Bölüm 2

MEMELİ GENİTAL FARKLILAŞMASININ GENETİK KONTROLÜNE GENEL BİR BAKIŞ

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GİRİŞ

Cinsiyet gelişimi bireyin fetal hayatı boyunca meydana gelen ve ergenlikle birlikte tamamlanan kompleks biyolojik bir süreçtir. Bu süreç temel özelliklerine göre birbirini takip eden dört aşamada incelenmektedir. Bunlar; kromozomal cinsiyet tayini, farklılaşmamış gonadın over yada testise dönüşümünün yer aldığı gonadal farklılaşma, iç genital ve dış genital organların gelişimini içeren birincil cinsiyet tayini ve puberte ile birlikte gonadların hormonal uyarısı sonucu dokuların cevabını içeren "ikincil cinsiyet tayini" olarak sıralanmaktadır (Lim & Hawkins, 1998).

Kromozomal cinsiyet tayini fertilizasyon sırasında bireyin sahip olduğu genotipteki gonozomlarca (XX/XY) belirlenmektedir. Bu süreçte iki adet X kromozomu taşıyan birey dişi fenotipte olması beklenirken, 1 adet X ve 1 adet Y kromozomu taşıyan bireyin ise erkek fenotipinde olması beklenmektedir(Sadler, 2005). Ancak, krossing over hataları ve bazı mutasyonlara bağlı olarak oluşan genotip beklenen fenotipi göstermeyebilir (Berkovitz & Seeherunvong, 1998). Diğer taraftan, her ne kadar genetik olarak cinsiyet tayini fertilizasyonda belirli olsa da, gelişimin ilk 7 haftalık sürecinde erkek ya da dişi olarak fenotipik herhangi bir cinsiyet gelişimi mevcut değildir. Farklılaşmamış evre olarak da bilinen bu süreçte öncelikle genital sırtlar oluşmaktadır ve bu sırtlara primitif germ hücreleri (PGH) göç ederek bipotansiyel gonad oluşumu sağlanmaktadır (Sadler, 2005). Farklılaşmamış gonadal primordiyum, çeşitli farklılaşmamış hücre tiplerini içermektedir. Bunlar; primitif germ hücreleri, steroidojenik öncül hücreler ve destek öncül hücreleri olarak sıralanmaktadır (Kim & Capel, 2006). Bu hücrelerin çeşitli transkripsiyon faktörleri ve sinyal yolaklarınca uyarılması sonucu bipotansiyel

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SONUÇ

Genital sırt oluşumuyla başlayan ve gonadal farklanma, iç genital ve dış genital organ oluşumuyla devam eden genital gelişim sürecinde başta SRY geni olmak üzere birçok gen önemli rol oynamaktadır. Yaklaşık 30 yıldır devam eden araştırmalarda bu genetik sinyal yolaklarının tanımlanabilmesi ve bu yolakların fonksiyonel olmasında etkili mekanizmaların açıklanması amaçlanmıştır. Genital farklanmayı sağlayan bu moleküler sinyal mekanizmalarının belirlenmesi ise fertilitenin uzun yıllarca korunabilmesi açısından oldukça büyük öneme sahiptir.

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