**Benign adult familial myoclonic epilepsy**

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**Introduction**

This article includes discussion of benign adult familial myoclonic epilepsy, autosomal dominant cortical myoclonus and epilepsy, BAFME, familial adult myoclonic epilepsy, and familial cortical myoclonic tremor and epilepsy. The foregoing terms may include synonyms, similar disorders, variations in usage, and abbreviations.

**Overview**

Benign adult familial myoclonic epilepsy is an inherited epileptic syndrome characterized by cortical hand tremors, myoclonic jerks, and seizures with no signs of early dementia. Worldwide prevalence is unknown, but is estimated to be less than 1/35,000. It is transmitted autosomal dominantly, and penetrance is high. This is a well-delineated disease with remarkable features that clearly distinguish it from other forms of myoclonic epilepsies. Genetic studies of the families show heterogeneity, and different susceptible chromosomal loci have been identified. Diagnosis is based on clinical and electrophysiological findings. It must be differentiated from epilepsy syndromes with prominent myoclonus features. Valproate, levetiracetam, and benzodiazepines are the most beneficial treatments.

**Key points**

- Benign adult familial myoclonic epilepsy usually presents in the second decade of life with a minor cortical hand tremor exacerbated by fatigue or emotional stress. Myoclonus usually appears around the same age and consists of erratic, arrhythmic, segmental jerks of the upper limbs heightened by posture and action. Rare tonic-clonic seizures are also a manifestation and are often precipitated by photic stimulation, emotional stress, and sleep deprivation.

- Diagnosis is based on clinical and electrophysiological findings. EEG findings include a photomyoclonic response along with abnormality of polyspikes and waves. Patients also display extremely enlarged cortical components of somatosensory-evoked potentials and an enhanced C-reflex. Jerk-locked average analysis reveals positive-negative, biphasic spikes preceding myoclonus.

- Benign adult familial myoclonic epilepsy is transmitted autosomal dominantly, and it must be differentiated from idiopathic epilepsy syndromes with prominent myoclonus features (eg, juvenile myoclonic epilepsy or Janz syndrome) and from progressive myoclonic epilepsies.

- Valproate, levetiracetam, and benzodiazepines are most beneficial in the treatment of cortical tremors and myoclonus due to their combined antiepileptic and antimyoclonic effects. In some cases, epilepsy may be difficult to treat.

**Historical note and terminology**

The term "cortical tremor" was introduced by Ikeda and colleagues to indicate a postural and action-induced shivering movement of the hands mimicking essential tremor but showing the electrophysiological findings of cortical reflex myoclonus, that is: (1) brief EMG burst of about 50 msec duration; (2) no definite synchronisation or reciprocity in the antagonist muscles; (3) positive spikes preceding EMG bursts at the jerk-locked averaging; (4) enlarged cortical components of somatosensory-evoked potentials; (5) enhanced long-loop reflex I (or C-reflex) (Ikeda et al 1990).

Uyama and colleagues first reported a patient with adult-onset fine finger tremulous movement, myoclonic jerks, and 2 generalized seizures coming from a family that was affected with the same condition through 3 generations with high
penetrance (Uyama et al 1985). None of the patients showed other neurologic signs or abnormal neuroradiological findings; electrophysiological study indicated cortical reflex myoclonus. Subsequently, the same group reported 4 unrelated families, including 27 affected members through 3 generations with high penetrance (Uyama et al 2005). In 1991, Yasuda used the term “benign adult familial myoclonic epilepsy” (BAPME) to describe 2 pedigrees in which affected members showed autosomal dominant hand tremor, myoclonus, and epileptic seizures with a nonprogressive course (Yasuda 1991). Also in these patients, electrophysiological studies showed evidence of cortical reflex myoclonus. In 1999, Plaster and colleagues and Mikami and colleagues mapped the disease on chromosome 8q24 (Mikami et al 1999; Plaster et al 1999).

Although this condition was exclusively reported from Japan until the 1990s, several reports on pedigrees with similar clinical features but with different genetic identifiers appeared from different European countries and worldwide over the past decade with different names, such as autosomal dominant cortical myoclonus and epilepsy, familial adult myoclonic epilepsy, familial cortical myoclonic tremor, familial essential myoclonus and epilepsy, familial benign myoclonus epilepsy of adult onset, and heredofamilial tremor and epilepsy. Despite phenotypic and genetic differences, the clinical and electrophysiological data point toward one syndrome (Striano et al 2005; Striano and Zara 2016).

