# BÖLÜM 3

# EMBRİYOLOJİ VE ETİYOPATOGENEZ

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## DİYAFRAM GELİŞİMİ VE KONJENİTAL DİYAFRAM HERNİSİNİN PATOGENEZİ

İnsan diyaframının gelişimi, halen tam olarak açıklığa kavuşturulamamış, son derece karmaşık bir embriyolojik süreçtir. Bu yapı, farklı embriyonik dokuların zamansal ve mekansal olarak koordine edilmiş etkileşimleri sonucunda oluşur. Diyaframın morfolojik temelleri gebeliğin yaklaşık dördüncü haftasında belirginleşmeye başlar. Klasik embriyolojik teoriye göre diyafram, dört ana embriyonik komponentin füzyonuyla meydana gelir: septum transversum, plöroperitoneal kıvrımlar, **özofageal mezenterin krural bölümü** ve dorsal vücut duvarı mezodermi (Şekil 1). Bu yapılar arasında gerçekleşen koordineli füzyon, torasik ve abdominal boşlukların birbirinden ayrılmasını sağlar (1,2).

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Bu iki olayın birleşimi, hem alveoler hem de vasküler hipoplaziye ve sonuçta pulmoner hipertansiyona yol açar (17).

### Klinik önemi ve prognostik değer

KDH-PH, mortalite ve morbiditenin en güçlü belirleyicilerinden biridir. Yenidoğan döneminde pulmoner arter basıncının doğumdan sonraki ilk haftalarda normal sınırlara inmemesi, prognoz açısından olumsuz bir göstergedir (45). Yapılan retrospektif bir analizde, yaşamın ilk üç haftasında pulmoner arter basıncı normale dönen bebeklerde %100 hayatta kalma oranı, buna karşın yüksek basınçların devam ettiği olgularda %100 mortalite bildirilmiştir (46).

Klinik olarak KDH-PH, genellikle sağdan sola şant (foramen ovale ve duktus arteriozus düzeyinde) ile seyreder. Bu durum, oksijenizasyonu ciddi şekilde bozar. İleri olgularda sağ ventrikül yetmezliği, sistemik hipotansiyon ve çoklu organ yetmezliği gelişebilir.

KDH'ye bağlı pulmoner hipertansiyonun gelişiminde, akciğer ve damar sistemlerinin embriyolojik olarak paralel ilerleyen iki sürecinin birlikte bozulması belirleyici rol oynar. Diyafram defekti yalnızca mekanik bir etken değil, aynı zamanda moleküler düzeydeki bir gelişimsel kusurun da göstergesidir. Bu nedenle gelecekteki tedavi stratejilerinin, pulmoner vasküler gelişimin erken dönemde modülasyonu ve retinoid sinyal yollarının düzenlenmesi üzerine yoğunlaşması beklenmektedir.

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