11. BÖLÜM

MİT AİLESİ TRANSLOKASYON RENAL HÜCRELİ KARSİNOM

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MiTF (Mikroftalmi assosiye transkripsiyon faktörü) ailesi translokasyon RCC'leri TFE3 ve TFEB genlerinin farklı partnerlerle füzyonu ve onkogenik aktivasyonu sonucu ile gelişen neoplazilerdir (1). Dünya Sağlık Örgütü tarafından 2004 yılında Xp11 translokasyonlu RCC'ler (t-RCC) translokasyon karsinomları kategorisi altında tanımlanmıştır (2). 2016 yılında ise literatürde tanımlanan TFEB translokasyon karsinomları da bu gruba dahil edilerek "MiT ailesi translokasyon renal hücreli karsinomları" grubu altında Dünya Sağlık Örgütü sınıflandırmasına dahil edilmiştir (3,4).

TFE3 geninin ASPSCR1(ASPL), PRCC, NONO (p54nrb), SFPQ (PSF), CLTC gibi farklı partner genlerle füzyonu sonucu Xp11 translokasyonlu RCC gelişirken; TFEB geninin MALAT1 geni ile füzyonu sonucu t(6;11) RCC gelişir (4).

Etyolojide sitotoksik kemoterapi maruziyetinin rolü olabileceği bildirilmiştir (5).

Çocukluk çağında izlenen RCC'lerin yaklaşık %40'ı, Xp11 translokasyonlu RCC iken erişkin çağda izlenen RCC'lerin %1,6-4'ü, Xp11 translokasyonlu RCC'dir (6). t(6,11) RCC'ler ise Xp11 translokasyonlu RCC'lerden çok daha az sıklıktadır. Literatürde yaklaşık 70 vaka bildirilmiştir. Ortalama görülme yaşı 31'dir (7-9).

MiT ailesi translokasyonlu RCC'lerin kendilerine özgü ayırıcı bir makroskopik görünümü yoktur (4).

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t(6;11) RCC'ler ise çok nadir görüldüğünden klinik davranışı henüz net değildir. Cerrahi tedavi önceliğini korumaktadır. Adjuvan kemoterapinin etkinliği henüz tartışmalıdır. Bu grup karsinomlarda da VEGF reseptörleri tedavide denenmektedir (50,55). Xp11 t-RCC'ler gibi uzun yıllar sonra metastaz ve rekürrens yapabildiği için uzun dönem takip önerilmektedir (9).

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