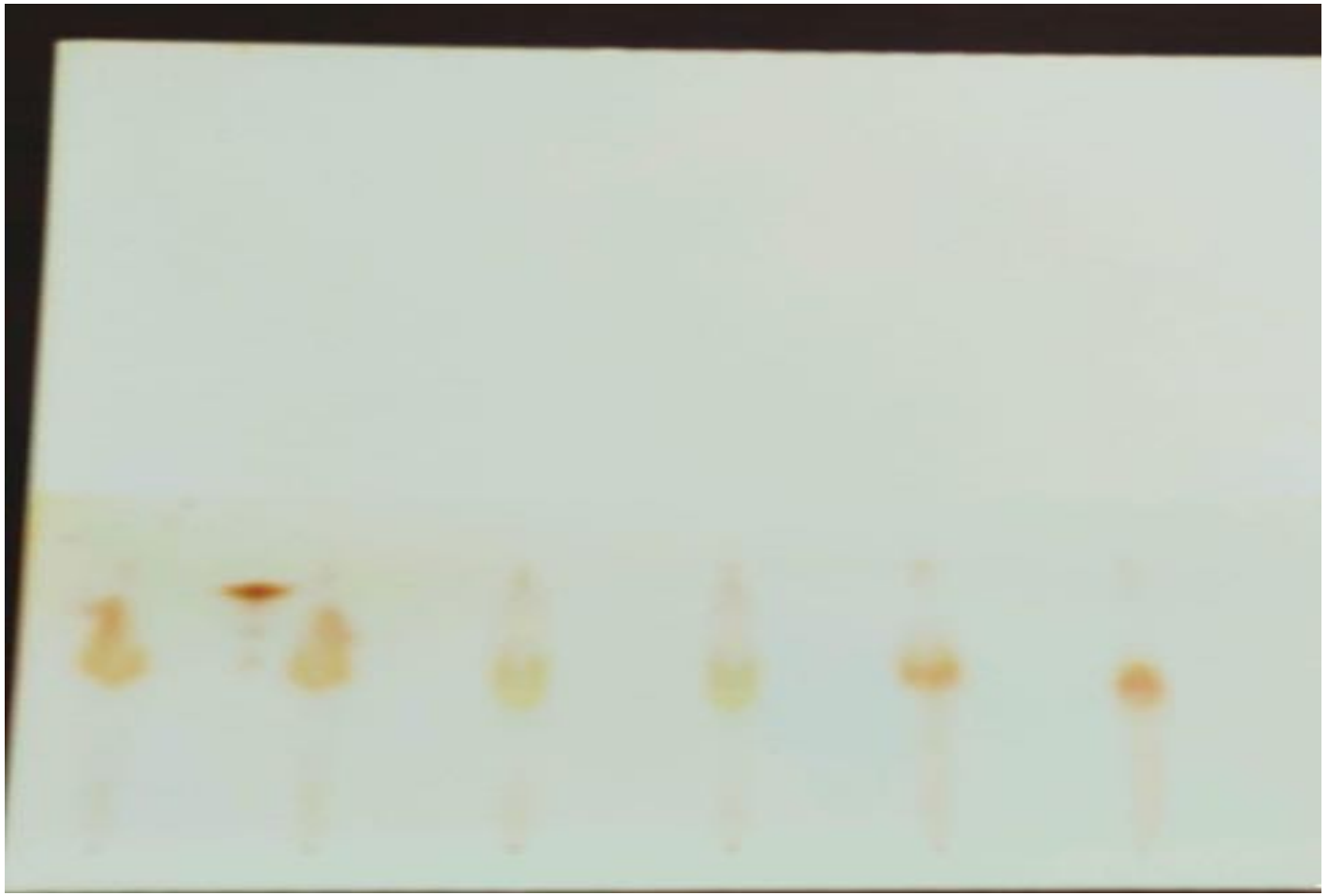
Possibilities

- Hyper-IgE syndromes-

- STAT3 LOF
- Autosomal recessive (DOCK8 or TYK2 mutation)

