44.
BÖLÜM

MULTİPL ENDOKRİN NEOPLAZİ TİP2B (TİP 3) VE FAMİLYAL MEDÜLLER TİROİD KANSERİ

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GİRİŞ

Multipl endokrin neoplazi tip 2 (MEN2) genel popülasyonda otuz bin de bir görülen otozomal dominant olarak kalıtılan, nadir bir kanser sendromudur. MEN2'de genel olarak medüller tiroid kanseri, feokromasitoma ve paratiroid hiperplazi görülür.

MEN2B, MEN2A ve familyal medüller tiroid kanserine 10. kromozom üzerinde taşınan RET proto-onkogeninde ki mutasyon neden olur .(1,2) MEN2B görülen hastalar tüm MEN 2 vakalarının %6'sı civarındadır.(3) MEN2B ve MEN2A aynı genlerle kalıtılır. Otozomal dominant sendromlardır. MEN2B'de medüller tiroid kanseri ve feokromasitoma MEN2A'ya benzer olarak görülür. Ancak paratiroid hiperplazisi MEN2B'de görülmez. Bunlara ek olarakta önemli klinik farklılıkları vardır. MEN2B'li hastalarda, tipik olarak dudakları ve dili tutan mukozal nöromalar ve bağırsak ganglionöromaları görülür. Kronik kabızlık ve megakolon gibi kolon fonksiyon bozuklukları yaygındır. Bu hastaların çoğunda gelişim anormallikleri, Marfanoid görünüm ve miyelinli bu nedenle kalınlaşmış kornea sinirleri vardır.

MEN2B'li hastaların % 95'inde bulunan en yaygın mutasyon RET proto-onkogeninin tirozin kinaz domaininde 918. kodonda ki metionin ile threonin yer değiştirmesidir.(M918T) Geri kalan hastalar ise genellikle RET A833F mutasyonu taşırlar.(4,5,6) MEN2B'li hastalarda mutasyonların % 90'ından fazlasının de novo olarak ortaya çıktığı tahmin edilmektedir.(2,7)

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profilaktik total tiroidektomiye verilebilir.(40) Özellikle M918T mutasyonu taşıyanlarda erken yaşlarda profilaktik total tiroidektomi önerilmektedir.(8) MEN2B ilişkili tümörler özellikle agresif seyirlidir. 10 yaşından önce metastatik hale gelebilirler bu nedenle MEN2B ilişkili RET mutasyonu taşıyıcıları feokromasitoma ve MTK açısından yakın takip edilmelidir.(40)

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