Bölüm
9

KONJENİTAL ADRENAL HİPERPLAZİ HİPERTANSİYON İLİŞKİSİ

Tülay OMMA[1]

GİRİŞ

Hipertansiyon (HT) erişkin hipertansif popülasyonun yaklaşık %15`inde hormonal bir bozukluk ile ilişkilidir. Bunların çoğunluğunu pirimer aldosteronizm oluşturur (1, 2). Bununla birlikte seçilmiş adölesan ve erişkin hipertansif hastalarda konjenital adrenal hiperplazi (KAH) olarak bilinen kalıtsal steroidojenik defektler de akla gelmelidir.

KAH, kolesterolden kortizol biyosentezine kadar olan biyokimyasal yolakta rol alan protein ve enzimlerden birinde genetik defekt ile karakterize bir bozukluktur. Kortizol sentezindeki eksiklik sonucu hipofiz bezi üzerinde kortizol negatif feedback aksı bozulur ve hipofiz bezinden kortikotropin (ACTH) üretimi artar. Artan ACTH ise adrenal hiperplazi ve adrenal steroidlerin fazla üretilmesine neden olur. Klinik özellikler defekt olan enzim bölgesine bağlı olarak kortizol, mineralokortikoid (MK) ve androjenlerin artmış ya da azalmış hızına göre şekillenir. KAH genel olarak klasik ve nonklasik (geç başlangıçlı, NKAH) olarak iki gruba ayrılır. Klasik tip ise enzimatik aktivitenin hemen hemen yok olduğu 'tuz kaybettiren form' ve % 1-2 enzimatik aktivitenin bulunduğu 'basit virilizan' tip şeklinde iki gruba ayrılır.

Tüm dünyada KAH'ın en sık nedeni (>%90) 21 hidroksilaz enzim eksikliğidir. Enzim eksikliklerinin prevalansı coğrafi bölgeye göre değişmektedir. Ülkemizde ise 11 beta hidroksilaz eksikliği daha fazla görülmektedir. Bu iki enzim sadece adrenal bezde eksprese edilirler. Diğer nadir tipler olan yan zincir klivaj (P450scc) enzimi, 17 alfa hidroksilaz ve 3 beta hidroksisteroid dehidrogenaz enzim eksikliğinde, bu üç enzim hem adrenal hem de gonadlarda eksprese edildiğinden hem

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Tablo 6: KAH'da yıllık takip planı			
	Sonlanım	**Gerekçe**	**Ölçüm**
Kısa Dönem	1.Büyüme hızı 2.Kilo değişikliği 3.Doğru tedavi doz 4.Kan basıncı 5.Puberte	1.Kontrol 2.Doz ayarı 3.Optimize tedavi 4.Tedavi etkileri 5.KAH da zamanlama değişebilir	1.>2cm/yıl ise dikkat 2.>2kg/yıl ise dikkat 3.1 ve 2.maddeler ve kan testleri 4.Kan basıncını karta işle 5.Muayene
Orta Dönem	1.Kemik maturasyonu 2.Pubertal durum 3.Hidrokortizon dozu 4.Fludrokortizon dozu 5.Testis ve over sağlığı 6.Metabolik durum	1.İskelet maturasyon hızı 2.Erken puberte, hızlı gelişim 3.Optimize tedavi 4.Hipertansiyondan kaçın 5.KAH'ın gonad etkisi 6.İnsülin duyarlılığı ve lipidler	1.Yıllık kemik yaşı 2.Tanner evrelemesi 3.Kortizol ve 17OHP 4.Plazma renin aktivitesi 5.Kızlarda pelvik USG Erkeklerde dikkatli muayene 6.Açlık glukoz, insülin,lipidler
Uzun Dönem	1.Büyüme 2.Kemik mineralizasyonu 3.Fertilite 4.Kardiyovasküler risk	1. Sonuçlar 2.KAH/tedavinin kemik üzerine etkisi 3.KAH etkisi 4.KAH/tedavi etkisi	1.Final boy 2.DXA tarama 3.Kızlarda menstrüel siklus, Erkeklerde testiste adrenal rest tümör 4.Açlık glukoz, insülin,kan basıncı,açlık lipidler

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