



## BÖLÜM 3

# Konjenital Trombositopeniler

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### Giriş

Trombositopeni, trombosit sayısının 150.000/MikroL'un altında olması olarak tanımlanır. Kolay morarma veya kanama öyküsü ile başvuran hastalarda, rutin değerlendirme veya başka nedenlerle yapılan tetkikler sırasında tesadüfi olarak ortaya çıkabilir. Konjenital trombositopeniler, erken başlangıçlı trombositopeni ile karakterize olup, hafif semptomlardan, yaşamı tehdit eden kanamalara kadar değişen klinik bulgular ile karakterize nadir, heterojen bir hastalık grubudur (1, 2). Bu sendromların heterojenliği göz önüne alındığında, birkaç sınıflandırma yöntemi vardır (3).

1. Altta yatan genetik mutasyon (Tablo 1'de),
2. Trombosit boyutu (Tablo 2'de)
3. Trombosit işlevi ile ilişkili anormallikler

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