Genetic studies of families have revealed clinical and genetic heterogeneity with at least 4 loci, including 8q24 (FCMTE1), 2p11.1-q12.2 (FCMTE2), 5p15.31-p15.1 (FCMTE3), and 3q26.32-3q28 (FCMTE4) (see section 4).

Clinical manifestations

Presentation and course

The primary clinical manifestations are persistent, continuing myoclonus and tonic-clonic seizures that only seldom occur. Age of onset is highly variable (11 to 50 years of age). The initial sign, myoclonus, usually develops between the third and fifth decades and persists through life. After onset, myoclonus persists not only at rest but also on posturing or in action and is increased by emotional stress, fatigue, lack of sleep, or intermittent light. Myoclonus is fragmentary or bilateral and most marked in the distal portion of the upper limbs. In rare cases, patients could also present with drug-resistant complex partial seizures and focal EEG abnormalities (Guerrini et al 2001).

The clinical course is apparently nonprogressive, allowing the majority of patients a normal lifespan. Daily activities are not disturbed except for some difficulties in skillful finger movements, such as writing, buttoning, and picking up small objects. However, prolonged follow-up evaluation revealed walking impairment and need for help in everyday life for individuals older than 80 years of age.

Preliminary data suggest higher prevalence of mood disorders and significant psychiatric burden compared to other forms of epilepsy and healthy controls, particularly depression and anxiety in association with pathological traits of personality, ie, paranoid, psychasthenia, schizophrenia, and hypomania (Coppola et al 2016). Therefore, a comprehensive psychiatric evaluation should be offered at the time of diagnosis to detect these comorbidities and to treat them.

Prognosis and complications

This condition usually has a benign course in patients taking antiepileptic therapy, although in some cases seizures may be frequent and unresponsive to treatment. As for other idiopathic generalized epilepsies, some antiepileptic drugs may precipitate myoclonic status. An unusually long-term (30 years) follow-up of a 4-generation South African family confirms that this condition does not cause additional progressive neurologic deterioration, and quality of life is mostly influenced by worsening of the cortical myoclonic tremor with age (van Coller et al 2017). However, in advanced age, a worsening of the myoclonus is possible as well as mild ataxia and difficult walking. In particular, a long-term observation study on 14 patients from 3 different families linked to 2p11.1-q12.2 showed evidence for an age-dependent progression and the presence of neuropsychiatric and neuropsychological dysfunction in these patients (Coppola et al 2011). Cortical tremor significantly worsened, and amplitudes of giant somatosensory evoked potential significantly increased with age even in Japanese patients, supporting progressive increase of cortical hyperexcitability and a progression with aging or over generation in such patients (Hitomi et al 2014).

Clinical vignette
A 68-year-old man had a family history of hand tremors and upper-limb jerks that began at 20 years of age and for which he had been treated for several years with propranolol (20 mg/day). Tremors consisted of continuous, rhythmic, distal fine twitches at the hands, enhanced by emotion or fatigue. Daily activities were not significantly disturbed except for some difficulties in skilful actions, such as writing, buttoning, and picking up small objects. Other than the tremor, the patient suffered from distal arrhythmic, mainly postural, myoclonic jerks, especially in the upper limbs.

The patient experienced 3 tonic-clonic seizures in adult age, precipitated by sleep-deprivation or emotional stress. Therefore, he was treated for many years with valproic acid (1000 mg/day) with partial benefit. The patient had no signs of cognitive impairment and had normal neurologic examination, except for hand myoclonus. His EEG revealed mild generalized paroxysmal abnormalities and photoparoxysmal response. Detailed electrophysiological investigations (jerk-locked averaging, giant somatosensory-evoked potentials, and long-loop reflex I) were consistent with the presence of cortical reflex myoclonus. Conventional brain MRI was unremarkable, but 1H-MR spectroscopy showed abnormal spectral choline peak in the right cerebellar hemisphere. Add-on with levetiracetam (1000 mg/day) improved myoclonus and EEG abnormalities.

