BÖLÜM 8

EPİDERMOLİZİS BÜLLOZADA PEDİATRİK BAKIM [1]

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GİRİŞ

Epidermolizis bülloza (EB), minimal travmalardan sonra deri ve müköz membranların aşınması, kabarması (bül oluşumu) ve dermo-epidermal cilt bütünlüğünün bozulması (yara oluşumu) ile tanımlanan bir grup genetik hastalıktır (1). Bül oluşumuna dermo-epidermal yapıdaki proteinlerin kodlanmasında görev alan genlerde gelişen mutasyonlar sebep olmaktadır (2,3). Epidermolizis bülloza adı, ilk kez Koebner tarafından 1886'da kullanılmıştır. Ancak bu tanı ile uyumlu vakaların başkaları tarafından daha önceden tanımlandığı görülmüştür(4). İnsidans ve prevalansa ilişkin net bir sayı olmamasına karşın, yaklaşık olarak EB prevalansının milyonda 11, insidansının bir milyon canlı doğumda 20 olduğu tahmin edilmektedir(5,6). Türkiye'de ise kayıtlara geçen yaklaşık 700 civarı hasta bulunmaktadır (7).

Pearson 1962'de elektron mikroskopik yöntemler kullanarak EB'yi üç büyük fenotipik grup olarak sınıflamıştır. Bunlar sırasıyla EB simpleks (EBS), junctional EB (JEB) ve distrofik EB (DEB)'dir

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