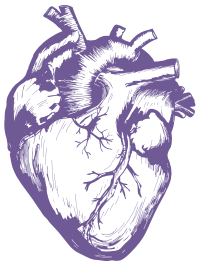

BÖLÜM 72

Kardiyak Amiloidoz

Tahir Alper CİNLİ [1]

GİRİŞ

Sistemik amiloidozlar, çeşitli doku ve organlarda proteinlerin yanlış katlanması sonucunda oluşan beta amiloid fibrinlerinin çökerek biriktiği geniş bir hastalık spektrumudur. Kardiyak amiloidozda, amiloid fibrinleri kardiyak miyositler arasındaki intertisyel alanda birikerek hücresel hasarına ve ileti bozukluğuna neden olur. İleri aşamada biriken amiloid fibrinleri restriktif kardiyomiyopati (RKM)'ye neden olur.

Sistemik amiloidozların adlandırması, amilodiozun "A" harfi ile başlar ve sonrasında yanlış katlanan proteinin baş harfi eklenerek kısaltmadan oluşturulur. Amiloid kardiyomiyopati (CM) vakalarının büyük çoğunluğunu, kemik iliği plazma hücre bozukluklarında üretilen monoklonal immünglobulin hafif zinciri (LC) ve karaciğer, koroid pleksus, retinal pigmente epiteli tarafından üretilen transtiretin (TTR) olarak bilinen taşıma proteini oluşturur. Kardiyak amiloidozun nadir nedenleri arasında serum amiloid A amiloidozu (AA), kalıtsal apolipoprotein A-1 amiloidozu bulunur(1).

Yaşlılıkta TTR yanlış katlanması ve birikimi artış gösterir. TTR' nin yanlış katlanması ve birikimi (ATTR CM), genetik olarak normal (wild type, vahşi tip) protein bağlamında gerçekleşir. Vahşi tip TTR amiloid kardiyomiyopatisi (ATTRwtCM) olarak adlandırılır. TTR'nin yerine geçme veya delesyon mutasyonları sonucu meydana gelen amiloid kardiyopatisi varyant transtiretin amiloid kardiyomiyopatisi (ATTRvCM) olarak adlandırılır (1).

ATTR CM' li hastaların en büyük problemi hastaların geç tanı almasıdır. Hastalar görünüşte birbiri ile alakasız kas-iskelet, nörolojik, gastrointestinal (GIS), renal bulguların göstermesi ve bu bulguların hekimler tarafından tek bir hastalıkla ilişkilendirilememesi sonucu tanı gecikir.

Kardiyak amiloidozda kardiyak belirtiler spesifik değildir. Artmış sol ventrikül duvar kalınlığı kardiyak amiloidoz ile birlikte hipertansif kalp hastalığı, aort stenozu, kosantrik hipertrofi, hipertrofik CM veya Fabry hastalığı gibi diğer infiltratif CM'lerde de görülür.

Özellikle monoklonal hafif zincir testi sonuçlarının yorumlanması, kardiyak sintigrafi taramalarının yapılması ve yorumlanması, biyopsi ve genetik testlerin endikasyonları ve yorumlanması konusunda çoklu disiplinli yaklaşım gerektirir.

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CM için, ekstra kardiyak etkilenme olan GIS tutulumu ve otonomik nöropati kontrendikasyon oluşturur. GIS tutulumu enfeksiyon riski, yara iyileşmesinde kötüleşme ve malnütrisyona neden olabilir. Nöropati, kalp nakli sonrasında düzelmez. TTR susturma tedavisinin ortaya çıkmasıyla birlikte ATTRvCM' li hastalarda kalp nakli sonrasında bir rolü olabilir(57).

Kalp nakli için %75'in altında veya 10 yılda %50'nin altında bir sağ-kalım oranı olan durum kontrendikasyon olarak kabul edilir. Sistemik AL amiloidozda kalp nakli sonrası sağ kalım yaklaşık olarak %75' tir ve 5 yılda %50'dir. Sağkalım yüksek riskli sitogenetik bulguların varlığında daha da azalır. Kalp nakli sonrası hafif zincirlerin derin ve uzun süreli baskılanmasının gereklidir bu yüzden kalp nakli ardından OKİT uygun hastalarda akılda bulundurulmalıdır(58).

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