BÖLÜM 48

NÖROKÜTAN HASTALIKLARA GİRİŞ

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GİRİŞ

Nörokütanöz sendromlar (fakomatozlar), nöroektodermal ve bazen mezodermal gelişim anormalliklerini içeren cilt, göz ve merkezi sinir sistemi tutulumu ile giden çeşitli doğumsal bozukluklar grubudur. Bulgular tipik olarak erken çocukluk veya ergenlik döneminde mevcuttur. Çoğu nörokütanöz sendrom, tek gen hastalıkları olarak sınıflandırılır, ancak otozomal dominant, otozomal resesif veya X'e bağlı kalıtım paternlerine bağlı olarak da ortaya çıkabilirler.[1] Spontan mutasyonlar yaygın olmasına rağmen, birçoğunun zaten tanımlanmış genetik anormalliği vardır ve geniş bir fenotipik yelpazesi vardır. Genetik teknolojilerdeki son gelişmeler, birçok nörokütanöz bozukluktan sorumlu olan spesifik genetik kusurlar ve protein işlevleri konusundaki bilgilerimizi artırmış böylece birçok ilişkili komplikasyon için biyolojik temelli, hedefe yönelik tedaviler geliştirilmesini sağlamıştır. Bu sendromların çoğu, birçok organ sistemini etkilediklerinden, doğaları gereği belirgin şekilde heterojendir. Bu hastalıkların tümör oluşumuna yatkınlıklarından dolayı hastaların multidisipliner yaklaşımla takip edilmesi, komplikasyonlar ve olası bir tümör gelişimi durumunda erken müdahale edilmesi açısından önemlidir.[1,2,3]

Nörokütanöz hastalıkların tarihçesi değerlendirildiğinde, milattan önce 200 yılında yapıldığı düşünülen bir heykelde göğüs ve kollarında nörofibromlara benzeyen cilt nodülleri mevcuttur.[3] Dr. Robert William Smith 1849'da ilk defa nöromun patolojik tanı ve tedavisinden bahsetmiş, daha sonra Von Recklinghausen 1882'de NF Tip 1'i tanımlamıştır. Yunanca kelime anlamı 'doğum lekesi' olan fakomatoz terimini ilk defa 1923 yılında Dr. Jan van der Hoeve kullanmış ve daha sonra 1932'de de nörokütanöz hastalıklardan bahsetmiştir.[4] Bourneville hastalığı olarak da bilinen Tübrosklerozis, Bourneville tarafından 1880 yılında çocukluğundan bu yana nöbetleri olan 15 yaşında bir vakada tanımlanmıştır. 1908'de Vogt hastalığın klasık triadı olan epilepsi, mental retardasyon, adenoma sebaseumları tanımlanmıştır.[2,4]

1879 yılında Sturge parsiyel epilepsisi ve beyinde vazomotor merkezde lezyonu olan bir vaka tanımlamıştır.1904'de Eugene von Hippel ve 1926'da Arvid Lindau tarafından Von Hippel–Lindau Hastalığı tanımlanmıştır.[5]

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ayırt etmede zorluk vardır. Asimetrik SWS'de hemiparezi ortamında uzuv atrofisi meydana gelebilir ve bu da tanı ve ayırıcı tanıyı daha da karmaşık hale getirebilir.[94]

Vasküler ektaziler, diseksiyon ve intrakranial anevrizmalar dahil olmak üzere KT ile ilgili inmelerin çoğunda serebrovasküler anormallikler rol oynar.[100] Serebral anevrizmalar, bazen bu durumla ortaya çıkabilen kronik tüketim koagülopatisinin oluşumunda özellikle olumsuz sonuçlara sahip olabilir.[101] KT'de baş ve boyundaki kütanöz olmayan vasküler anormalliklerin hastaların yaklaşık %10'unu etkilediği tahmin edilmektedir.[101] Bu hastalarda MR anjiyografi, BT anjiyografi veya konvansiyonel anjiyografi ile anjiyografik taramanın dikkate alınması gerekir. En yaygın MSS bulgusu, genellikle kas-iskelet hipertrofisi ve deri lezyonları ile aynı tarafta beyin boyutundaki değişikliktir.[102] Serebral ve serebellar hipertrofi ile hemimegansefali bildirilmiştir.[102,103] Bu hastalar ayrıca serebral AVF ortamında tarif edilen nöbetlerden ve bazen kortikal gri madde malformasyonları veya kortikal heterotopilerle ilişkili hemimegalensefaliden de etkilenebilirler.[100,103] KT'de oftalmik tutulum nadirdir ve hem retinal hem de koroidal vasküler anormallikleri (diffüz koroidal anjiyom gibi), maküler telenjiektazileri, retina displazisini ve retinal arteriyovenöz iletişimi içerebilir.[9]

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