MECHANOBULLOUS SKIN DISEASE

BY Dr ADEJUMO O.A

OUTLINE

- Introduction
- Types
- Clinical Features
- Diagnosis
- Treatment
- Complications
- Prognosis
- Conclusion

INTRODUCTION

- Is a skin disease caused by inherited abnormalities in structural proteins of skin leading to skin fragility
- Presentation varies in severity from mild to lethal
- Blistering may occur following minor trauma and usually shortly after birth
- Hot weather may also precipitate blistering.

- It is a rare heritable skin disease
- There is no sex, ethnic or racial predilection
- Incidence is about 1: 50,000 population
- Mode of inheritance is mainly AD, but may be AR too. The latter is associated with more severity

TYPES

- Epidermolysis Bullosa Simplex: 92%
- Epidermolysis Bullosa Dystrophica: 5%
- Junctional Epidermolysis Bullosa: 1%
- Unclassifed

EPIDERMOLYSIS BULLOSA SIMPLEX

- Is a genodermatosis characterized by superficial blistering of the skin with or without the mucous membrane owing to genetic mutations of cytoskeleton proteins within the basal layer of the epidermis
- Affected proteins are keratin 5, 14
- Autosomal dominant and recessive mode of inheritance

- There are few reported cases in Nigeria: Shehu and Uloko reported a case of Recessive Epidermolysis Bullosa Simplex in Kano.
- Useni et al reported 3 cases between 1999- 2002 in Owo.
- Odey et al reported a case of Junctional EB in Calabar in 2009
- 2 cases of EBS have been seen in UBTH in the last 4 years

TYPES OF EBS

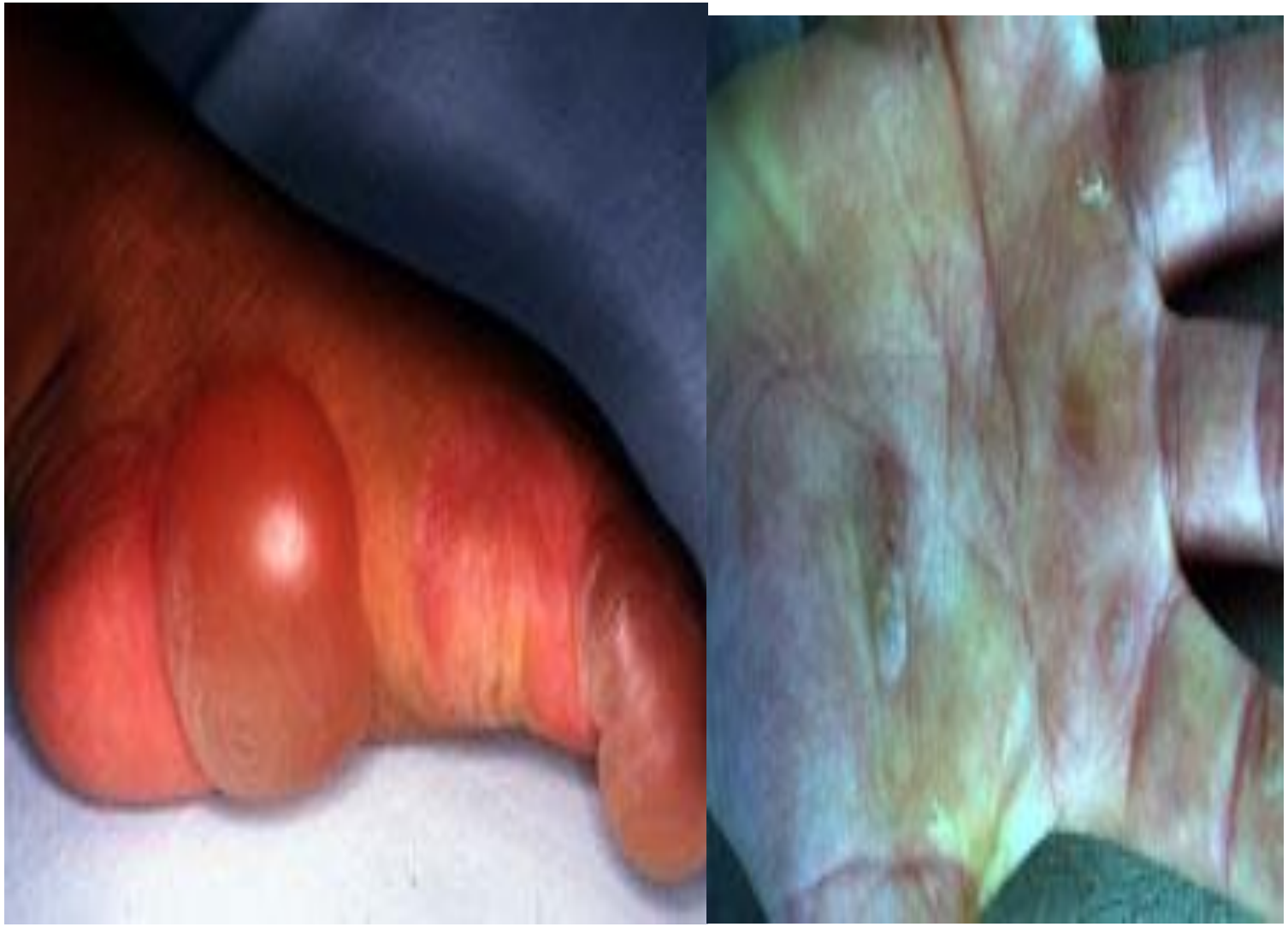
- Generalized EBS : Koebner variant
- Localized EBS Weber Cockayne variant
- EBS of Oga
- EBS with Muscular Dystrophy
- EBS with Mottled Pigmentation
- Epidermolysis Bullosa Herpitiformis: Dowling Meara EBS