### Biological basis

**Etiology and pathogenesis**

**Genetics.** The inheritance pattern of benign adult familial myoclonic epilepsy is compatible with an autosomal dominant trait. Males and females are equally affected. An affected person has, on average, an equal number of affected and normal children, normal children of an affected parent have normal children, and affected patients occur every generation. Japanese authors reported clinical anticipation in families with benign adult familial myoclonus epilepsy (BAFME), with a higher degree of clinical anticipation associated with maternal transmission, suggesting that the disease is associated with unstable expanding repeats (Hitomi et al 2013; Hitomi et al 2014).

Genetic studies of worldwide available families have revealed clinical and genetic heterogeneity with at least 4 loci, including 8q24 (FCMTE1), 2p11.1-q12.2 (FCMTE2), 5p15.31-p15.1 (FCMTE3), and 3q26.32-3q28 (FCMTE4). By linkage analysis in a large Japanese kindred with benign adult familial myoclonic epilepsy, Mikami and associates assigned the gene responsible for this disorder to the distal portion of 8q (Mikami et al 1999). A maximum 2-point LOD score of 4.31 with D8S555 was obtained at a recombination fraction of 0.0; the maximum multipoint LOD score was 5.42 for the interval between D8S555 and D8S1779. Mikami and associates also assigned a cytogenetic localization to 8q23.3-q24.11 (Mikami et al 1999). To date, 1 large pedigree and 4 small pedigrees originating from Japan were localized on this FCMTE1 locus (Mori et al 2011). This FCMTE1 locus was also confirmed in a Chinese family, and the mapped genomic region was narrowed into an 18.4 Mb region at 8q22.3-q24.13, but whole genome scan failed to identify the causative gene (Cen et al unpublished). The FCMTE1 locus was confirmed also in a Chinese family, including 12 living affected members. The mapped genomic region was narrowed into an 18.4 Mb region at 8q22.3-q24.13, but whole genome scan failed to identify the causative gene (Cen et al 2015).

In non-Japanese benign adult familial myoclonic epilepsy patients, Labauge and colleagues reported in a pedigree in Spain absence of linkage to 8q24 (Labauge et al 2002). Subsequently, Guerrini and colleagues disclosed negative linkage to 8q24 but positive linkage to 2p11.1-q12.2 in 8 patients in Italy with autosomal dominant cortical myoclonus and epilepsy (Guerrini et al 2001). However, the phenotype was not identical with benign adult familial myoclonic epilepsy as the affected patients showed complex partial seizures of frontotemporal origin along with generalized seizures. Linkage analysis in a Dutch family presenting with familial cortical tremor with epilepsy accompanying cognitive deterioration excluded linkage to chromosome 8q3.3-q24 (van Rootselaar et al 2002). De Falco and coworkers described 2 benign adult familial myoclonic epilepsy families showing linkage to the same chromosomal region and suggested allelism with autosomal dominant cortical myoclonus and epilepsy, suggesting genetic heterogeneity (de Falco et al 2003). Locus heterogeneity in benign adult familial myoclonic epilepsy and the related disorder is present, although in most families with classical signs and symptoms, the disorders appear to result from a single gene on chromosome 8q (Uyama et al 2005).

In 2004, Striano and colleagues presented a family pedigree in Italy indicating an autosomal dominant inheritance characterized by cortical tremor, myoclonic jerks, and generalized seizures with nonprogressive course (Striano et al 2004). The family showed linkage to chromosome 2p11.1-2q12.2 (lod score value=1.55). This observation would confirm that benign adult familial myoclonic epilepsy is a genetically heterogeneous condition, with Japanese families
Electrophysiological study confirmed the presence of cortical reflex myoclonus in all affected members, and giant somatosensory-evoked potentials and enhanced long loop reflex were found in 3 presymptomatic members who manifested hand tremor and myoclonus in the upper limbs after 1.5 years of follow-up. Genetic study revealed a significant linkage on chromosome 2p (maximum lod score=5.9).

