

8. BÖLÜM

DİYABETİK RETİNOPATİ VE GENETİK

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GİRİŞ

Diyabetik retinopati (DR) dünya çapında görme kaybı ve bozukluklarının başlıca nedenidir. Dünya çapında yedinci en yaygın körlük nedeni olarak tahmin edilmektedir. DR'ye birden fazla faktörün katkıda bulunduğu bilinmektedir. Bunlar arasından glisemik maruziyet, hipertansiyon, hiperlipidemi en iyi bilinen değiştirilebilir risk faktörleridir. Bunun yanında vücut kitle indeksi (VKİ)'ninde hastalığın gelişmesinde rol oynadığı kabul edilmiştir. Yapılan çalışmalarda DR'nin gelişiminde önemli oranda genetik duyarlılığın olduğu gösterilmiştir. DR'li olan hastaların aile üyelerinin de DR öyküsü olmayan ailelere göre DR oluşma olasılığı 2-3 kat daha fazladır. Genetik katkının DR için %27 ve proliferatif DR (PDR) için %52 kadar yüksek olduğu hesaplanmıştır. Son on yılda, DR'nin genetik belirleyicilerini tanımlamaya odaklanan çok sayıda araştırma yapılmıştır. Genetik bağlantı analizleri, aday gen çalışmaları, genom çapında ilişkilendirme çalışmaları ve yeni nesil dizileme gibi moleküler yöntemler retinopatinin gelişimine katkıda bulunabilecek genler ve lokuslarla ilgili zengin bilgiler ortaya çıkarmıştır.

Çok sayıda çalışma, genetik faktörlerin bir bireyin DR gelişimi ve proliferatif DR'ye ilerleme riskini belirlemede önemli bir rol oynayabileceğini göstermiştir. Bununla birlikte, DR ile geniş popülasyonlarda tutarlı ve kesin genetik ilişkiler kurulamamıştır. DR ile ilişkileri için çok sayıda gen çalışılmıştır ve bu araştırmaların sonuçları özellikle DR gelişimi ve ilerlemesinde yer alan üç spesifik gene işa-

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