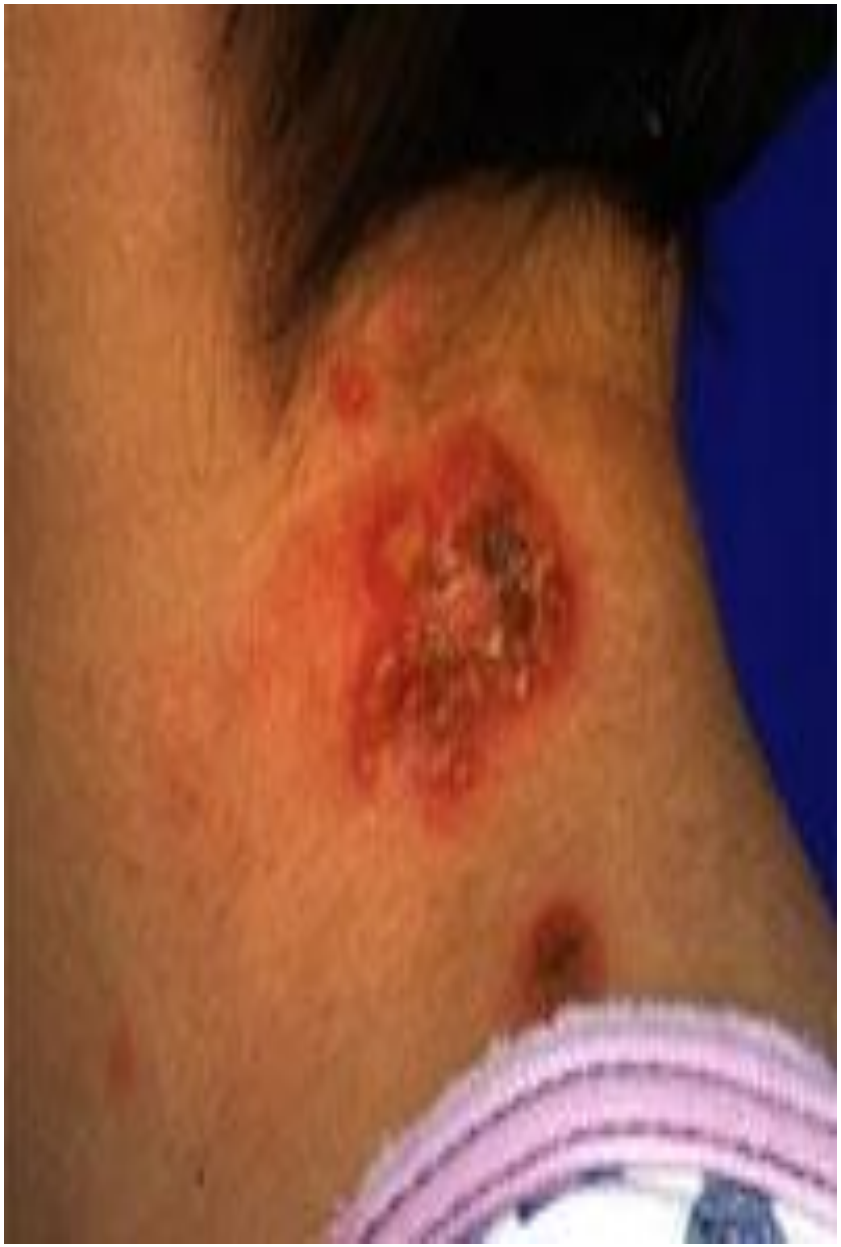
PATHOPHYSIOLOGY

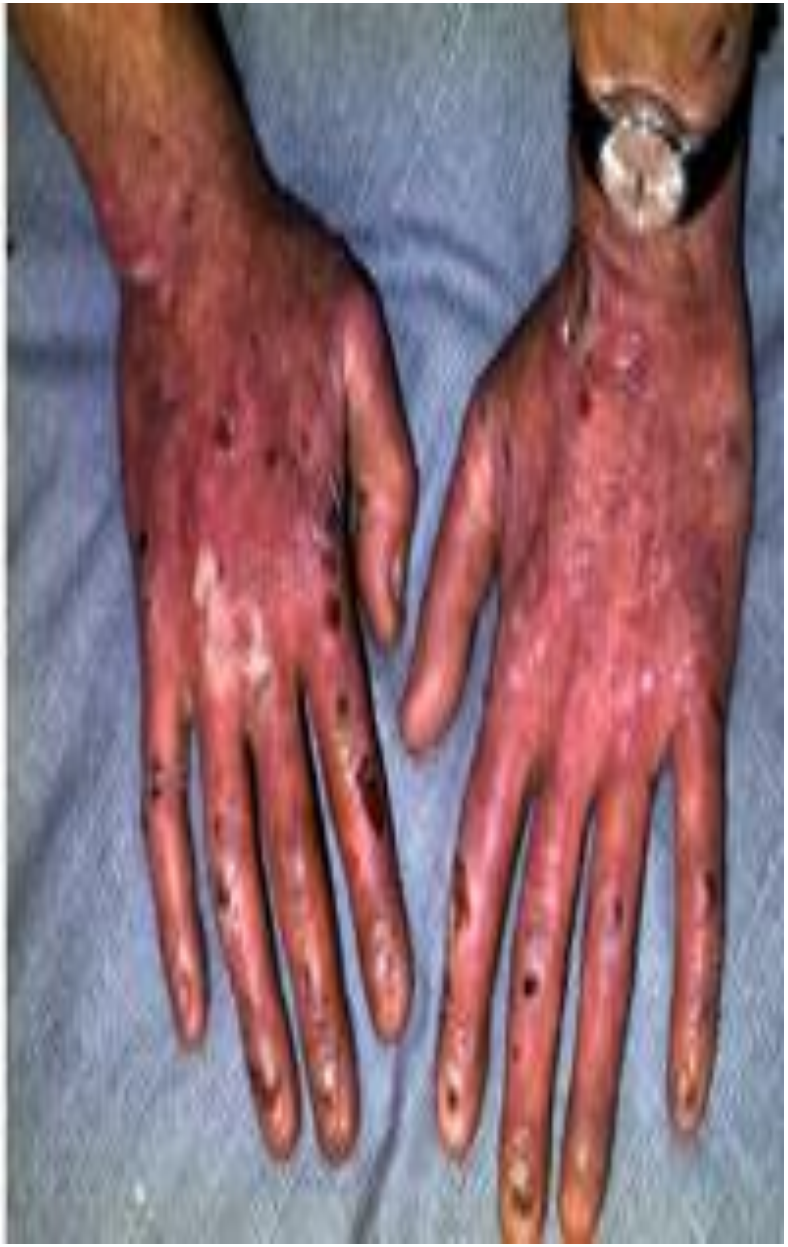
- There is mutation of genes such as KRT5, KRT14, PLEC that code for keratin4, keratin14 and Plectin respectively
- These proteins provide tensile strength and resilence to the epidermis. They also anchor the 2 layers of the skin so that they do not move independently
- Absence of these proteins makes the skin to be fragile, allows independent movement, hence causing shearing, blister formation with minimal trauma.

