

PRECISION MEDICINE IN HEALTH AND DISEASE INTERNATIONAL SYMPOSIUM

Clinical Applications of Precision Medicine Session

Case Presentations 1 : Precision Medicine in Rare Disease

Date : 03.10.2024

Time : 13:00-14.00



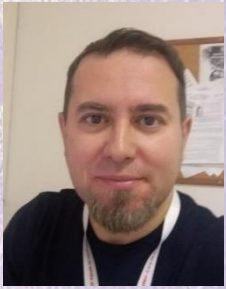
Transthyretin (TTR) Cardiac Amyloidosis, Early Findings

Asst. Prof. Dr. Erhan Pariltay, Ege University, School of Medicine, Department of Medical Genetics



A Patient with SMA: How RNA-based Diagnosis Works

Prof. Dr. Ahmet Okay Çağlayan, Dokuz Eylül University, School of Medicine, Department of Medical Genetics; Izmir Biomedicine and Genome Center



Two Cases and Prenatal Diagnosis Of A Rare UFM1 Associated Hypomyelinating Leukodystrophy

Assoc. Prof. Dr. Berk Özyılmaz, Izmir City Hospital, Genetics Diseases Diagnosis Center



Adult Onset GalC Krabbe Disease Case

Ravza Nur Yıldırım, PhD, Izmir Biomedicine and Genome Center



Molecular Diagnosis Enables Effective Treatment in a Case of Late-Onset NAXE Deficiency

Assoc. Prof. Dr. Esra Işık, Ege University, Faculty of Medicine