■ Original article

Epileptic Disord 2021; 23 (2): 313-324

Epileptic Disorders

Unmasking the entity of 'drug-resistant' perioral myoclonia with absences: the twitches, darts and domes!

Pavan Kumar Rudrabhatla, Sunesh ER, Ashalatha Radhakrishnan, Ramshekhar N. Menon

Sree Chitra Tirunal Institute for Medical Sciences and Technology, Dept of Neurology, Thiruvananthapuram, Kerala, India

Epileptic Disorders

Unmasking the entity of 'drug-resistant' perioral myoclonia with absences- the twitches, darts & domes!

Epileptic Disorders

- Peri-oral myoclonia with absences (POMA) is an epilepsy syndrome characterised by the presence of short absences similar to genetic generalized epilepsy (GGE) syndromes

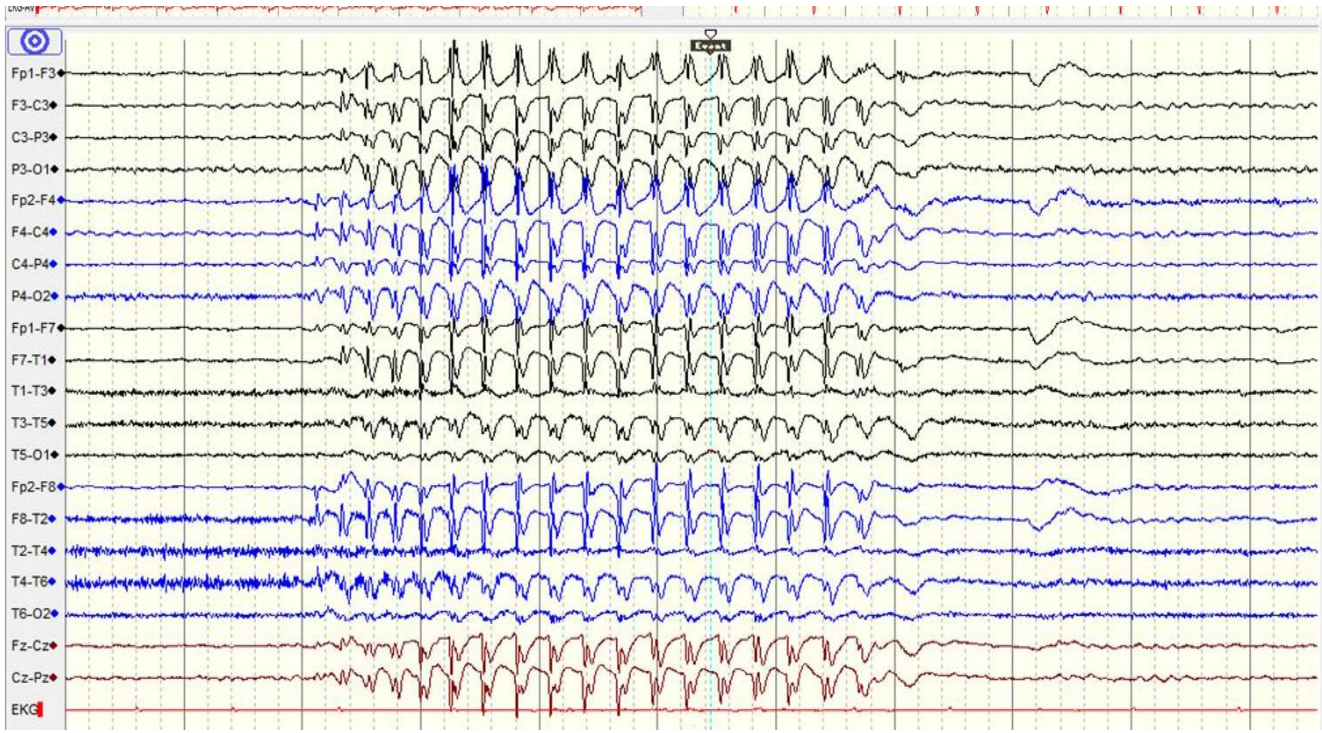
- Its distinctive feature is the associated pronounced rhythmic contractions of the perioral muscles, mainly involving the orbicularis oris

- Other features include generalized tonic–clonic seizures which follow or appear concomitant with absences, absence status, unsatisfactory response to antiseizure medications (ASM), and persistence of seizures into adult life

- Ictal rhythm is characterised by typical generalized 3-4-Hz spike/polyspike and wave discharges with a dart-dome morphology and occasionally shifting asymmetry

- The presence of clinical features suggestive of focal-onset opercular seizures or EEG asymmetry frequently leads to the misdiagnosis of POMA as focal epilepsy and mismanagement

- The actual prevalence of POMA is under-reported and the reported drug resistance is often a 'pseudo-resistance' due to misdiagnosis and management as focal epilepsy



- Management of POMA as focal epilepsy can lead to seizure aggravation and absence status complicating the clinical picture

- Hence, syndromic diagnosis is paramount and video-telemetry recordings with documentation of a characteristic 'primary absence-like' ictal rhythm and facial myoclonia are necessary to recognize POMA

- Background-slowing, atypical ictal-rhythms and pharmaco-resistance, particularly to first-line ASM for GGE syndromes such as sodium valproate are 'red-flags' in the diagnosis of this unique absence epilepsy and may suggest focal epilepsy of frontal-opercular origin