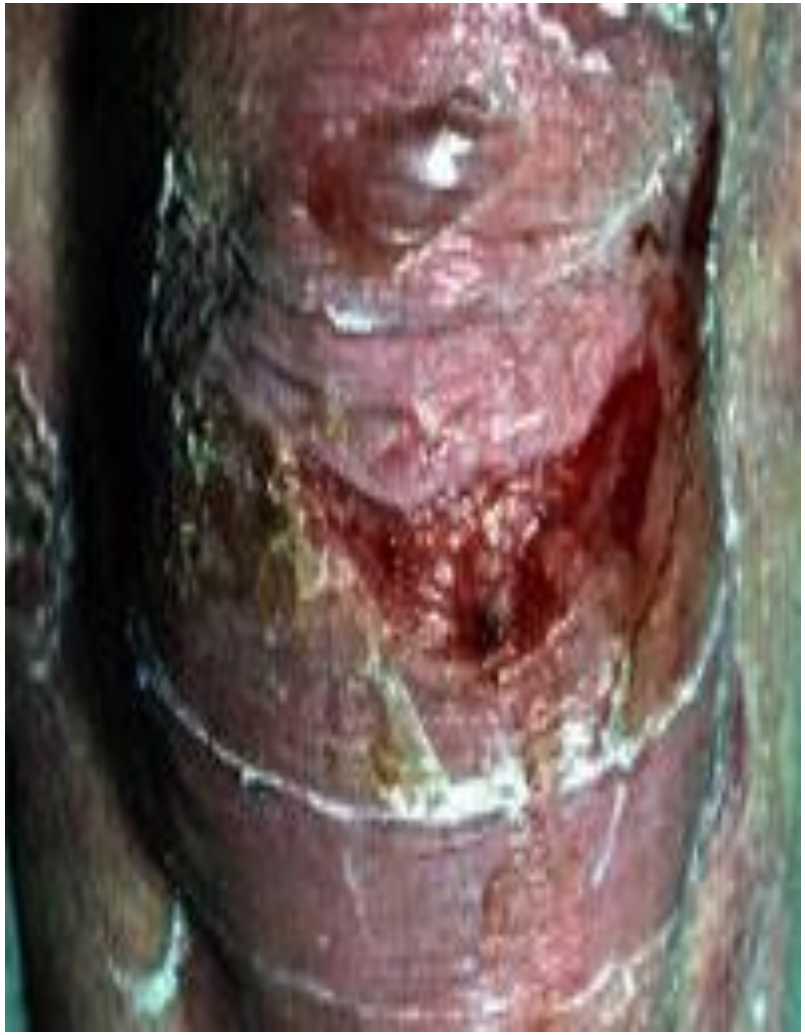
CLINICAL FEATURES

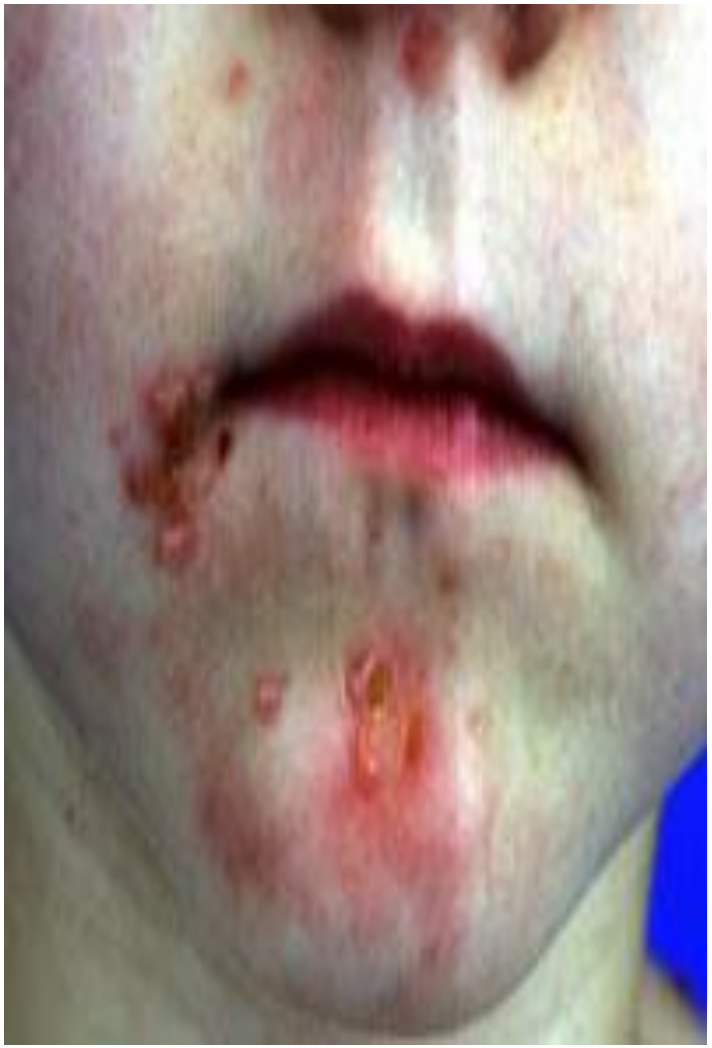
- Skin Blistering of varied severity
- Alopecia
- Nail Deformity or Loss
- Tooth Decay
- Dysphagia
- Hoarseness
- Joint Deformity

