BÖLÜM 54

DERİ VE SUBKUTAN DOKU ANOMALİLERİ İLE GİDEN NÖROKÜTAN SENDROMLAR

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GİRİŞ

Nörokütan hastalıklar genellikle genetik geçişlidir. Ektoderm kaynaklı olup hem deriyi hem de sinir sistemini etkiler. Sıklıkla yaşamın erken döneminde bulgu verir, ancak yaşamın daha ileri aşamalarında tanı alan hastalar da vardır. Bu grup hastalıklara 'nörokütan sendromlar' adı verilir. Bir diğer adı ile 'fakomatozlar' olarak adlandırılır ve Latincede doğum lekesi anlamına gelmektedir.

Nörokütanöz hastalıklar heterojen bir grup olup, nörojenik ve kütanöz rahatsızlıklar birarada olduğu için çoğu zaman sinonim olarak değerlendirilmiştir. Bu hastalıkların bir kısmının genetik geçişi bulunmakta iken bazıları sporadik olarak ortaya çıkabilir. Bu durum gelişimsel geriliğe, epileptik nöbetlere, beyinde displazilere neden olabilmektedir. Nörokütanöz sendromlar olarak bilinen hastalıklar displastik yapılarıyla ve özellikle deri ve sinir sistemi gibi çeşitli organlarda tümör oluşturmaya eğilimleriyle dikkat çekmektedir.[1] Bu bölümde daha sıklıkla karşımıza çıkan, deri ve subkutan bulgularıyla seyreden nörokütan hastalıkları ele alacağız.

Nörofibramatosis

Bu grubun en sık görülen hastalığıdır. Tek bir hastalık olmayıp çok geniş spektrumludur. Nörofibramatosis (NF) en sık görülen tipleri; nörofibramatosis tip 1(NF-1displazilere neden olabilmektedir tip 2 (NF-2) ve schwannomatosis olarak sıralanabilir. Bu hastalık grubunda santral sinir sisteminde tümür oluşumu riski yüksektir.[2] Nörofibramatosis tip 1 ve tip 2'nin genetik geçişi %50 oranında otozomal dominanttır, kalan %50'si de novo mutasyon olarak karşımıza çıkar. De novo mutasyon oranı diğer bir tip olan schwannomatosiste çok daha yüksektir.[3]

NF-1

Tüm NF tiplerinin %85'ini kapsar. Ortalama 3000 kişide 1 sıklığında görülür. NF-1 Von Recklinghausen hastalığı olarak da bilinir.[1] Çeşitli malignite oluşumu ve diğer komplikasyonlardan ötürü normal sağlıklı insan yaşam süresinden yaklaşık 15 yıl daha az yaşam süresi beklenir.[4] Hastalığın tanımlanmış tanı kriterleri mevcuttur. NF tanı kriterlerinden ikisini karşılaması hastalara NF tip-1 tanısını koydurur. Hastaların %97'si 8 yaşına geldiği zaman bu kriterlerden ikisini karşılayarak tanı almış

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hastalıktır.[71] İlk olarak Aksiyel hipotoni, eklem hipermobilitesi eşlik edebilir. PIK3CA mutasyonu ve varyantları genetik incelemede bazı vakalarda gösterilmiştir. Tedavi semptomatiktir.[72]

Bannayan- Riley- Ruvalcaba Sendromu

Nadir görülen bir hastalık olup, overgrowth ve kütanöz bulguları içerebilen bir hastalıktır. Gerçek prevelansı bilinmemekle beraber, genetik olarak PTEN geninde 10q23.3 %60 hastada raporlanmıştır.[73] Klinik bulguları; makrosefali, genital melonotik maküller, hamartamatöz intestinal polipler, vaskuler veya lipomatoz hamartomlar (anjiokeratom, lenfanjiokeratom, fasial, oral veya akral verrukoz papüller), fasial dismorfoloji, oftalmolojik anomaliler, kas-iskelet veya sinir sistemi anomalileri eşlik edebilir. Göz anomalilerinden strabismus, ambliyobi, pseudopapilödem gözlenebilir.

Tedavi oluşan komplikasyona yöneliktir. Cilt bulguları için eksizyon, lazer kriyoterapi uygulanabilir.[73-74]

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