Condition: Epidermolysis bullosa

Inheritance:

Genetically heterogeneous, autosomal dominant and autosomal recessive.

Genetic etiology:

The simplex form is due to mutations in either KRT5 or KRT14, which encode two different forms of keratin proteins. Inheritance of EB simplex usually is autosomal dominant. The dystrophic form includes both dominant and recessively inherited types, due to mutation in the *COL7A1* gene that encodes a subunit of type 7 collagen. Other forms of EB include junctional (genetically heterogeneous, with mutations in various proteins comprising basement membrane), and hemidesmosomal (involving mutation in proteins that anchor keratinocytes to basement membrane).

Frequency:

EB simplex is estimated to affect 1/30,000 – 1/50,000. Dystrophic EB is estimated to affect 1/6.5M live births.

Clinical features:

The epidermolysis bullosa phenotypes involve fragility of the skin, leading to blistering with minor friction or trauma. In the simplex types, the blistering occurs within the basal layer of skin and does not result in scarring. Usually the major involvement is in the hands and feet. In dystrophic EB, blistering occurs below the basement membrane and leads to scarring all over the body.

Management:

Symptomatic care of blisters; avoidance of trauma if possible.

Genetic counseling:

Depends on mode of genetic transmission (autosomal recessive or sex-linked). Genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.