

Progressive myoclonus epilepsy (PME) is a group of conditions involving the central nervous system and representing more than a dozen different diseases. These diseases share certain features, including a worsening of symptoms over time and the presence of both muscle contractions (myoclonus) and seizures (epilepsy). Patients may have more than one type of seizure, such as petit mal or grand mal. PME is progressive, but the rate of progression may be quick or slow, depending on the underlying disease. Progressive myoclonus epilepsy is diagnosed by clinical findings and Electroencephalogram (EEG) results. Molecular genetic testing is available for genes associated with EPM1, EPM2A, and for some of the genes associated with other types of PME.