Bölüm
2

ENDOMETRİOZİS ETKENİ OLARAK MOLEKÜLER BİYOLOJİ VE GENETİK POLİMORFİZM

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Endometriozis endometrium gland ve stromasının rahim dışında bulunması olarak tanımlanmaktadır. Genelde kronik pelvik ağrı, dismenore, disparoni ve infertiliteye neden olmaktadır. Benign, inflamatuar bir hastalıktır ve östrojen bağımlıdır. Tanıda altın standart laparaskopi ve histopatolojik tanıdır.

Hastalığın prevalansı değişmektedir. Çünkü hastaların bir kısmı asemptomatik olabilir. Ayrıca çalışılan populasyona göre hastalık sıklığı değişmektedir. Asemptomatik tubal ligasyon nedeniyle laparoskopi yapılan hastalarda %1-7 arasında sıklık görülmüştür (1). Cerrahi yapılan hastalarda ise endometriozis nedeniyle cerrahi yapılan hastaların %57 sinde ,pelvik ağrı nedeniyle cerrahi olan hastaların %21' inde ve pelvik ağrı ya da endometriozis ön tanısı olmadan cerrahi yapılan hastaların %8'inde endometriozis mevcuttur (2). Semptomatik hastalarda sıklığı daha fazladır, pelvik ağrı olan adolesanlarda sıklık %70 iken (3),infertil hastalarda %50 (4) sıklıkta görülmektedir.

Hastalığın cerrahi olarak evrelemesi American Society for Reproductive Medicine evreleme sistemine (5) göre yapılmaktadır.

Evre 1: Hastalık izole implantlarla sınırlıdır. Adezyon yoktur.

Evre2: 5 cm altında süperfisyel implantlar vardır. Adezyon yoktur.

Evre 3: Multipl süperfisyel ve derin implantlar vardır. Peritubal ve periovaryen adezyonlar vardır.

Evre 4: Multipl süperfisyel ve derin implantlar vardır. Büyük ovaryen endometriomalar vardır. Film ve dens yapışıklıklar mevcuttur.

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Gen polimorfizmi toplumdan topluma farklılık göstermektedir. Bu nedenle çalışmalara aynı ırksal özellikteki hastaların alınması gerekmektedir.

Yine son zamanlarda yapılan bir çalışmada endometriozis dokularında somatik mutasyon varlığına bakılmıştır. %26 oranında mutasyon saptanmıştır. Bunlar da genellikle kanserle ilişkili mutasyonlardır(48).

Sonuç olarak endometriozis kalıtsal olan ama komplike bir hastalıktır. 8 gen bölgesi tanımlanmıştır ve ileri evre hastalarda daha çok mutasyona rastlanmıştır. Konunun daha net ortaya konabilmesi için daha fazla çalışmaya ihtiyaç duyulmaktadır.

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