- Prolidase deficiency

Thin layered chromatography for urine proline



Yellow colour shows proline positivity.First two healthy control; next two patient and last two of the sibling

Whole exome sequencing

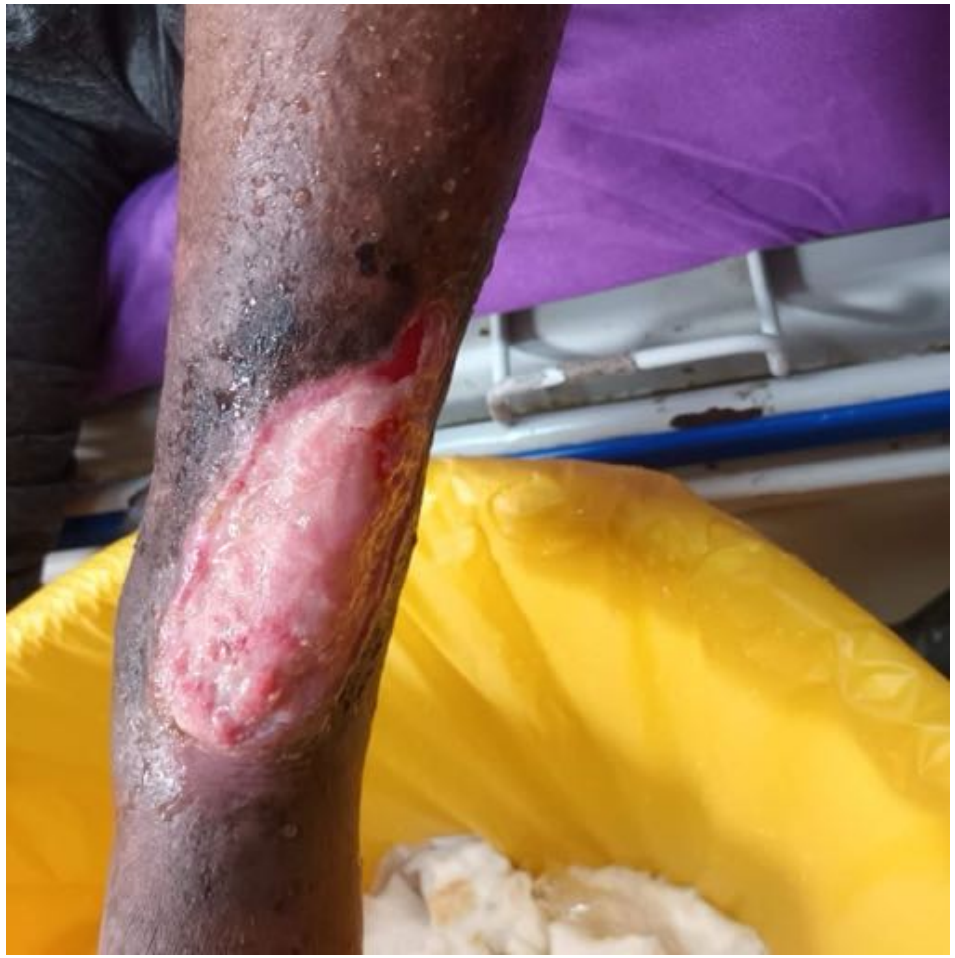
- Homozygous pathogenic variant in PEPD gene c825del(pPhe275Leufs*46)

- Diagnosis:AR prolidase deficiency

Management of index child

- Prolonged sensitive IV followed by oral antibiotics
- Local site dressing and application of Debridace (papain-urea) ointment
- Prophylactic antibiotics
- Short course of oral Steroids followed by yopical steroids for eczema
- Coconut oil for application in skin
- Antihistaminics

Clinical image on follow-up



CASE 9

DR RITA VORA

A CASE OF EXTENSIVE GIANT TUBEROUS XANTHOMAS IN A 12 YEAR OLD BOY

CLINICAL HISTORY :

- A 12-year-old, non-obese boy, born of **second degree** consanguineous marriage.
- He presented with asymptomatic swellings over the elbows, hands, buttocks, posterior thighs, popliteal fossa, knees, and ankles since 7 years.
- Lesions started over the buttocks, were initially small and gradually increased in size and number while progressing to other body parts.
- His maternal uncle had similar complaints.