Striano and colleagues also reviewed a familial case presenting autosomal cortical tremor, myoclonus, and epilepsy (Striano et al 2005b). They analyzed the phenotypic differences between the pedigrees according to the genetic acquisitions. They concluded that benign adult familial myoclonic epilepsy, familial adult myoclonic epilepsy, familial essential myoclonus and epilepsy, familial cortical tremor epilepsy, and autosomal dominant cortical myoclonus and epilepsy are the same clinical entity even if genetically heterogeneous. A founder effect may possibly explain the high frequency of families coming from the same topographic area of Japan and Southern Italy (Uyama et al 2005; Madia et al 2008; Mori et al 2011). In 2013, a genetic study in a large Italian family allowed to narrow the critical interval to a 10.4 Mb segment, which included 59 genes, at the FCMT2 locus on chromosome 2p11.1-2q12.2. Haplotype analysis confirmed a founder haplotype identical to that previously observed in families from the same geographic area (Licchetta et al 2013). Notably, an in-frame insertion/deletion in the alpha2-adrenergic receptor subtype B gene (ADRA2B), encoding the alpha2-adrenergic receptor subtype B, has been reported in 2 apparently unrelated ADCMC pedigrees of Italian origin (De Fusco et al 2014). This mutation alters several conserved residues of the third intracellular (3i) loop and alters the binding with the scaffolding protein called spinophilin on neurotransmitter activation, thus, increasing the epinephrine-stimulated calcium signaling. However, ADRA2B is not the major gene associated to the families linked to chromosome 2p11.1-2q12.2.

The efforts to identify potential causal variants at 2p11.1-2q12.2 have, so far, been unsuccessful by linkage analysis. An international effort on a FAME cohort of 10 European families and 1 Australian/New Zealander family has narrowed the FAME2 locus to a 9.78 megabase interval within 2p11.2-2q11.2, confirming the founder effect in 4 Italian families and allelic heterogeneity with at least 4 distinct founders responsible for FAME at the FAME2 locus (Henden et al 2016).

Additional disease loci have been mapped on 5p15.31-p15 and 3q26.32-q28 in single families of French, Thai, and Chinese origin (Depienne et al 2010; Yeetong et al 2013; Li et al 2014; Liu et al 2015).

Stogmann and colleagues described a consanguineous Egyptian family with an autosomal recessively inherited condition featuring focal epilepsy, neuropsychiatric features, borderline cognitive level, and myoclonus (Stogmann et al 2013). Exome sequencing in this family revealed a homozygous deletion (c.503_503delG) leading to a frameshift in the coding region of CNTN2 and segregating in the 5 affected family members. The gene CNTN2 encodes for contactin 2, a glycosylphosphatidylinositol-anchored neuronal membrane protein, which is necessary to maintain voltage-gated potassium channels at the juxtaparanodal region. Given the severity of the mutation and the function of the protein, the authors considered this mutation the most likely cause of the clinical phenotype in this family (Stogmann et al 2013). However, this family showed clinical, electrophysiological, and neuropathological findings, which were only partly overlapping with the criteria for familial cortical tremor, myoclonus, and epilepsy (Striano et al 2013b).

Martí-Massó and colleagues identified mutations in the ACMSD gene, which encodes for a critical enzyme of the kynurenine pathway of the tryptophan metabolism, in a large atypical family featuring cortical myoclonic tremor with epilepsy and associated parkinsonism (Martí-Massó et al 2013).

Pathophysiology. The pathophysiological and biochemical bases of this condition remain speculative. Both clinical and electrophysiological features of the syndrome suggest a cortical hyperexcitability, which can be the result of a decreased cortical inhibition by the cerebellum via its cerebello-thalamo-cortical projections (Striano et al 2005; Striano et al 2013a). Sporadic postmortem histological studies support the evidence of cerebellar pathology in these patients, and a few imaging investigations suggest involvement of the cerebellum and its projection areas in these patients (van Rootselaar et al 2002; Uyama et al 2005). MRI spectroscopy demonstrates elevated choline/creatine ratio in the cerebellum cortex of patients compared with controls (Striano et al 2009; Long et al 2015). A MR diffusion tensor imaging study compared cerebellar fiber density between patients with familial cortical myoclonic tremor with epilepsy (FCMTE) and essential tremor (ET), and it revealed significantly decreased mean fractional anisotropy (ie, microstructural damage of the cerebellar white matter) in familial cortical myoclonic tremor patients (Buijink et al 2013). Finally, resting-state functional magnetic resonance imaging (fMRI) revealed significant differences in the
bilateral frontal lobe and fusiform gyrus correlated with duration of tremor in familial cortical myoclonic tremor patients (Wang et al 2015).

