BÖLÜM 22

KORTİKAL GELİŞİM MALFORMASYONLARI

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GİRİŞ

Kortikal gelişim malformasyonu (KGM) terimi ilk olarak 1996 yılında insan serebral korteksinin normal gelişimsel sürecindeki bozukluklardan kaynaklanan ve kortekste çok çeşitli gelişimsel bozukluklara neden olan hastalık grubunu tanımlamak için kullanıldı. Nörogelişimsel gecikmenin yaygın nedenlerinden biridir ve sıklıkla epilepsiye neden olur.[1]

Kortikal gelişim malformasyonları; genetik, enfeksiyöz, vasküler veya metabolik etiyolojilerin neden olduğu bozulmuş serebral korteksle ilişkili heterojen büyük bir grup hastalığı tanımlamak için kullanılmaktadır. Anormal kortikal yapı veya anormal beyin boyutu (mikrensefali ya da megalensefali) ile ilişkilidir. Her yaşta önemli morbiditeye neden olabilir. En yaygın görülen semptomlarından olan epilepsi erken çocukluktan adölesan döneme kadar değişen dönemde başlayabilir. Gelişimsel gecikme, entelektüel bozukluklar veya hemi-kuadri parezi veya pleji şeklinde de kendini gösterebilir. Özellikle, çocuklarda cerrahi ile tedavi edilen ilaca dirençli epilepsilerin %40-50'sinin KGM kaynaklandığı tahmin edilmektedir.[2]

Kesin tanısı nöropatolojik bulgulara dayansa da epilepsi cerrahisi veya otopsi materyalleri ile değerlendirildiğinden nadiren patolojik tanı konur. Bu nedenle çoğu zaman en pratik yol nörogörüntüleme yöntemleridir. Alt sınıflandırmada görüntülemeye ek olarak ilişkili klinik fenotipleme ve genetik bulgular da kullanılmaktadır.[2] Manyetik rezonans görüntüleme (MRG) kortikal displazilerin tanısında değerli bir yöntemdir ve bozuklukların %50-70'inin tanınmasını sağlar. Volümetrik MRG kullanılabilir ve en uygun anatomik değerlendirme için T1 ağırlıklı serilerde devamlı ince kesit görüntüler gerekir.[3] Nörogörüntülemenin iyi değerlendirilmesi ile hedefli genetik testler planlanarak ve/veya yeni genotip-fenotip korelasyon keşifleri ile tanı oranı artırılabilir. Yaş grubuna uygun olan teknikte, yüksek çözünürlüklü alınan MRG görüntülerinin patern tanıma konusunda uzmanlaşmış ehil kişilerce yapılması da tanı için önemlidir.[2] Elektroensefalogram (EEG) tanıya yardımcıdır ancak bulgular spesifik değildir.[3]

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