42.
BÖLÜM

MULTİPLE ENDOKRİN NEOPLAZİ TİP 1

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GİRİŞ

Multiple Endokrin Neoplazi Tip 1 (MEN1) nadir görülen otozomal dominant kalıtsal bir sendromdur. Klasik olarak paratiroid bezi, ön hipofiz bezi ve pankreas adacık hücre adenomlarına (3P; parathyroid, pituitary, pancreas) yatkınlık ile karakterizedir. Klinik olarak bir hastada iki veya daha fazla majör MEN1 tümör tipinin bir arada bulunması veya MEN1 tanısı olan bir hastanın aile bireylerinde MEN1 ilişkili tümör görülmesi ile tanı konulur (1,2). Gastrointestinal ve adrenokortikal adenomlar, timik ve bronşiyal karsinodiler, benign cilt ve santral sinir sistemi tümörleri eşlik edebilir. Paratiroid bezi adenomları %90, enteropankreatik tümörler %30-70, ön hipofiz bezi adenomları %30-40, cilt kollajenomları ve anjiofibromları %60-65 oranında görülür (2,3). Populasyonda görülme sıklığı 2/100 000 olup, kadın erkek dağılımı benzerdir ve beş yaş sonrası bütün yaş gruplarında görülebilir (2).

GENETİK

Klasik MEN1, 11. kromozom uzun kolunda yer alan *MEN1* geninde (11q13) veya bu geni etkileyen diğer bölgelerde oluşmuş germline mutasyonlar sonucunda gelişir. *MEN1* geni tümör baskılayıcı bir gen olup protein ürünü "menin" olarak adlandırılır (4,5). Hastaların yaklaşık %80-90'nında *MEN1* geni germline mutasyonları tespit edilebilir. Geri kalan kısmında gen delesyonları, promoter bölge veya

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