

Bölüm 28

SİSTEMİK MASTOSİTOZ VE KARDİOVASKÜLER BULGULAR

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GİRİŞ

Mastositoz; deri, kemik iliği ve iç organlarda anormal mast hücrelerin (MH) birikimi ile tanımlanan heterojen bir hematolojik neoplazm grubudur.

Literatürde mastositoz ile ilişkili ilk olgu raporu 1869 yılında Nettleship tarafınca kutanöz mastositoz olgusu olarak yayımlanmıştır (1). Sistemik mastositoz (SM) ise ilk defa 1946 yılında Ellis tarafından otopsi preparatlarında organ infiltrasyonunun gösterilmesi ile tanımlanmıştır (2). 1987 yılında Schwartz ve ark. triptazı biyomarker olarak göstermiş (3), 1993 yılında Furitsu ve ark. MH'de KIT D816V mutasyonunu saptamış (4), Escribano ve Orfao MC'lerin anormal immünofenotiplerini tanımlamışlardır (5).

Danimarka'da yapılan bir çalışmada prevalansı 1/10.000 olarak saptanmıştır (6). Erkek ve kadınların eşit oranlarda etkilendiği hastalığın net insidansı bilinmemektedir. Çocukluk çağında cilt mastositozu (CM) yaygın görülmekteyken, erişkin yaşlarda sistemik formu daha sık görülmektedir (7).

Mastositozlu olgularda farklı klinik bulgular görülür. Bu bulgu çeşitliliği, MH mediyatörlerinin kronik ve/veya epizodik salınımına ek olarak, bir veya daha fazla dokuda aşırı MH birikiminden kaynaklanır (8). Böylece zaten nadir olan bu hastalığın tanınması güçleşir.

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D816V KIT mutasyonuna karşı aktiviteye sahip bir multikinaz inhibitörü olan midostaurin FDA tarafından SM tedavisi için onaylanmıştır(12, 20). Başka bir tirozin kinaz inhibitörü olan imatinib D816V mutasyonu dışındaki mutasyonlar ve eozinofilik lösemi varyantında (PDGFR mutasyonu) etkilidir (12).

SM'da salınan mast hücre mediyatörlerine bağlı kardiyovasküler semptomlar siktir. Bir semptom, birkaç farklı mast hücre aracısından kaynaklanabilir. Farklı birçok sisteme ait bulgu verebilen sistemik mastositozun kardiyak tutulumu yaşamı tehdit edici olabilir ve akla getirilmezse tanı koymak zordur. Kardiyovasküler açıdan; tekrarlayan açıklanamayan senkop epizodları veya derin hipotansiyon durumlarında, açıklanamayan aritmi ve kalp yetmezliklerinde akla mastositoz getirilmelidir.

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