- There was no history of breathlessness, palpitation and easy fatiguability.
- No H/O joint pain or muscle pain
- There was no history suggestive of premature cardiac arrest or stroke in family members.
- No H/O similar complaints in other siblings

On cutaneous examination:

- There were multiple skin-colored nodular lesions, ranging in size from 0.5 cm to 10 cm in diameter and distributed symmetrically. (Figure a & b)

- Lesions were non tender, immovable and did not change their size and shape with application of pressure.

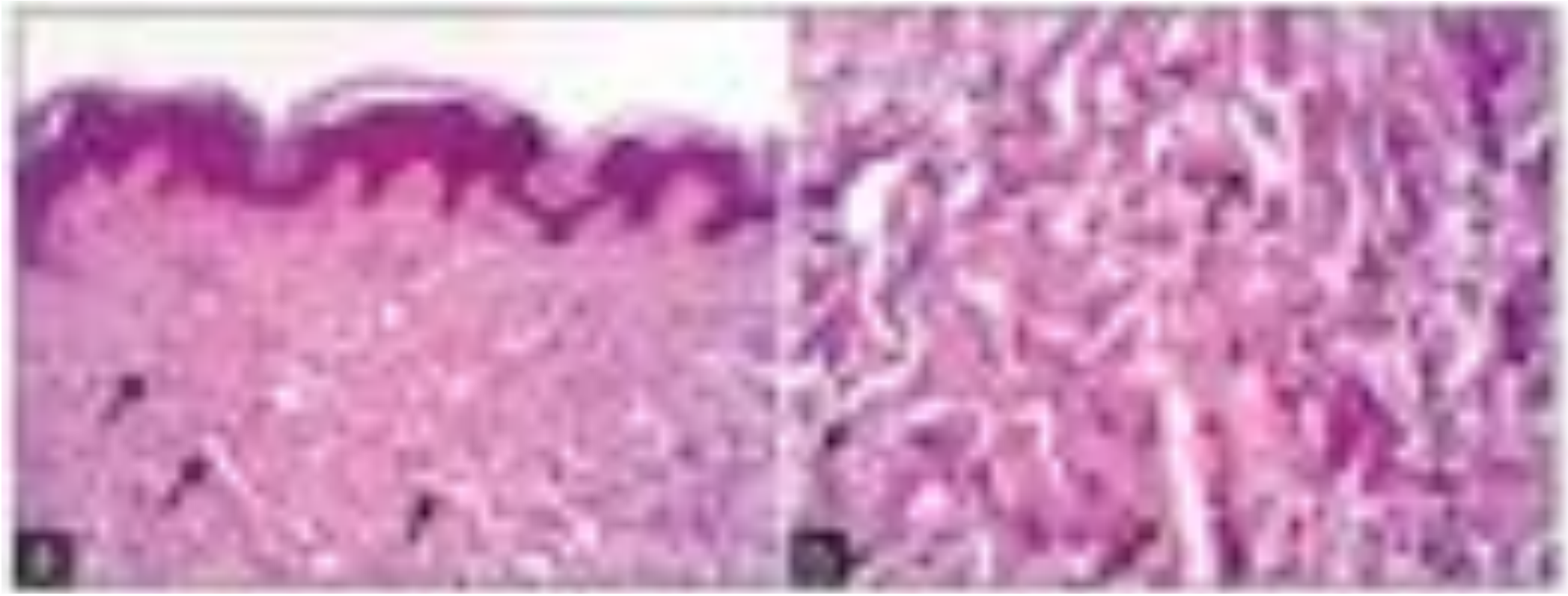
INVESTIGATIONS:

- Routine hematological investigations (CBC, LFT, RFT) were normal.

