Bölüm
19

GAUCHER HASTALIĞI

Elif BAYRAKTAR[21]

GİRİŞ

Splenomegali, daima önemli bir bulgu olarak ele alınmalı ve mutlaka incelenmelidir. Splenomegali yapan major nedenleri konjestif, malignite, infeksiyon, inflamasyon, hematolojik ve malign olmayan infiltratif olmak üzere 6 grupta toplayabiliriz. Gaucher hastalığı, neimann-pick, mukopolisakkaridozlar gibi lizozomal depo hastalıkları malign olmayan infiltratif grupta yer alır (1).

Metabolizmal hastalıklarda klinik tablo farklılıklar gösterebilmektedir. Spektrumun bir ucunda çok hafif bulgularla seyreden hastalar bulunurken, diğer ucunda masif organomegali ile birlikte ağrılı kemik krizleri ile yaşam kalitesi düşen hastalar yer alabilmektedir. Bazen de açıklanamayan splenomegali, metabolik hastalıklarda hastalığın tanısını koymada çıkış noktası olabilir. Nitekim gaucher hastalığını 1882 yılında ilk kez tanımlayan Phillippe Ernest Gaucher de, açıklanamayan masif splenomegalisi olan 32 yaşında bir kadın hasta tariflemiş ve otopsi sırasında dalakta gözlenen büyük ve anormal hücreler saptamıştır (2).

Son yıllarda geliştirilen yeni tedavi yöntemleri, metabolik hastalıkların bazılarının kaderini değiştirmiş ve yaşam süresini artırmıştır. Eskiden pediatristlerin görebildiği bu hastalıklarla iç hastalıkları uzmanları da karşılaşmaya başlamıştır. Bu bölümde yetişkinlerde ortaya çıkan lizozomal depo hastalığı gaucher hastalığından bahsedilmiştir.

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