2. Bölüm

VENÖZ TROMBOZLARDA GENETİK RİSK

Nehir SELÇUK[1]

Venöz tromboembolizm (VTE), çocuklukta yıllık insidansı 100.000'de 1 olan ve yaşlılıkta yaklaşık 100'de 1'e yükselen yaygın bir hastalıktır. En sık görülen klinik bulgular, bacağın derin ven trombozu (DVT) ve pulmoner embolidir (PE). Nadiren başka yerlerde (üst ekstremite, karaciğer, serebral sinüs, retina ve mezenter) trombüs oluşumu meydana gelir. VTE'nin başlıca sonuçları ölüm, rekürrens, post-trombotik sendrom ve antikoagülan tedaviye bağlı majör kanamadır. VTE, birden çok çevresel ve genetik risk faktörünü içeren çok nedenli bir hastalıktır. (1) Aile ve ikiz çalışmaları VTE' nin kalıtsallığını yaklaşık% 50-60 (2 - 4) olarak tahmin etmiş ve bu da genetiğin önemli bir rol oynadığını göstermektedir.

Venöz tromboembolizm (VTE), genellikle kalıtsal ve edinilmiş risk faktörlerinin müdahalesinin sonucu olan çok faktörlü bir hastalıktır.

Genetik risk faktörleri hiperkoagülabiliteye (kalıtsal trombofili) yatkınlık yaratır ve özellikle 50 yaş altında görülen VTE'de en önemli etiyopatojenik rolü oynar. (5)

[1] Op. Dr., Siyami Ersek Eğitim ve Araştırma Hastanesi, Kalp Damar Cerrahisi Kliniği, nehirtandogar@gmail.com

dan bile tespit edilmemiştir. [80,81] Birkaç sözde yeni nesil dizileme (NGS) veya Yüksek Verimli Nükleotid Dizileme platformlarının geliştirilmesi, [87] hızlı, ucuz ve doğru genomik bilgi üretme olasılığını sunmaktadır . Bu tür birkaç çalışma yayınlanmıştır. [88-97] Önemli bir problem, incelenen mutasyonların nadir olması ve genlerdeki mutasyon zenginleştirmesi üzerinde çalışılsa bile, büyük bir çalışma boyutunun gerekli olmasıdır.

Sonuçta VTE karmaşık bir hastalıktır .VTE'nin genetik patogenezinde sadece pıhtılaşma ve antikoagülasyon değil, aynı zamanda trombositler, eritrositler ve bağışıklık sistemi ve VTE'ye bilinmeyen bağlantısı olan bir dizi gen de rol oynar. GRS, risk değerlendirmesi için yararlı araçlar olacaktır. Umarım, daha büyük GWAS ve farklı çalışmalar VTE'nin kalan kalıtsallığını çözecektir.

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