- **S. Fasting Lipid Profile**

- ❑ High total cholesterol (619 mg/dl)
- ❑ High low-density lipoprotein (LDL) - 537 mg/dl
- ❑ Normal high-density lipoprotein (HDL) - 60 mg/dl
- ❑ Normal Very low-density lipoprotein (VLDL) - 20 mg/ dl
- ❑ Normal Triglycerides - 102 mg/dl

- Lesional punch biopsy was done and was suggestive of foamy macrophages and cholesterol clefts along with collagen bundles in the dermis. (Figure a & b)



- As elevated cholesterol and LDL may cause atherosclerosis so ECG was done and was suggestive of left ventricular hypertrophy. Patient was advised furtherer cardiac evaluation but was lost to follow up.

- S. TSH and S. amylase and lipase and fasting blood glucose done to rule out secondary hyperlipidemia.

- Abdominal ultrasound was done to rule out biliary involvement and it was found to be normal.

- Chest X ray was done to rule out sarcoidosis and found to be normal.

- Slit lamp and fundus examination of the eyes were done to detect corneal arcus. It was found to be normal.

- Patient was advised Doppler echocardiography but did not turn up in follow up.

- Lipoprotein electrophoresis is expensive and is unnecessary for the diagnosis of hyperlipidaemia.

- LDL receptor analysis can be used to identify the specific LDL receptor defect but it is also expensive have no impact on management.

- Other tests which can be done:

- ❑ Radiographic imaging of Achilles tendon to measure the Achilles tendon xanthoma but findings do not change lipid management.

- ❑ Beta liquefaction (ultracentrifugation and electrophoresis) when diagnosis of hyperlipidaemia is in doubt.

