Bölüm 2

KANSER TANI VE TEDAVİSİNDE LİKİT BİYOPSİ

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Günümüzde sağlık hizmetlerinde 'Genomik Tıp' kavramında sadece bireysel farklılıkları göz önünde bulunduran kişiselleştirilmiş tıptan değil, aynı zamanda da toplumun karakteristik özelliklerini dikkate alan hassaslaştırılmış/kesinleştirilmiş tıptan bahsedilmelidir. Burada bireyden bireye geniş ölçüde farklılıklar gösteren, ölçülebilen, klinik olarak değerlendirilebilen özelliklere göre sağlık hizmetinin özelleştirilmesi ile hastalıklarda etkin ve kişiye özel tedavi seçeneklerinin sunulabilmesi hedeflenmektedir. Hassaslaştırılmış/ Kesinleştirilmiş tıbbın uygulanabilirliğini sağlayan ise Omikler ve buna bağlı teknojilerdir.

Omiklerin temelini genleri, gen ürünlerini (transkriptler ve proteinler) ve metabolitleri biyobelirteç olarak kullanan bir yaklaşım oluşturmaktadır. Bu yaklaşımda moleküler düzeydeki farklılıkları saptayabilen, güvenilirliği ve verimi gün geçtikçe artan omiks teknolojileri kullanılmaktadır. İyi klinik uygulamaları kapsamında bu teknolojiler, hipotez gerekmeksizin biyolojik sistemlerin işleyişini her aşamada incelemeyi mümkün kılmaktadır.

Medikal onkoloji de bu yeni teknolojilerin kullanımı için önemli bir alanı oluşturmakta olup, hastalıkların patofizyolojilerinin çözümlenmesinde önemlidir. Ancak hastaların sağaltımında, popülasyon-birey etkileşimi gibi gen-çevre etkileşimlerinin de bilinmesi gerekmektedir.

Omiks teknolojileri hücre, doku veya organlardaki molekülleri bütünsel olarak incelemekle birlikte, insan genomunun tümünün sekanslanarak dizisinin belirlenmesi bu teknolojinin gelişmesine ön ayak olmuştur. Kelimelerin sonuna getirilen 'omik' eki, biyolojik sistemlerin bir bütün olarak analizini ifade eder. İn-

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12.*PIK3CA*: PI3K yolağını düzenleyerek hücrenin sağkalımında rol alan lipid kinaz kodlayan onkogenik bir gendir. Endometriyal kanserlerde % 28,4, meme kanserinde % 27,9, safra kesesi kanserinde % 18,7 oranında somatik değişim rapor edilmiştir (Kompier & ark 2010, Cizkova & ark 2012, Kandoth & ark 2013). Ayrıca, Cowden Sendromu tip 5 gibi herediter neoplastik sendromlarda germ-line mutasyonlar bildirilmiştir. Bu gendeki değişimler, evre I-III kolerektal kanser hastalarında düşük sağkalım oranı ile ilişkilendirilmiştir (De Roock & ark 2011).

Bütün bu bilgiler ışığında, özellikle kanserde oldukça umut vaat eden likit biyopsi ve yeni nesil dizileme yöntemlerinin birlikte kullanımı genetik tanı laboratuvarları açısından büyük önem arz etmektedir. Bu sayede geniş kapsamlı ve hassas bir çalışma ile etkinliği yüksek, bireye özgü sağlık hizmetinin kanser hastalarına verilebilmesi mümkün olabilecektir.

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