BÖLÜM 1

İNFERTİLİTE İLE İLİŞKİLİ SENDROMLAR

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DE LA CHAPELLE SENDROMU

Tanım

De La Chapelle sendromu, ilk olarak 1964 yılında A. de la Chapelle ve arkadaşları tarafından tanımlanmış nadir bir genetik bozukluktur (1). Bu sendrom, kromozomal cinsiyet ile gonadal ve fenotipik cinsiyet arasındaki uyumsuzlukla karakterizedir (2). Geçmişte De La Chapelle sendromu olarak bilinen bu durum, günümüzde genellikle 46,XX erkek sendromu veya 46,XX testiküler cinsiyet gelişimi bozukluğu (DSD) olarak adlandırılmaktadır. 2006 yılında Chicago Uzlaşma Grubu tarafından resmi olarak "46,XX testiküler cinsiyet gelişimi bozukluğu" ismiyle anılması önerilmiştir (1,3).

De La Chapelle sendromu, fenotipik olarak erkek olan ancak 46,XX genotipine sahip bireylerde görülen nadir bir genetik durumdur (3). Normalde kadınlara özgü olan 46,XX kromozom yapısına rağmen, bu bireyler erkek dış genital organlarına ve testis

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Erkek hastalarda penis boyutunu artırmak ve virilizasyonu desteklemek için yüksek doz testosteron veya dihidrotestosteron (DHT) denenebilir, ancak sonuçlar sınırlı ve değişkendir. Gonadektomi yapılan kadın hastalarda ise puberteyi başlatmak ve ikincil seks karakterlerini geliştirmek için östrojen replasman tedavisi zorunludur (49).

Cinsel kimlik, fertilite ve yaşam kalitesi gibi konularda hastalar ve aileleri için psikolojik destek tedavinin ayrılmaz bir parçasıdır (50).

Sonuç olarak, Reifenstein sendromu (PADS), androjen reseptör fonksiyonundaki kısmi bozukluktan kaynaklanan ve infertilite ile yakından ilişkili, geniş bir klinik yelpazeye sahip bir durumdur. Özellikle hipospadias, kriptorşidizm ve jinekomasti gibi ürolojik bulgularla karakterizedir. Tanı ve tedavisi, multidisipliner bir yaklaşım gerektirir ve hastanın yaşam kalitesini artırmak için erken tanı, uygun cerrahi ve hormonal müdahaleler ile psikososyal destek kritik öneme sahiptir.

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