CASE 10

DR SUDARSHAN P GAURKAR

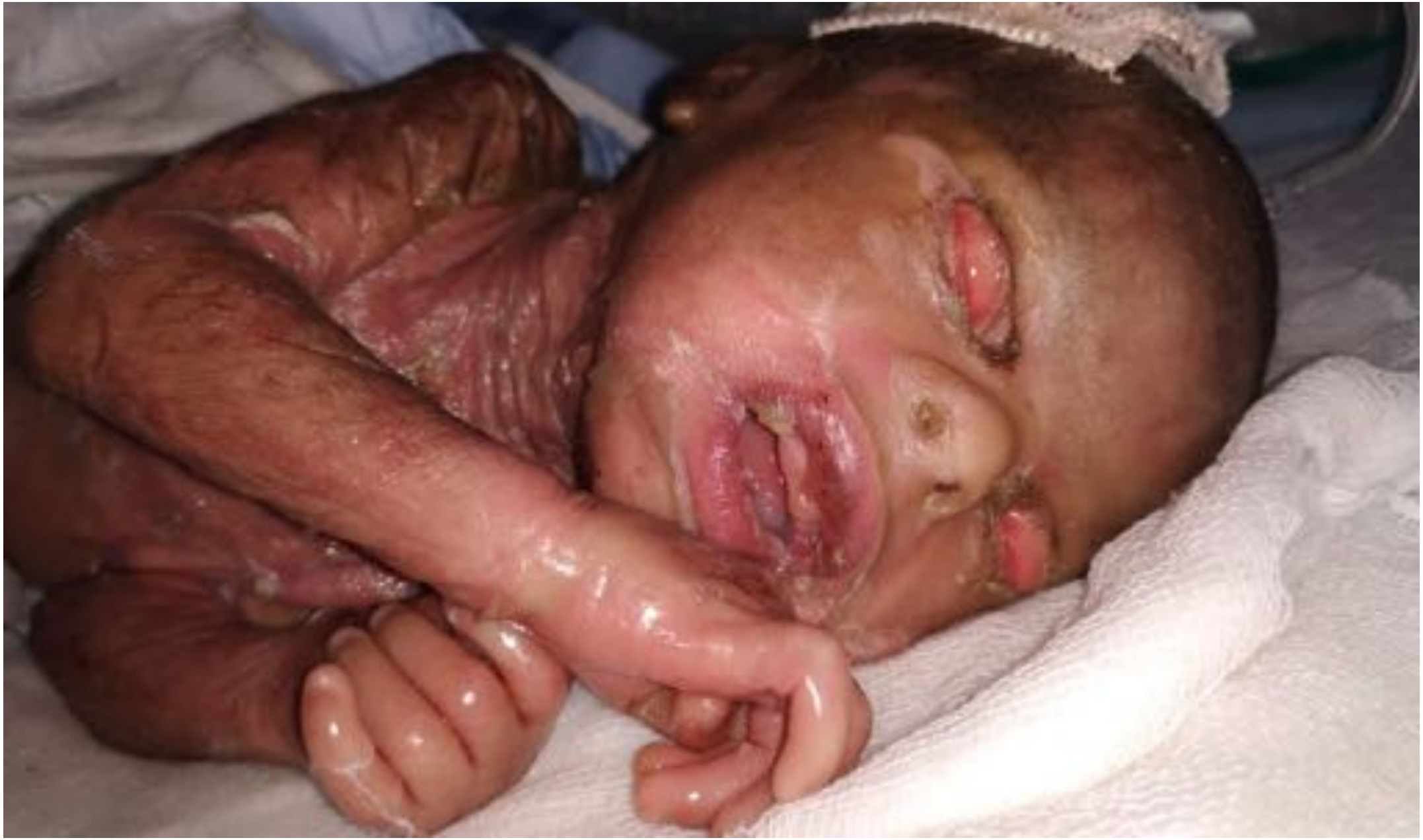
CASE Presentation

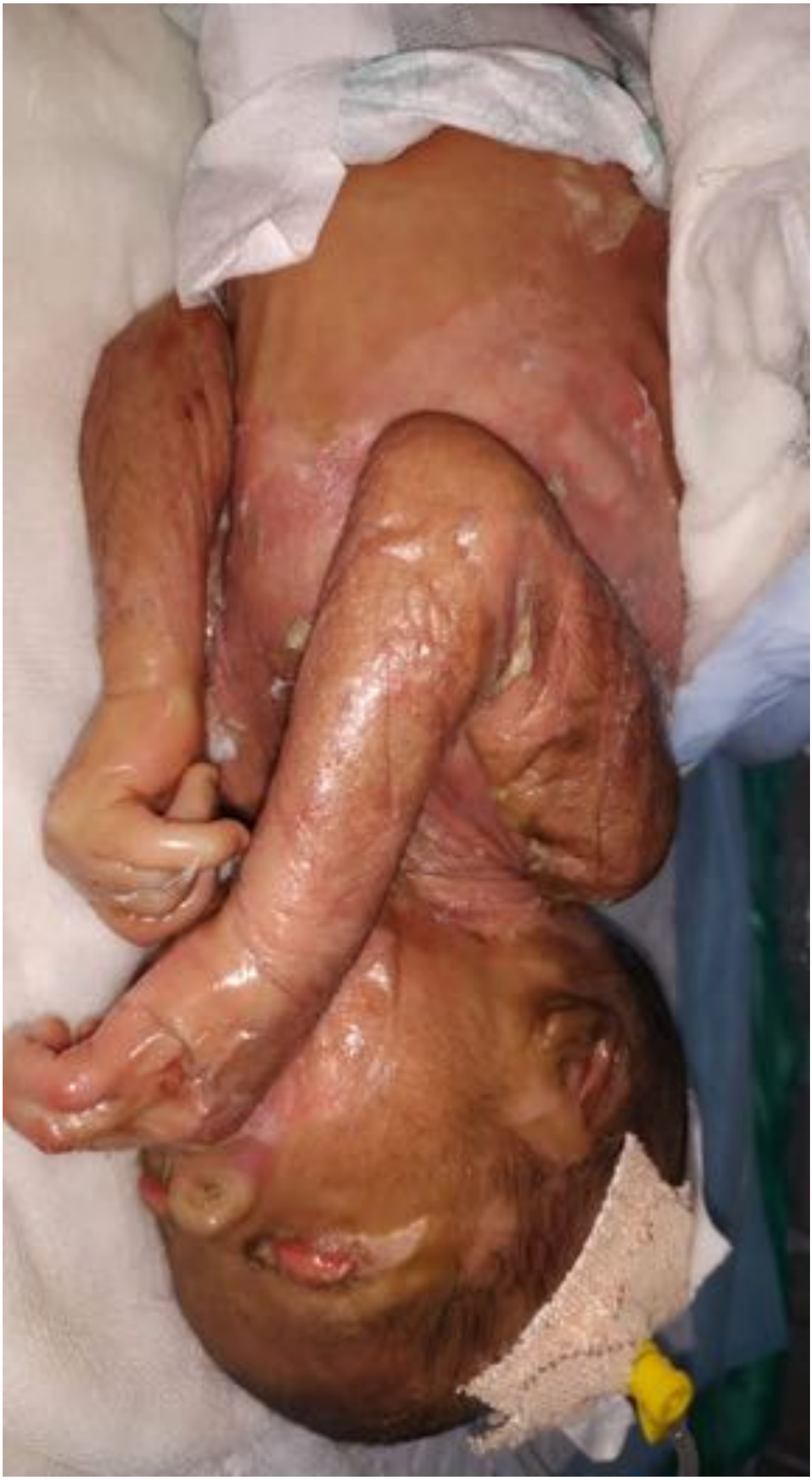
A 32 weeks gestational age male child, born of consanguineous marriage of birth weight 2100 grams, born to a primipara of age 29 years by caesarean section had –

