



BÖLÜM 5

DİSTONİ'DE BOTULİNUM NÖROTOKSİN UYGULAMALARI-1

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DİSTONİDE BOTULİNUM NÖROTOKSİN UYGULAMALARI

Distoni; sürekli veya aralıklı kas kasılmaları sonucu gelişen anormal, sıklıkla repetitif, istemsiz hareket veya duruş veya her ikisinin birlikte görüldüğü nörolojik bir durumdur. Distoni, Parkinson hastalığı ve esansiyel tremordan sonra görülen üçüncü sıklıktaki hareket bozukluğudur (1).

Distoni tarihçesinde ilk yayınlar 1890'lı yıllara denk gelmesine rağmen "Distonia Musculorum Deformans (Famılyal Jeneralize Distoni, DYT)" tanımlanması ve "distoni" kelimesinin literatüre kazanılması 1911 yılında Oppenheim tarafınca olmuştur (2). Bu tanımlamadan sonra distoni ile ilgili yayınlanan literatürler sonucunda distoni şemsiyesi altına birçok heterojen hastalık grubu girmiştir. İlk distoni konsensusu 1984 yılında "Distoni Medikal Araştırma Derneği" tarafınca açıklanmıştır. Bu konsensus 2013 yılında Uluslararası Parkinson ve Hareket Bozuklukları Topluluğu (International Parkinson and Movement Disorder Society) tarafınca revize edilmiştir (3). 1994 yılında ilk olarak Otozomal dominant-Dopa yanıtı distoninin (DYT5A) GCH1 mutasyonu sonucu geliştiği tanımlandıktan sonra günümüze kadar halen devam eden genetik çalışmalarda otozomal dominant (TOR1A (DYT1), THAP1 (DYT6), GNAL (DYT25), ANO3 (DYT24), CIZ1, ANO3, TUBB4A, SGCE (DYT11), ATP1A3 (DYT12), ADCY5) ve resesif (HPCA, VPS16, COL6A3, PRKRA (DYT16)) bir çok gen mutasyonları ve X'e bağlı distoni-parkinsonizmde TAF1 mutasyonu (DYT3), erken başlangıçlı jeneralize distonide KMT2B mutasyonu gibi birçok mutasyonlar tanımlanmıştır (2). Distoni tedavisinde;

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