

Bölüm 7

OTOZOMAL DOMİNANT POLİKİSTİK BÖBREK HASTALIĞI VE HİPERTANSİYON

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Otozomal dominant polikistik böbrek hastalığı (ADPKBH) böbrek yetmezliğine neden olan genetik hastalıklar arasında en sık görülenidir. Ortalama olarak 400-1000 canlı doğumdan birinde görüldüğü bildirilmiştir (1). Böbreklere ek olarak karaciğerde ve pankreas gibi diğer organlarda da kistler görüldüğü için multi-sistemik bir hastalık olarak değerlendirilmektedir. Ayrıca hastalarda intrakranial arteriyel anevrizma ve vasküler diseksiyon sıklığında artış mevcuttur. Kistlerin çok sayıda olması ve progresif olarak büyümesine bağlı gelişen kronik böbrek hastalığı ve erken yaşlarda ortaya çıkan hipertansiyon hastalığının en önemli klinik bulgularıdır.

EPİDEMİYOLOJİ VE GENETİK ÖZELLİKLER

ADPKBH tahmini sıklığı 1/400-1/1000 aralığında değişmektedir. Klinik olarak sıklıkla sessiz seyretmekte, vakaların yarısından daha azı tanı alabilmektedir. Hastalığın görülme sıklığının cinsiyet ile ilişkisi yoktur. Hastaların %6-8' inde son dönem böbrek hastalığı gelişmektedir.

ADPKBH patogenezinde böbrek tubulus epiteli, safra kanalları, pankreas kanalları ve serebral damarlarda gösterilen polikistin-1 ve yapısı kalsiyum kanallarına benzeyen polikistin-2 membran proteinlerinin sentezinde rol oynayan PKD-1(kromozom 16) ve daha hafif formda hastalığa neden olan PKD-2 (kromozom 4) genlerindeki mutasyonlar suçlanmaktadır (2). Hastaların %78'inde PKD-1 mutasyonu, % 14'ünde PKD-2 mutasyonu tespit edilmiştir. Yakın dönemde son dönem böbrek hastalığına ilerlemeyen hastalığın hafif formu ile ilişkili GANAB

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oranı arasında anlamlı bir farklılık gösterilememiştir(32). Yine enalapril ve atenolol ile yapılan bir diğer çalışmada da her iki grupta da böbrek fonksiyonları ve mikroalbuminüri düzeyleri benzer bulunmuştur (33)

Sonuç olarak; ADPKBH en sık görülen kalıtsal böbrek hastalığıdır. Bu hastalar erken yaşlarda hipertansiyon gelişimi açısından dolayı yüksek risklidir. ADPKBH'da en önemli mortalite nedeni olan kardiyovasküler hastalıklar ile hipertansiyon arasında yakın bir ilişki mevcuttur. Hasta ilk tespit edildiği andan itibaren hipertansiyon yönünden değerlendirilmelidir. Bu hastalara ADPKBH ve hipertansiyon hakkında eğitim verilmesi, diyet ve yaşam tarzı değişiklikleri açısından teşvik edilmesi ve hipertansiyon tedavisinin erken dönemde başlanması ileri dönemde ortaya çıkabilecek komplikasyonları engellemek açısından önemlidir.

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