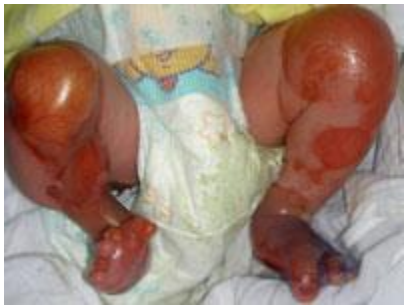


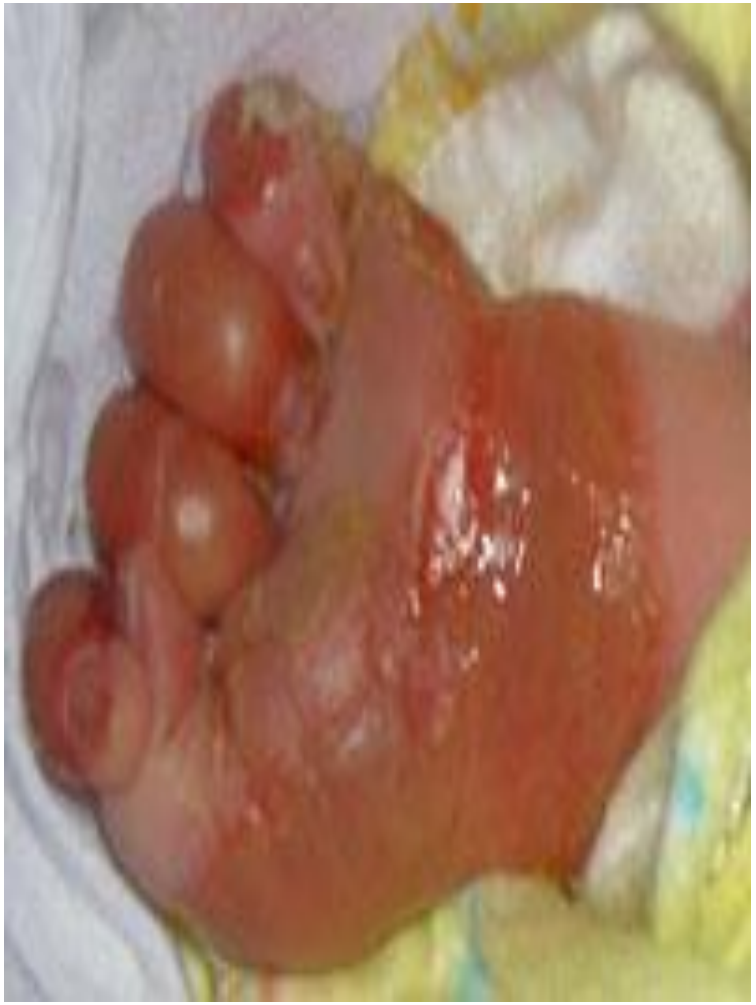


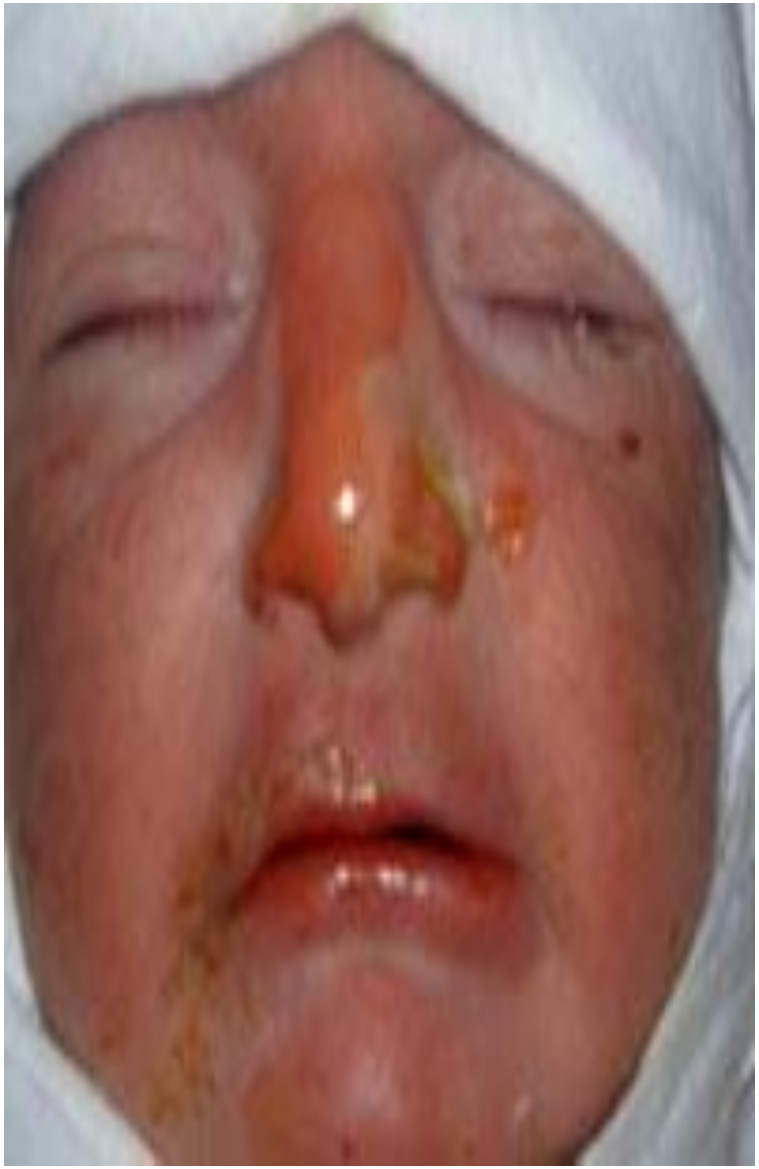


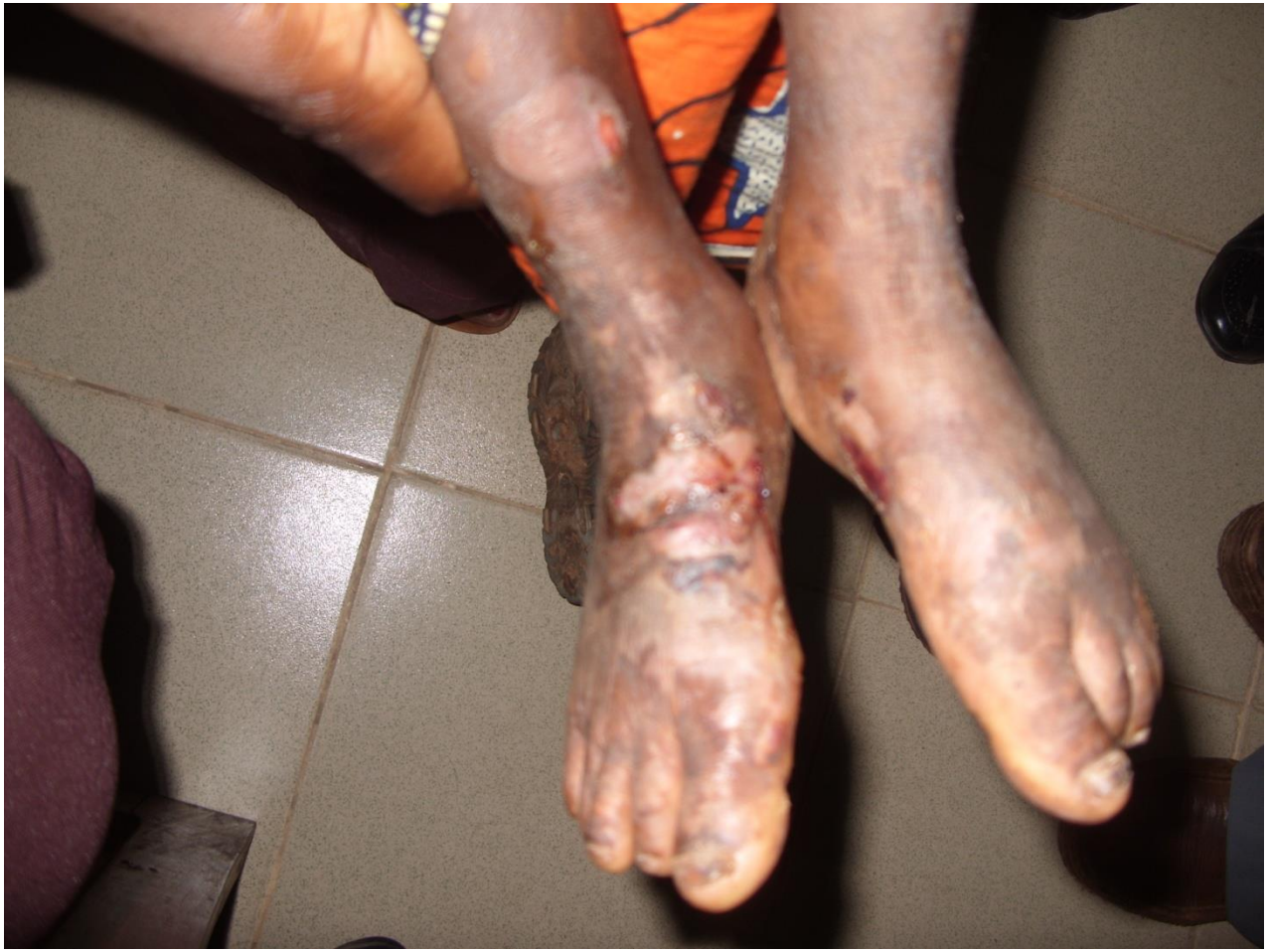


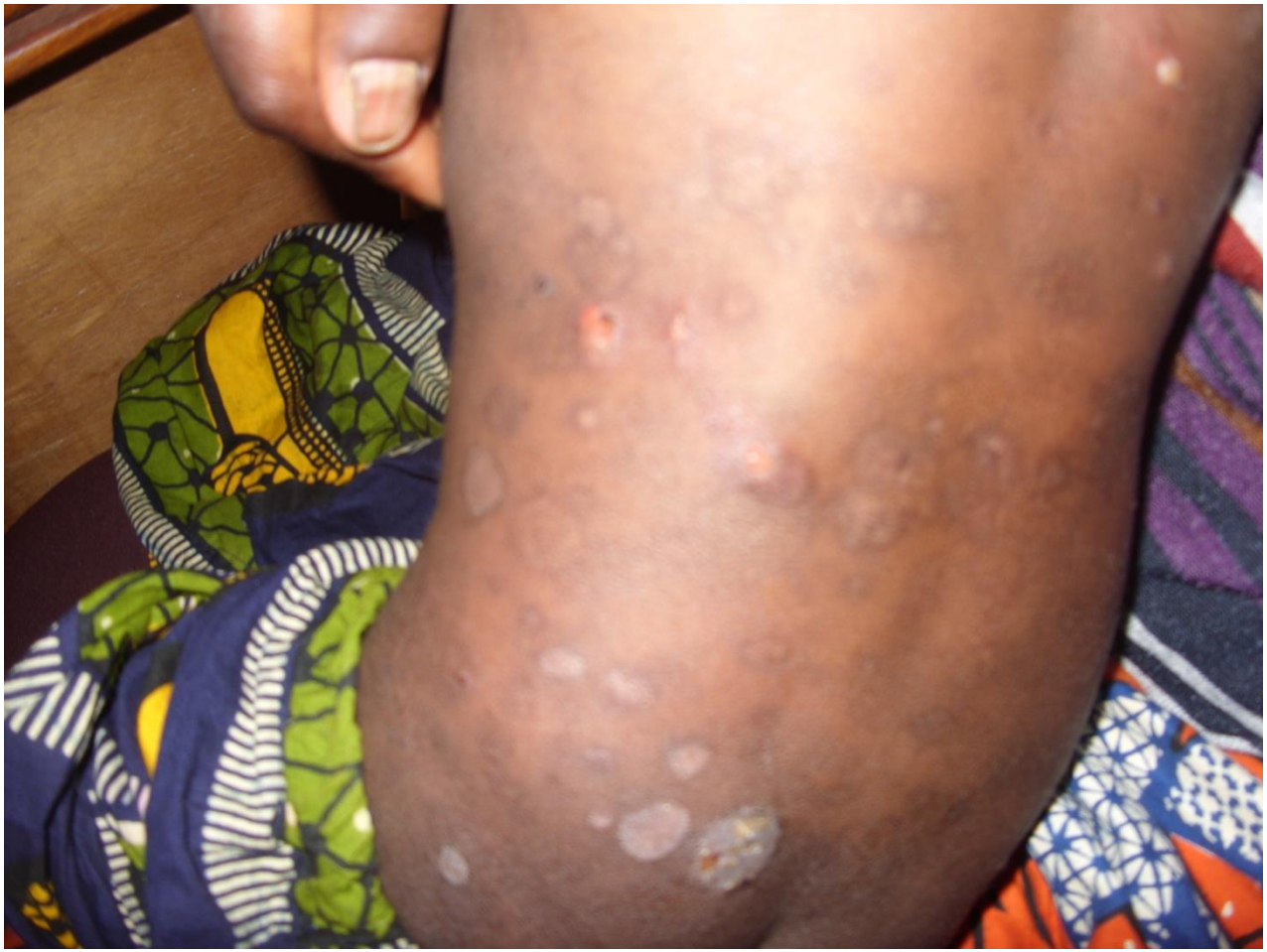


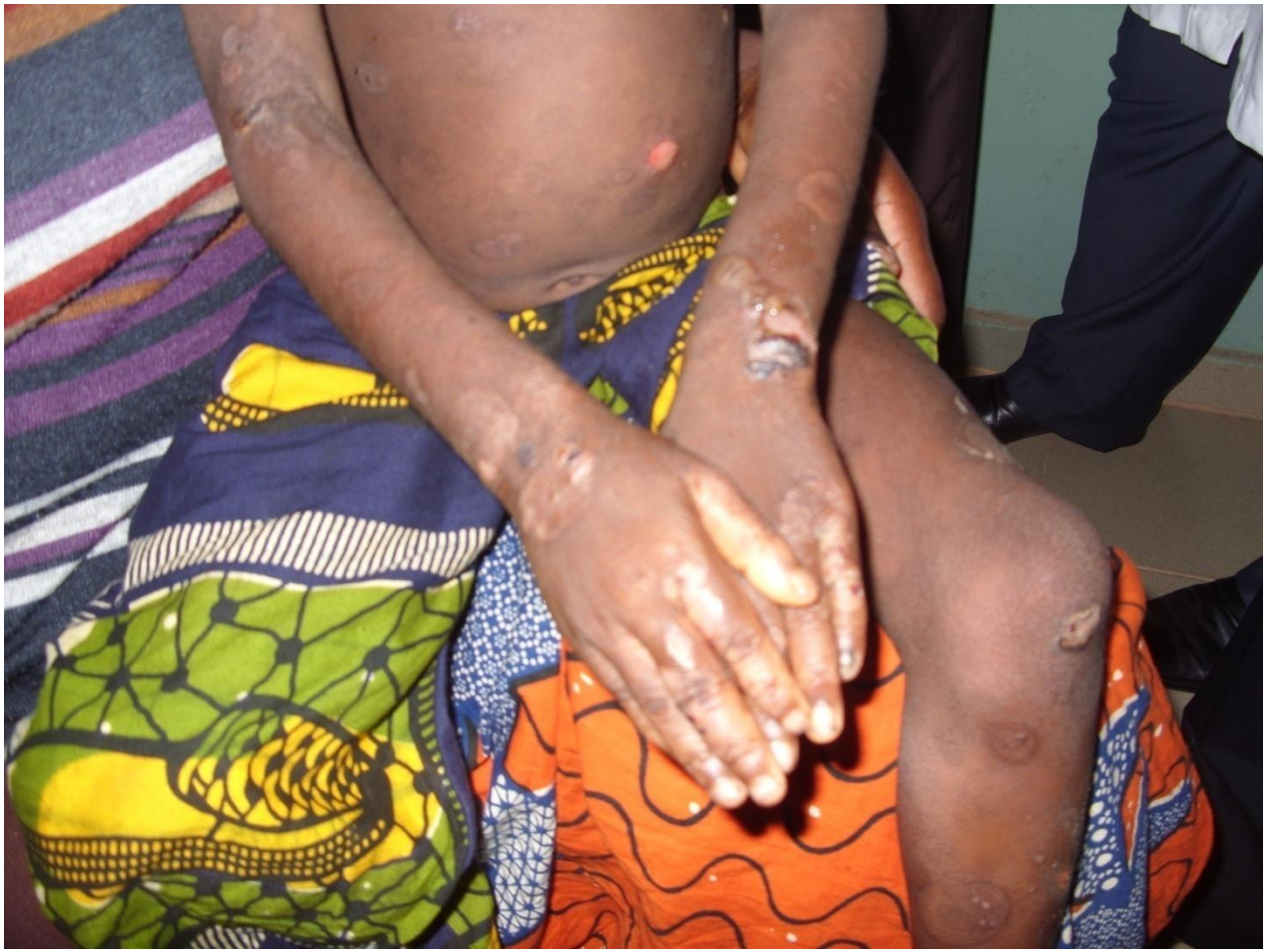












DIAGNOSIS

- History
- Physical Findings
- Skin Biopsy and Electron Microscopy
- Skin Swab MCS
- Full Blood Count

TREATMENT

- Goals of treatment is to prevent formation of blisters and to prevent complications
- Treatment is Multidisplinary in approach
- Topical Antibiotics
- Wound Dressing
- Antifungal
- Good Oral Hygiene
- Steroids with or without Ca and vit D supplement

TREATMENT

- Nutrition
- Physiotherapy
- Skin Grafting
- Oesophageal Dilatation
- Excision of Cancerous Skin Ulcers
- Sulforaphane
- Gene Therapy
- Bone Marrow Transplant

PROGNOSIS

- This is dependent on the subtype and the severity

COMPLICATIONS

- Anaemia
- Eosophageal Stricture
- Blindness
- Sepsis
- Muscular Dystrophy
- Peridontal Disease
- Malnutrition
- Malignacy especially Sqamous Cell Carcinoma
- Failure to Thrive

PREVENTION

- Genetic Counselling
- Prenatal Diagnosis; Chorionic Villous Sampling

CONCLUSION

- Epidermolysis Bullosa Simplex is a rare genadermatosis with varied degree of skin affectation.
- Treatment presently is conservative, although definitive treatment is at the experimental phase and is quite promising.

- THANK YOU FOR LISTENING