BÖLÜM 21

ALVEOLAR SOFT PART SARKOMLAR VE PERİFERİK SİNİR KILIFI TÜMÖRLERİ

Seval AY ERSOY[1]

GİRİŞ

Alveolar soft part sarkomlar (ASPS) genelde yumuşak doku sarkomların %1'inden azında görülen ve genellikle erişkinlerde alt ekstremite yumuşak dokularında, çocuklarda baş ve boyun bölgesinde ortaya çıkan nadir bir neoplazmadır. Öncelikle yavaş büyüyen bir kitle olarak ortaya çıkar, teşhisi zor olabilir. Genellikle erişkinlerde tanı anında metastatik hastalık olarak ortaya çıkar. Spesifik bir kromozomal alterasyon olan der(17)t(X:17)(p11:q25) ile karakterizedir. Bu kromozomal alterasyon transkripsiyon faktör E3 (TFE3) ile alveolar soft part sarkom kritik bölge 1(ASPSCR1) füzyonu sonucu oluşur. Bu translokasyon tanısal olarak önemli ve spesifiktir çünkü tümör çekirdekleri immünohistokimya boyama ile TFE3 için pozitiftir.

Periferik sinir kılıfı tümörleri (PSKT) periferik sinirlerden kaynaklanan ve sarkomların yaklaşık %4'ünü oluşturan tümörlerdir. Ana risk faktörlerinden birisi nörofibromatozis tip I (NF1) genetik hastalığı ile birlikteliği olmasına rağmen PSKT vakalarının yaklaşık %45'i sporadik olarak ortaya çıkar. Sporadik vakalarda tanımlanan en önemli risk faktör ise radyasyona maruziyettir. Her iki cinsiyette eşit sıklıkta olarak saptanmakla beraber genelde otuzlu yaşlarda pik yapma eğiliminde olmaktadır. Duysal sinirler ön planda etkilenir ve hastalarda en önemli semptom paraparezi yani ilgili uzuvlarda yorgunluk, uyuşukluk ve hareket güçlüğü ile karakterizedir.

ALVEOLAR SOFT PART SARKOMLAR

Epidemiyoloji ve İnsidans

ASPS'lar tüm yumuşak doku tümörlerinin ortalama %1'inden azında görülen bir alt tiptir (1). ASPS'ler genç erişkinlerde genelde 15-30 yaş arasında daha sık tanı

[1] Uzm. Dr., Balıkesir Atatürk Şehir Hastanesi, Tıbbi Onkoloji Kliniği, drsevalay@gmail.com

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