Bölüm
5

AĞRI GENETİĞİ

Duygu ONUR CURA[1]

GİRİŞ

Ağrı, fiziksel, duygusal, davranışsal ve sosyokültürel faktörlerin karmaşık etkileşimleriyle şekillenen bir deneyimdir (1). Birçok hastalığın ana bileşeni olup polikliniк başvurularında karşılaşılan en sık semptomlardan biridir. Araştırmalar, toplumun % 15–50'sinin yaşamının herhangi bir döneminde ağrı deneyimlediğini göstermektedir (2). Ağrıya neden olan uyaranlar, nosiseptörler denilen periferik ağrı reseptörleri tarafından tespit edilir ve bu nosiseptif sinyaller aksonlarla beyine iletilerek ağrı olarak algılanır. Bu bölümde, genetik temelleri bilinen ailesel ağrı sendromları yanı sıra çok sayıda gendeki varyantlarla ilişkilendirilmiş kronik ağrı durumları, temporomandibular eklem hastalıklarının genetik temeli ve epigenetik değişikliklerin ağrıdaki rolü güncel genetik çalışmalar ışığında irdelenmiştir.

AĞRI VE GENETİK

Ağrı, dokusal bütünlüğün zarar görmesine neden olabilecek dış etkenlere karşı doğal bir koruyucu mekanizma olarak görev yaparken ağrının kronik formu, içsel patoloji işareti olarak görülür ve bireylerin işlevselliğini olumsuz yönde etkiler (3). Ağrı algısı, bireyler arasında farklılıklar göstermektedir. Bu farklılıklar cinsiyetler arasında belirgin olduğu gibi ırk ve etnik kökenle de ilişkili olduğu gösterilmiştir (2). Ayrıca çeşitli ağrı durumlarında, genetik olarak tanımlanmış farklı periferik nöron gruplarının ve mekanizmaların rol aldığı bilinmektedir (4).

Genomdaki varyasyonlar, bireyler arasındaki farklılıkların temelini oluşturmaktadır. Bu varyasyonlardan en yaygın olanı tek nükleotid değişiklikleridir (SNP: Single Nucleotide Polimorphism). Genomda belli bir yerdeki bir nükleoti-

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