BÖLÜM 16

Hipertrofik Kardiyomiyopati

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GİRİŞ

Hipertrofik kardiyomiyopati (HKMP), hipertansiyon ve kapak hastalığı gibi sekonder nedenlerin yokluğunda, özellikle sol ventrikül (LV) (duvar kalınlığı ≥ 15 mm) olmak üzere kardiyak hipertrofi ile karakterizedir. Özellikle sarkomer proteinlerini kodlayan çok sayıda gendeki mutasyonlar nedeniyle en yaygın kalıtsal kardiyomiyopatidir. genç atletlerde ani kardiyak ölümün bir numaralı sebebidir. HKMP ile ilişkili en önemli anormallik, hipertansiyon veya aort stenozu gibi bir sebep yokken ortaya çıkan sol ventriküler hipertrofidir. En yaygın gözlemlenen patern anterior interventriküler septumda asimetrik kalınlaşmadır (asimetrik septal hipertrofi). Bu patern, klasik olarak, mitral kapak sistolik anterior hareketi ve dinamik sol ventrikül çıkış obstrüksiyonu ile ilişkilidir. Buna rağmen, vakaların çoğunda (%75) hipertrofik kardiyomiyopati sol ventrikül çıkış obstrüksiyonu ile ilişkili değildir ve bu sebepten isim Hipertrofik Obstruktif Kardiomiyopati'den HKMP'ye değiştirilmiştir. Ayrıca 1:500 oranında görülür ve genellikle otozomal dominant geçiş gösterir (1).

ETİYOLOJİ

HKMP"nin ailesel formu, genetik geçiş gözlenen otozomal dominant hastalıktır. Çocuk ve erişkinlerde HKMP'ye sebep olacak birçok hastalık vardır (Tablo 1). Bunlara örenek olarak Noonan sendromu, Pompe hastalığı, diğer metabolik hastalıklar, Danon hastalığı verilebilir.

Kardiyomiyopatiler içinde en sık genetik geçiş gösteren HKMP'dir. Ayrıca aile öyküsü olmayan bireylerde De Novo mutasyona bağlı olarak da gözlemle-

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