Bölüm 19

ENDOMETRİOZİS VE GENETİK

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Aile ağacı çalışmaları ve ikiz çalışmaları endometriozisin genetik bir alt yapısının olduğunu ortaya çıkarmıştır. Yapılan çalışmalarda endometriozisin poligenik bir kalıtım gösterdiği, birinci derece akrabalarda %4-9 oranında bulunduğu bildirilmiştir. Klinik gözlemlerde endometriozis gelişimi ile ilişkili genler ile kısırlığa neden olan genlerin bağlantılı olabileceğini düşünülmüştür. Otöpik endometrium ile normal endometrium gerek peritoneal endometriozisli olgularda, gerekse endometriomalı hastalarda farklı reseptif gen haritalarına sahiptir. Endometriozis/endometrioma cerrahisi ötopik endometrium resptif gen dizilimine olumlu yönde katkıda bulunarak implantasyon oranlarını artırabilir. **Editorial**

Giriş

Endometriozis genellikle disparoni, dismenore, pelvik ağrı ve infertilite gibi jinekolojik sorunlara yol açan endometrial dokunun uterus dışında bulunması olarak tanımlanan bir hastalıktır (1). Üreme çağındaki kadınlarda yapılan prevalans çalışmaları hastalığın %10-15 kadar yaygın olduğunu hatta infertilite nedeniyle araştırılan kadınlarda bu sıklığın %30 lara kadar yükseldiğini bildiren yayınlar mevcuttur (2-10). Aslında endometriozis için "muamma" kelimesinin kullanılması bu hastalık hakkında ne denli fazla bilinmeyen olduğunu göstermektedir. Keza bir hastalık olduğu konusunda dahi tartışmalar vardır ki bazı görüşlere göre adenomyozis ayrı bir klinik antite, uterus dışı bulunan pelvik peritoneal yüzeylerdeki yüzeyel lezyonlar, over içerisinde bulunan endometrioma ve derin endometriozis de ayrı birer hastalık olarak olarak düşünülmektedir. Bunun nedeni etyopatogenezin net ayrıştırılmamamsından kaynaklanmaktadır. Belki de bu hastalık faklı etyopatogenez temelli değişik temel etyolojilerin ortak klinik yansımasıdır (11-24). Sık görülen bu jinekolojik durum için genetik orjinli olduğunu destekleyen kanıtlara rağmen, temel etiyoloji ve endometriozis patogenezi henüz tam olarak aydınlatılamıştır. Endometriotik lezyonların heterozigosite kaybı olduğunu inceleyen moleküler DNA çalışmaları endometrioid tip over kanserleri için endometriozis odaklarının malign dönüşümünde rol alan 5q, 6q, 9p, 11q ve 22q dahil aday tümör baskılayıcı gen lokusları tanımlamıştır (25-36). Epitelyal over kanserinde rol alan tümör baskılayıcı PTEN genindeki mutasyonların benign endometriotik hücrelerinde de görülmesi bu noktada diğer bir kanıttır. Genetik faktörler de endometriozis için bireysel yatkınlık olasılığı da vardır. Allelik farklılıkların kalıtsal olduğuna dair henüz kanıt yoktur. Aile bireyleri arasındaki çalışmalar ve ikiz çalışmaları akrabalarda endometriozis sıklığının daha fazla olduğunu göstermiştir (37-49). Çevresel faktörler ve genetik faktörlerin araştırıldığı çalışmalarda ise her ikisininde endometriozis etyopatogenezinde rol aldığı ve aralarında etkileşim olduğu ortaya konulmuştur. Son yıllarda yapılan çalışmalar ile endometriozis gelişiminde rol oynayan yeni aday genlerin de ortaya konması

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