KALITSAL ERİTROSİT ENZİM EKSİLİĞİ İLİŞKİLİ HEMOLİTİK ANEMİLER

18. BÖLÜM

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Giriş

Yaşam döngülerinde eritrositler, olgunlaşma sırasında nükleus, mitokondri ve diğer organellerini kaybetmelerinden dolayı oksidatif fosforilasyon, protein ve lipit sentezi yapamazlar. Hücresel fonksiyonların sürdürülebilmesi için enerji gereksinimini anaerobik glikoliz ile sağlarlar ve yine hücre bütünlüğünün ve esnekliğinin korunması, oksijenin taşınması, eritrositler için toksik bir takım nükleotid öncüllerinin uzaklaştırılması, hücrenin oksidatif hasardan korunması birçok enzimin görev aldığı metabolik yolaklar ile sağlanmaktadır. Bu yolaklar Embden-Meyerhof yolu, heksoz monofosfat şantı, glutatyon yolu, Rapoport-Luebering şantı ve nükleotid metabolizmasıdır. Bu yolaklar ve görevli enzimler **Şekil 1' de** gösterilmiştir (1-5).

Enzimleri kodlayan genlerde oluşan mutasyonlar sonucu bu yolaklarda görevli enzimlerde eksiklikler ya da defektler oluşur. Bu durum eritrositlerin enerji dengesini bozarak oksidatif strese yol açar ve hücre bütünlüğü bozulur. Enzim eksikliğinin tipine ve rol aldığı yolağa göre hematolojik ya da non-hematolojik klinik durumlar oluşur. Bu hematolojik klinik durumlardan bir tanesi de non-sferositik hemolitik anemilerdir (5,6).

Hemolitik anemi oluşturan en sık enzim eksiklikleri ise adenozin trifosfat (ATP) enerji gereksiminin %90' ını sağlayan yolak olan Embden-Meyerhof yolağında görevli olan piruvat kinaz eksikliği ve enerji üretiminin geri kalan kısmından sorumlu olan yolak olan heksoz monofosfat şantında görevli enzim olan glukoz-6 fosfat dehidrogenaz (G6PD) eksikliğidir (6,7).

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Sonuç

Enzim eksikliklerine bağlı hemolitik anemiler nadir görülseler de immun olmayan hemolitik anemi, açıklanamayan sarılık, dalak büyüklüğü gibi durumlarda akılda tutulması gereken hastalık grubudur. Çoğunluğu asemptomatik seyretse de erken müdahale edilmediğinde mortal kliniklere neden olabilirler.

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