# BÖLÜM 87

# MEGALOENSEFALİ İLE GİDEN METABOLİK HASTALIKLAR

Meltem UZUN [1]

## GİRİŞ

Pediatrik nörodejeneratif ve nörometabolik hastalıklarda klinik ve genetik olarak heterojen çok geniş bir grup hastalıktır. Ayrıntılı olarak hastanın şıkayetleri, prenatal, natal postnatal,-ve aile öyküsü, gelişim basamakları, muayene bulguları, labaratuvar, görüntuleme, metabolik ve genetik testler ve klinik bulguları değerlendirilerek tanı, tedavi ve izlem planlanmaktadır.

Bazı norodejeneratif ve nörometabolik hastalıklarda klinik olarak anahtar bulgulardan birisı fizik muayenede makrosefali ve/veya megalensefalinin varlığıdır.

Bu bölümde megalensefalinin eşlik ettiği nörometabolik hastalıklarda klinik bulgular ve tanısal yaklaşımlar özetlenecektir. Bu nedenle bazı tanımları yapmak faydalı olacaktır.

Makrosefali, baş çevresi ölçümünün yaşa göre iki standart sapmadan (>98 persentil) daha büyük olmasıdır[1]. Her 50 çocuktan biri bu tanımlamaya uyduğu için, makrosefali yaygın bir durum olarak kabul edilir[2]. Rölatif makrosefali ise baş çevresi büyüklüğü iki standart sapmadan daha az olup, boy kiloya göre orantısızlığı mevcuttur. Baş çevresi ölçümü alnın ortasından oksiputa kadar olan daire seklinde ölçülüp yaş cinsiyet boya göre baş çevresi büyüme kartlarından kontrol edilip belirtilmelidir[3].

Hidrosefalide de artmış beyin omurilik sıvısı ventrikülleri genişleterek makrosefaliye yol açar[4]. Ventriküllerde bu genişleme ve bazen kominike ya da nonkominike olmasına göre değ,şik klinik gösterebilmektedir. Kominike hidrosefali, genellikle yapısal bir anormallik nedeniyle ventriküler sistemde tıkanma sonucu gelişirken nonkominike hidrosefali ise beyin omurilik sıvısı (BOS) nın araknoid villuslardan anormal emilimi neticesinde gelişir.

Yine bir diğer makrosefalı nedeni subdural hematomlardır. Kronik subdural hematomlar en sık kazara olmayan travmanın sonucudur. Frontal ve parietal daha belirgin olarak saptanmaktadır[5].

Makrosefali, ayrıca çok sayıda genetik sendromlarla birliktedir. Bunlar; Akondroplazi ve diğer iskelet displazileri (orantısız olması nedeniyle göreceli makrosefali ile ortaya çıkabilir), neurofibromatosis type I, Sotos syndrome, Fragile X ve birçok kromozal anormalliklerdir.[6,7]

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