

Bölüm 7

POLİKİSTİK OVER SENDROMU VE GENETİK ÇALIŞMALAR

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GİRİŞ

Polikistik over sendromu (PCOS) genetik altyapısı tam olarak çözülememiş, poligenik, multifaktoriyel ve üreme çağındaki kadınlarda en fazla görülen bir endokrinopatidir. İçerisinde klinik/biyokimyasal hiperandrojenizm, dislipidemi, oligo/anovulasyon ve polikistik over morfolojisi gibi kriterlerin incelendiği 3 farklı tanı kriteri ile PCOS tanısı konulmaktadır. Tanı kriteri olarak yer almasa da insülin direnci hastaların büyük bir kısmında bulunmaktadır.

Otozomal dominant kalıtım gösterme potansiyeli diğer mendelyan kalıtım tiplerden daha sık olan PCOS'da hem genetik düzey hem de biyokimyasal markır bulma düzeyinde çalışmalar devam etmektedir. Genetik çalışmalarda kullanılan yöntemler geliştikçe daha fazla sayıda aday gen daha fazla sayıda PCOS'lu hastanın aynı anda çalışılmasını sağlamıştır. Dolayısıyla her geçen gün daha fazla sayıda aday gen PCOS'lu hastalarda ortaya çıkarılmaktadır.

Çalışmamızda başlangıcından son güncel gelişmelere kadar PCOS'dan sorumlu aday genler, aday varyasyon veya mutasyonların hem spesifik popülasyonlarda hem de meta-analiz çalışmalarında PCOS'u ve çeşitli fenotiplerini açıklama potansiyelleri verilmiştir.

Polikistik over morfolojisini gösterebilmek için hala altın standart olarak kullanılan transvajinal ultrason görüntülemenin yerini alabilecek daha az iş/insan/maliyet yükü gerektiren markır bulma çalışmalarında umut vaadedici gelişmeler ortaya çıkarılmaya devam etmektedir.

ETİYOLOJİSİ VE TANI KRİTERLERİ

Polikistik over sendromu (PCOS) ilk kez 1935 yılında Stein ve Leventhal'ın obezite, kıllanma, infertilite ve amenore semptomları ile birlikte, genişleyen

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Son olarak NGS teknolojisini PCOS'lu kadınların mitokondriyal DNA'larında kullanan bir araştırma, hem mitokondriyal DNA kopya sayısını PCOS'lu hastalarda kontrol grubuna daha düşük bulmuş hem de *MT-ATP6* geninde (pThr67Ala) ve *MT-ATP8* geninde (p.Asn46Ser) mutasyonlarını içine alacak şekilde oksidatif fosforilasyon genlerinde 183 farklı varyant ortaya çıkarmıştır (141).

SONUÇ

PCOS, multifaktoriyel, poligenik ve farklı tanı kriterlerine göre tanı konan ve kadınlarda üreme sistemi ile birlikte diğer bazı sistemlerin de etkilendiği metabolik sendrom bulgularının gözlendiği bir endokrinopatidir.

Aday gen bulma çalışmaları hala devam etmektedir. Meta analiz çalışmaları ile birlikte daha homojen hasta gruplarının dahil edildiği NGS çalışmaları hastalık için gerekli olan biyomarkır bulma çabalarını sonuçlandırarak gibi görünse de hala transvajinal ultrason görüntülemenin önüne geçebilmiş değildir.

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