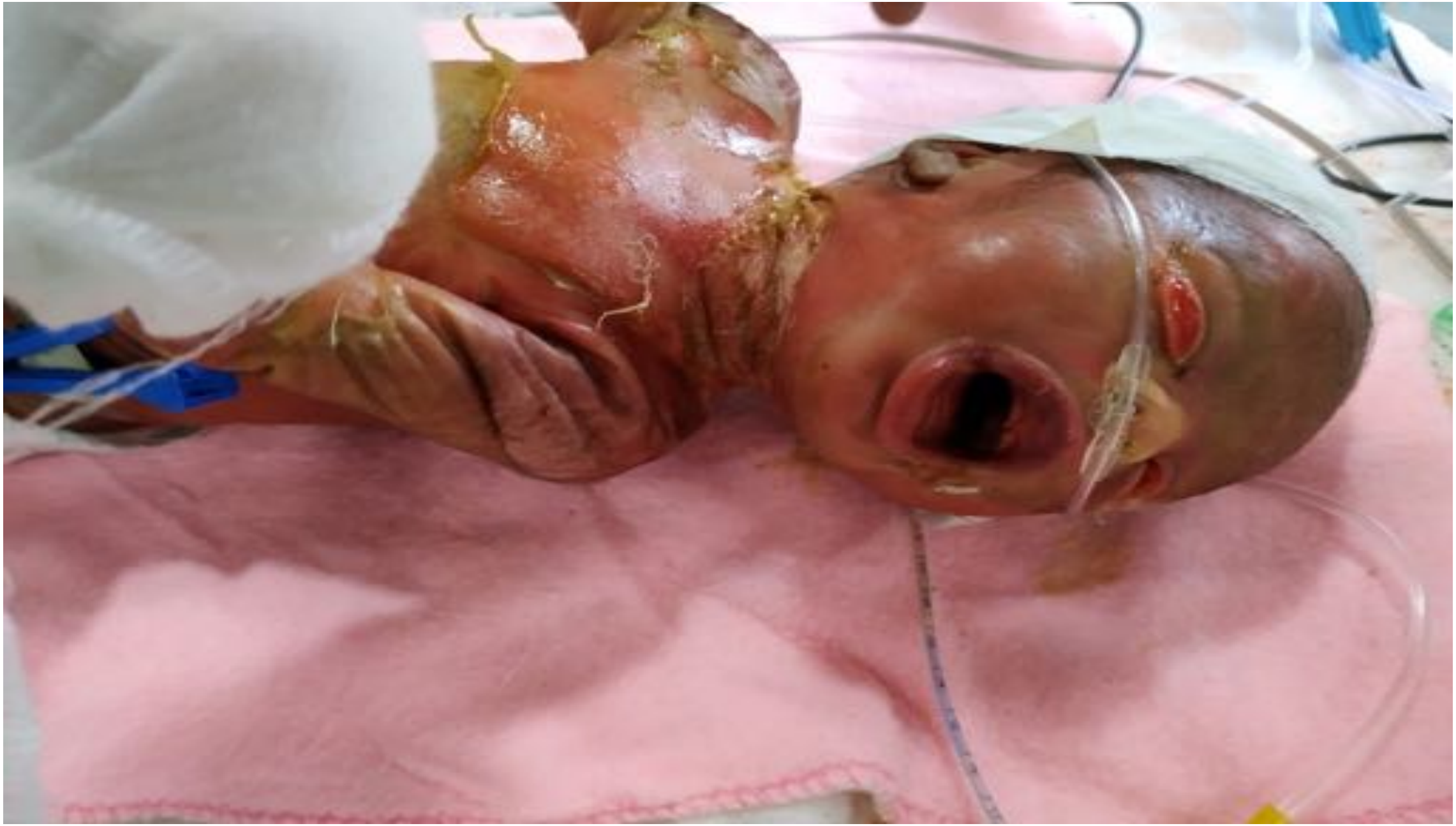
An Thick armor like appearance of skin with deep fissures at places