**Epidemiology**

Epidemiological features of benign adult familial myoclonic epilepsy are uncertain. Uyama and colleagues reported 54 patients in 7 families, estimating a prevalence of approximately 1:35,000 based on their observation in Kumamoto Prefecture (Uyama et al 2005).

A unique south Indian community including 241 patients with ADCME belonging to 48 families has been described (Mahadevan et al 2016). The 48 families are domiciled in 2 southern districts of Tamilnadu, India, which belongs to a community called “Nadar,” whose nativity is confined to these southern districts, with reported unique genetic characteristics. This is the largest single report on ADCME worldwide.

**Differential diagnosis**

Myoclonus is an important symptom of many neurologic disorders. When myoclonus is combined with generalized tonic-clonic seizures in adults, especially when familial hereditary trait is present, progressive myoclonus epilepsies must be considered first (Marseille Consensus Group 1990). Progressive myoclonus epilepsies are characterized not only by myoclonus but also by progressive neurologic complications, such as dementia and cerebellar ataxia. In contrast, patients with benign adult familial myoclonic epilepsy demonstrate myoclonus and generalized tonic-clonic seizures without early signs of dementia and cerebellar ataxia.

The majority of myoclonus epilepsies are generally progressive, causing dementia, cerebellar ataxia, or other extrapyramidal symptoms in addition to myoclonus and seizures. To the contrary, benign adult familial myoclonic epilepsy shows myoclonus as a primary symptom and is nonprogressive without accompanying cerebellar ataxia and mental deterioration. Benign adult familial myoclonic epilepsy is transmitted by an autosomal dominant trait with a high rate of penetrance, whereas a number of progressive myoclonus epilepsies show autosomal recessive inheritance. There are a limited number of progressive myoclonus epilepsies transmitted by autosomal dominant inheritance, such as dentatorubro-pallidoluysian atrophy, May-White syndrome, and progressive myoclonus epilepsy with lipoma, among which dentatorubro-pallidoluysian atrophy is most frequently found in Japan.

In dentatorubro-pallidoluysian atrophy, the onset age varies from the first decade to the sixth. In affected patients with earlier onset, myoclonus and mental deterioration predominate, whereas in patients whose initial signs develop after middle age, cerebellar ataxia and choreoathetosis predominate, but myoclonus and dementia are rather mild. The diagnosis can be confirmed by postmortem findings of atrophic features in the dentatorubro-pallidoluysian system.

In myoclonus epilepsy with ragged-red fibers, myoclonus, cerebellar ataxia, and mitochondrial myopathy are the triad of this condition with maternal transmission. The serum lactate and pyruvic levels are usually high, and the diagnosis can be confirmed by muscle biopsy.

Differentiation of benign adult familial myoclonic epilepsy from juvenile myoclonic epilepsy is not difficult because of their clinical features. In juvenile myoclonic epilepsy, myoclonic jerks occur frequently and often culminate in generalized tonic-clonic seizures. In benign adult familial myoclonic epilepsy, myoclonus persists for years, but the occurrence of generalized tonic-clonic seizures is rare, occurring only several times in a lifetime. In juvenile myoclonic epilepsy, the seizures are prone to occur on awakening. Such a circadian trait is not seen in benign adult familial myoclonic epilepsy. The onset age of benign adult familial myoclonic epilepsy is from the third to the fifth decade, whereas the peak age of onset of juvenile myoclonic epilepsy is in the second decade. An autosomal dominant inheritance is not often seen in juvenile myoclonic epilepsy. Autosomal dominant cortical myoclonus and epilepsy may show complex partial seizures, which are not seen in benign adult familial myoclonic epilepsy, and the results of linkage analysis are different from those found in benign adult familial myoclonic epilepsy.

