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.

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.

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