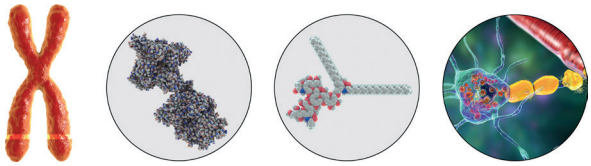

BÖLÜM 14

KONJENİTAL DEMİYELİNİZAN HASTALIKLAR

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GİRİŞ

Demiyelinizan hastalıklar, santral ve periferik sinir sisteminde aksonları saran miyelin kılıfın, konjenital veya akkiz nedenlerle yıkıma uğraması sonucu gelişen, geniş bir klinik spektruma sahip hastalıklar toplamıdır.

Demiyelinizan hastalıklar; otoimmun, enfeksiyöz, toksik/metabolik, vasküler ve myelin metabolizmasının herediter hastalıkları şeklinde sınıflandırılabilir (1).

Konjenital demiyelinizan hastalıklarda genellikle oluşmuş miyelin kılıfın hasarlanmasından ziyade, miyelin kılıfın yapım aşamasında genetik bir defekt söz konusudur. Kliniğin ortaya çıkması genelde hastalığın erken evrelerinde görülür. Burada herediter miyelin hastalıklar alt başlıklar halinde incelenecektir.

ADRENOLÖKODİSTROFİ (ALD)

Uzun zincirli yağ asidi metabolizmasında defekt ile ilişkili tablo, en yaygın kalıtsal peroksizomal hastalıktır. Progresif seyirde; santral sinir sisteminde beyaz cevher lezyonları ve adrenal gland disfonksiyonu bulguları eşlik eder.

Santral sinir sisteminde genellikle korpus kallosum ve oksipitoparyatel bölgeyi içine alan kranyal, nadiren de spinal kordda simetrik beyaz cevher lezyonları gözlenir. İnflamatuvar süreç öncelikle miyelin kılıf ve oligodendrosit hasa-

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Ayrıca hastalığın medikal tedavisinde fenilalanin düzeyinin düşürülmesine yarayan sapropterin, uygun diyet tedavisine ek olarak kullanılabilmektedir. Ne yazık ki tüm hastalarda uygun tedavi yanıtı vermemektedir. 2018'de FDA tarafından onaylanan enjekte edilebilir bir pegile fenilalanin amonyak liyaz olan ve halen oldukça yeni olan terapötik pegvaliaz, hastalığın tedavisi için yeni bir umut oluşturmaktadır (75).

SONUÇ

Herediter demiyelinizan hastalıklar, genellikle miyelinin yapım aşamasında bir defektle karakterize, çoğunlukla enzim eksikliğine sekonder gelişen hastalıklardır. Tutulum yerine ve etkilenim şiddetine göre değişik klinik bulgu verebilirler. Tedavileri çok yüz güldürücü olmasa da genetik danışmanlık, erken tanı ve uygun destek tedavileri ile hastalık yönetimi mümkün olmakta, tanı ve tedavi yöntemlerine dair güncel çalışmalar devam etmektedir.

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