12.
BÖLÜM

Herediter Retina Hastalıklarında Gen Tedavileri

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GİRİŞ

Herediter retina hastalıkları (HRH) retina hücre dejenerasyonuna bağlı görme kaybı ile karakterize oküler hastalıklar grubudur. HRH insidansı dünya çapında 1/2000-3000' dir (1,2).

HRH hasarlı retinal tabakaya, hastalığın seyrine (durağan veya progresif) ve kalıtım paternine (ailesel, sporadik) göre birçok alt gruba ayrılır (Tablo 1). HRH'nin %65'i fotoreseptör hasarı ile karakterize iken, diğer %28'i maküler distrofilerdir (3). Buna ek olarak, HRH bazı sendromların ek bulguları olarak da karşımıza çıkabilir (4).

HRH'nin başlangıç yaşı ve klinik karakterizasyonu bu hastalık grubunun tanımlanmasında önemli bir dayanak noktası iken, günümüzde moleküler genetik analizler rehberliğinde yapılan sınıflandırmalar önem kazanmaktadır. Genetik test seçenekleri: tek gen testlerini, birçok HRH ile ilişkili gen panellerini, tüm ekzom sekanslama (WES) ile tüm genom sekanslamayı (WGS) kapsamaktadır (5). Bu analizlerle, 260'dan fazla genin HRH'ye (RPE65, LRAT, MERTK, MYO7A, ABCA4, CNGA3, CNGB3, PDE6B, REB1, SPATA7 ve TULP1 vs.) neden olduğu tespit edilmiştir (5,6). Böylelikle, başlangıç klinik bulguları benzer olabilen ancak görme potansiyelleri açısından birbirinden çok farklı prognoza sahip olan LCA, akromatopsi, albinizm ve konjenital durağan gece körlüğü gibi HRH alt grupları genetik analizlerle birbirinden ayırt edilebilmektedir.

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