

BÖLÜM 10

SANTRAL SİNİR SİSTEMİ VASKÜLER MALFORMASYONLARI

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GİRİŞ

Santral sinir sistemi (SSS) vasküler malformasyonları; sporadik olabilir veya bir genetik sendromun bileşeni olarak bulunabilir. Yüksek derecede morbidite-mortaliteye sebep olabilmekte ve cerrahi veya endovasküler girişim gerekebilmektedir (1). Uygun görüntüleme çalışmaları, malformasyonların tanısında ve bu hastaların tedavi planlarının düzenlenmesinde kritik bir role sahiptir. Yeni gelişmeler ve görüntüleme tekniklerinin kullanımındaki artışla kraniospinal çoğu vasküler malformasyon tespit edilebilir hale gelmiştir. Bu durum lezyonların tanısında, karakterizasyonunda ve zamanında tedavisinde uzmanlaşmayı gerekli kılmaktadır (2).

Yetişkinlerde en yaygın görülen vasküler beyin lezyonları arterio-venöz (AV) şantlar ve kavernöz malformasyonlar olup sırasıyla tahmini tespit edilme oranları yılda 100.000 yetişkinde 1.0 ve 0.5'tir (3,4). Spinal kord vasküler lezyonları nadir olup bunların %70'ten fazlasını spinal dural AV şantlar oluşturmaktadır (5). Vasküler malformasyonların altında yatan ana patoloji

kapiller, venöz ve arteryel yatakların bütünlüğünün bozulmasıdır. Bu bütünlük kaybı, mekanik hasar gibi dış sebeplere bağlı olabilir ve/veya anjiogenez, damar gelişimi ve matürasyonu esnasında vasküler gelişimdeki defektlere bağlı olabilir (6).

Geçen yıllarla beraber birçok SSS vasküler malformasyonunun genetik temeli daha iyi açıklanmıştır. Bununla birlikte vasküler malformasyonların çoğu sporadik olarak oluşur. Bireyleri vasküler malformasyon oluşumuna ve komplikasyonlarına duyarlı kılan birçok genetik varyant tanımlanmıştır (7). Bu genetik varyantlar başlıca kan-beyin bariyer bütünlüğünü, tumor growth factor-B (TGF-B) sinyal yolunu, lokal enflamatuvar cevabı, anjiogenezi ve doku remodelingini düzenler. Ayrıca Endoglin, activin-like kinase (ALK) receptor 1, somatic-activating KRAS ve RASA-1 geni SSS vasküler malformasyonu oluşumuna katkıda bulunurlar (8-10).

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