GENETICS AND GENODERMATOSES

EXUBERANT GRANULATION TISSUE: BOON OR BANE? - A RARE CASE OF LARYNGO-ONYCHO-CUTANEOUS SYNDROME.

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Background- Laryngo-onycho-cutaneous (LOC) syndrome is a subtype of junctional epidermolysis bullosa showing autosomal recessive inheritance, presenting with abnormalities of the voicebox (laryngo-), nails (onycho-), and skin (cutaneous).It is a rare syndrome with fewer than 50 cases reported to date. It is important to diagnose this syndrome so as to be prepared to deal with its grievous and fatal complications.

Observation- A 20yr old unmarried muslim female patient was referred from the ENT hospital with complaints of bleeding from the nose, hoarseness of voice and eye complaints since infancy. She gave history of fluid filled lesions over the knees and neck in infancy and nail changes since 17 years. There was history of similar lesions in the patient's sister. On examination- red granulation tissue in the anterior nares, symblepharon of both eyes, nails were dystrophic, teeth-mild enamel hypoplasia, scalp and hair - normal, genitalia- normal. Based on clinical examination a diagnosis of laryngo- onycho-cutaneous syndrome was made.

Key message- It is important to counsel the patient and family about the poor prognosis and complications of this syndrome such as the increased risk of mucosal and skin granulation tissue formation that leads to delayed wound healing, laryngeal obstruction and blindness. At present there is no cure for this syndrome but novel treatment strategies such as cultured keratinocyte grafts, fibroblast and stem cell therapies are being studied.