BÖLÜM 53

VASKÜLER KOMPONENTLERİN BELİRGİN OLDUĞU NÖROKÜTAN SENDROMLAR

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A) STURGE WEBER SENDROMU

Sturge-Weber sendromu (SWS), fasiyal kapiller malformasyon (porto şarabı lekesi) ile karakterize, kapiller-venöz malformasyonlarla ilişkili olarak beyni ve gözü etkileyen nadir görülen bir konjenital vasküler bozukluktur. Kalıtsal bir bozukluk olmadığından tekrarlama olasılığı düşüktür.[1,2]

SWS'nin nedeninin, normal doku örneklerinin ve tam genom diziliminin incelendiği bir çalışmada tanımlandığı gibi GNAQ genindeki somatik mozaik mutasyonlardan kaynaklandığı gösterilmiştir.[3] GNAQ geni kromozomal olarak 9q21.1 bölgesinde lokalize olup, kan damarlarının gelişiminde önemli bir role sahiptir. Genin aktive edici mutasyonları aynı zamanda, apoptozun inhibisyonuna ve hücresel proliferasyonun uyarılmasına yol açmaktadır.[1,3] Bir çalışmada, SWS'li 26 katılımcıdan 23'ünden (% 88) ve sendromik olmayan (izole) porto şarabı lekesi olan 13 katılımcıdan 12'sinin (% 92) etkilenen doku örneklerinde mutasyon bulundu, ancak diğer serebrovasküler malformasyonları olan 4 katılımcıda ve 6 sağlıklı kontrolde mutasyon bulunmadı. Bu bulgular, SWS'nin kapiller venöz malformasyonlarının, kapiller kan damarı oluşumunun uygunsuz kontrolüne ya da olgunlaşmasına neden olan fetal ektodermal dokulardaki somatik mutasyonlardan kaynaklandığına dair uzun süredir devam eden hipotezi doğrulamaktadır.[4,5]

SWS insidansı ~1:50.000 olarak tahmin edilmektedir, erkekler ve kadınlar arasında anlamlı bir fark yoktur. Kalıtsal bir patern veya yatkınlık ve malign transformasyon gösterilmemiştir.[6-9]

Trigeminal sinirin birinci dalı boyunca tek taraflı fasiyal porto şarabı lekesi (Şekil 1), hemiatrofi, dirençli nöbetler, kontralateral hemiparezi, mental yetersizlik, hemianopsi ve ipsilateral glokomdan oluşan SWS'nin klasik klinik bulguları olduğunda tanı kolaylıkla konur. Ancak, Waelchli ve arkadaşları, PWS dağılımının aslında trigeminal sinirden ziyade yüzün embriyonik vasküler dağılımını takip edebileceğini gösterdi. Nörogörüntüleme teknikleri tanı koymakta yardımcı olur ve ayrıca parietal ve oksipital lobları tutan giriform kalsifikasyonları, leptomeningeal anjiyomatozu ve beyindeki astrogliosisi gösterir. SWS tanısı, klasik triadın üç belirtisinden en az ikisi (leptomeningeal anjiyom, porto şarabı lekesi ve okü-

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lesel kavernöz anjiomatozis, POEMS sendromu), kılcal hemanjiyomlar (Rubinstein-Tayabi, Coffin-Siris), telenjiektazi (konjenital telenjiektatik kutis marmorata, Cockayne, De Sanctis-Cacchione) ve anjiyokeratom (Fabry hastalığı, Fucosidosis) olarak sınıflandırılabilir.[135]

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