PS2.004

Epidermolysis bullosa or child butterfly

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It is considered rare disease with low prevalence 2 / 1000,000 live births. It is an autosomal recessive / dominant inheritance by type. The problem is alteration of proteins epidermo-dermal junction that alters the binding of the epidermis to the dermis, resulting in blisters and skin and mucous erosions. There is a type associated with Crohn acquired and Lupus.
The clinic is characterized by fragile skin with blisters and ulcers that cause different alterations, from alt alopecia. nails, esophageal strictures that can alter nutrition with growth retardation, blindness and serious consequences depending on the type.
Diagnosis:
Clinical: blisters and sores on skin very fragile
Skin biopsy of affected skin and healthy skin
Search family involvement with consanguinity
The prognosis ranging from mild to severe, depending on the type of epidermolysis as the single or the slightest junctional which is the most serious. Infection is the most common complication and causing death.
The tto. is prevention with genetic counseling, care of the skin and prevent infections and complications.
Surgery of the aftermath of the flanges, scars, mergers fingers preventing the functionality of hand.
The tto rehabilitation is based on improving the arch joints from flanges and scars.
Clinical Case:
54 year old male patient suffering from dystrophic epidermolysis bullosa. He is the first in his family and his children (2 children) unaffected.
Clinic: MMss skin and lower limbs with great fragility that have caused scarring flanges hands with syndactyly of the fingers significantly deformed. (Pictures)
Esophageal stenosis esophageal dilations motivated to improve nutrition.
Currently the patient has limited extension fingers of both hands, fingers deformity, fist fully functional, but has important fragility of both hands and feet (photos)
Make an active life and work desk in school, always keeping minor trauma care to avoid abrasions/ulcerations.