

## MERRF Syndrome

MERRF Syndrome is a mitochondrial disease causing oxidative phosphorylation dysfunction that most often presents in young adults. Patients present with myoclonic epilepsy, dementia, and ataxia. On histopathology of muscle, ragged red fibers are commonly found.



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### Characteristics

#### Mitochondrial Disease

##### Damaged Mitochondria

MERRF occurs from a defect in mitochondrial genes that leads to mitochondrial dysfunction. These are either sporadic or germline mutations, and are maternally transmitted.

#### Oxidative Phosphorylation Dysfunction

##### Dysfunctioning Oxidative-Ox with Pi badge

This is a key pathway which uses a transmembrane proton pump gradient to generate ATP. When this system is impaired, such as in MERRF, cells are unable to produce adequate ATP.

#### Young Adults

##### Young Adult

This disease usually presents in young adults.

### Clinical Findings

#### Myoclonic Epilepsy

##### Mayo-clown Caesar

Myoclonic epilepsy is characterized abnormal jerks involving a muscle or groups of muscles. This is due to abnormal electrical activity in the brain and is a form of seizure. Patients experiencing myoclonic epilepsy remain conscious.

#### Dementia

##### Demented-D-man

Dementia is neurocognitive symptom involving memory loss, and is a long term complication of MERRF.

#### Ataxia

##### A-taxi

Ataxia in patients with MERRF leads to difficulties with balance and ambulation.<br>

#### Ragged Red Fibers

##### Ragged Red Fibers Carpet

Muscle biopsies stained with Masson's Trichrome stain will reveal ragged red fibers surrounding the muscle fibers. These fibers are made up of aggregates of dead mitochondria.<br>