Associated with ectropion and eclabium giving a fish mouth appearance.

low set fused dysplastic ears associated with laboured breathing and inability to close the mouth.







Clinical differentials

Differentials include causes of a collodion baby

1. Congenital ichthyosiform erythroderma
2. Lamellar Ichthyosis
3. collodion baby
4. Sjogren Larsson syndrome

INVESTIGATIONS

Complete blood count:

Haemoglobin- 13.1 g/dl

Total leucocyte count-19,000/cmm

Platelet count- 2,50,000/cmm

Renal function tests:

Serum creatinine- 0.8 mmol/l

Blood urea- 34g/dl

Liver function tests:
Total serum bilirubin- 0.8
Direct bilirubin- 0.3
Indirect bilirubin-0.4
Serum electrolytes:
Serum sodium levels- 151 mmol/l
Serum potassium levels-3.1 mmol/l
Random blood glucose levels- 140g/dl

The laboratory investigations were suggestive of electrolyte imbalance

Rest all the labs were within normal limits

Chest Xray , ECG and USG abdomen Pelvis were also within normal limits

Follow up

The baby was diagnosed with Harlequin ichthyosis based on the classical clinical picture.

Other differentials were ruled out based on the clinical picture .

The baby was managed with oral isotretinoin at a dose of 5mg/day
Improvement in the cutaneous lesions was seen within 2 weeks with reduction in ectropion and eclabium and reduction skin fissures with reduction in scales.

Response after 2 weeks of starting isotretinoin

DISCUSSION

Acitretin is the drug of choice

The patient being from very poor background was non affording for Acitretin

So we managed the baby with isotretinoin 5mg along with the correction of fluid electrolyte imbalance under a paediatrician in a neonatal intensive care unit.

Mode of administration -5mg capsule to was opened,contents Were mixed With honey,given orally daily for 1month.
Patient survived 1month period and was dicharged from NICU. Was better 15 Days after Later on was lost to follow up.

ABCA12 gene(on chromosome 2q35) product is a protein that functions in the intracellular lipid transport in keratinocytes.

Defective gene causes inability to transport important lipids and enzymes to interstitial space between the keratinocytes.

Harlequin Ichthyosis is associated with premature birth and is frequently lethal due to neonatal complications of fluid loss and sepsis.

However, oral retinoids combined with intensive neonatal care could prove to be promising to reduce the mortality associated with this rare disorder.

THANK YOU.