

Bölüm 8

NEONATAL DİYABET

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GİRİŞ

Neonatal diyabetes mellitus (NDM), yaşamın ilk altı ayı içinde, bozulmuş insülin fonksiyonu nedeniyle hipergliseminin olduğu, çoğunlukla pankreas beta hücrelerinin işlevsel veya hücresel fonksiyonunu etkileyen tek gende meydana gelen mutasyonlardan kaynaklanan bir hastalıktır (1,2). NDM, infant dönemde mevcut olan monojenik diyabet formlarını tanımlamak için kullanılan terimdir. Bazı hastalar posnatal ilk 30 gün içinde tanı alsalarda, çoğunlukla yaşamın ilk altı ayında, bazen de 12. aya kadar tanı yaşı uzayabilir (3,4). NDM'si olan bebekler asemptomatik hiperglisemi veya klinik olarak dehidratasyon, intrauterin büyüme geriliği, kilo alamama, glukozüri, ketoasidoz ve ozmotik diürez bulguları ile başvurabilirler. İnsüline bağımlı hiperglisemili bebeklerde NDM, diğer nedenler dışlandıktan sonra kan glukoz düzeyleri 250 mg/dL'den daha yüksek seyrettiği (yedi ila on günden daha uzun süre devam eden) durumlarda düşünülmelidir (4,5). Polijenik otoimmün Tip 1 DM yaşamın ilk altı ayında ortaya çıkması beklenen bir durum değildir. Bu nedenle literatürde ve klinik olarak NDM terimi, monojenik DM ile başvuran yaşamın ilk bir yılında olan hastalar için kullanılmaktadır (1).

EPİDEMİYOLOJİ

Neonatal diyabetes mellitus (NDM), infantil hipergliseminin nadir bir nedendir ve insidansı yaklaşık olarak 90.000-160.000 canlı doğumda bir olarak bildirilmiştir (4,6). Akraba evliliğinin fazla olduğu ülkemizden yapılan bir çalışmada bu oran 30.000 doğumda bir olarak belirtilmiştir (7). Yaşamın ilk altı ayında DM ile başvuran bebeklerin çoğunda (%80-85) monojenik bir neden olmaktadır (3,8). NDM fenotipik özelliklerle geçici (%45), kalıcı (%45) ve sendromik (%10) formlara ayrılabilir (3,9). NDM genetik olarak heterojendir, bilinen 22 genetik nedeni vardır (21 gendeki mutasyonlar ve 6q24 lokusundaki metilasyon bozuklukları),

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