**Diagnostic workup**

No hematological or biochemical findings specific to benign adult familial myoclonic epilepsy have been found. Electrophysiological investigations including EEG, somatosensory-evoked potential, C-reflex, and jerk-locked back averaging are essential to confirm the cortical origin of myoclonus. However, some of these electrophysiological features can be masked by antiepileptic treatments (Striano et al 2005b). The EEG background activity is usually
normal or slightly slowed in the slower alpha band. Generalized paroxysmal abnormalities and photoparoxysmal response are frequently found in patients not receiving therapy (Striano et al 2005; Uyama et al 2005). Focal paroxysmal activity can occur in some patients in addition to generalized EEG abnormalities (Guerrini et al 2001). Polymyographic recording helps to differentiate tremor and myoclonus. The EMG pattern is consistent with irregular, arrhythmic or semi-rhythmic and high-frequency (around 10/s) myoclonic jerks. EMG bursts last about 50 ms and are usually synchronous between agonist and antagonist muscles, not showing the regular agonist/antagonist alternance as in essential tremor. Jerk-locked averaging analysis commonly discloses a positive-negative, biphasic, premyoclonic spike or a more complex pattern of wavelets related to myoclonus on the contralateral sensorimotor regions. The evaluation of somatosensory-evoked potentials and of long-loop reflex I may show an enlargement of cortical components (P25-N33 amplitude larger than 8.5 to 15 µV) and enhanced long-latency (40 ms) C reflex response evoked by stimulation of the peripheral nerve. A reduction of the resting motor threshold intensity and the post-motor-evoked potential silent period has been reported in few patients evaluated by transcranial magnetic stimulation, indicating that central motor inhibitory mechanisms are impaired in these cases (Guerrini et al 2001). MRI study is usually normal, even if minor, aspecific abnormalities (eg, mild enlargement of the subarachnoid spaces of the lateral ventricles) are sometimes reported. An MRI spectroscopy study demonstrated elevated choline/creatine ratio in the cerebellum cortex of patients compared with controls (Striano et al 2009; Long et al 2015).

**Management**

Cortical tremor is not responsive to alcohol, levodopa, or carbidopa but improves with antiepileptic drugs (Ikeda et al 1990; Striano et al 2005; Uyama et al 2005). Valproate, levetiracetam, and benzodiazepines produce the most benefit on cortical tremor and myoclonus as combining both antiepileptic and antmyoclonic activity. The effect of phenobarbital and primidone to control myoclonus is limited. As for other idiopathic generalized epilepsies, some antiepileptic drugs may precipitate myoclonic status. In these cases, a correct diagnosis and prompt discontinuation of the drug may reverse a potentially severe, life-threatening condition (Striano et al 2007). In advanced age, worsening of the myoclonus is possible, as well as difficulty in walking and mild ataxia (Coppola et al 2011; Striano and Zara 2016). It is advisable for patients with photosensitivity to wear sunglasses in sunlight or to avoid places where light is reflected on the sea or snow.

**Special considerations**

**Pregnancy**

The U.S. Food and Drug Administration (FDA) is advising health care professionals and women that the antiseizure medication, valproate sodium and related products—valproic acid and divalproex sodium, are contraindicated and should not be taken by pregnant women due to the evidence that these medications can cause decreased IQ scores in children whose mothers took them while pregnant. More information can be accessed at http://www.fda.gov/Drugs/DrugSafety/ucm350684.htm.

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**References especially recommended by the author or editor for general reading.

**Former authors**

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**ICD codes**

ICD-9:
Myoclonus: 333.2

ICD-10:
Myoclonus: G25.3
OMIM numbers

Epilepsy, familial adult myoclonic, 1: 601068
Epilepsy, familial adult myoclonic, 2: 607876
Epilepsy, familial adult myoclonic, 3: 613608

Profile

Age range of presentation

06-12 years
13-18 years
19-44 years
45-64 years

Sex preponderance

male=female

Family history

family history may be obtained

Heredity

autosomal dominant

Population groups selectively affected

European
Japanese

Occupation groups selectively affected

None selectively affected

Differential diagnosis list

progressive myoclonus epilepsies
juvenile myoclonic epilepsy

Other topics to consider

Autosomal dominant inherited ataxias
Myoclonic status epilepticus
Myoclonus
Valproic acid