

MITO PROFILE

MERRF INFORMATION

PROVIDED BY THE UNITED MITOCHONDRIAL DISEASE FOUNDATION

MERRF- Myoclonic Epilepsy associated with Ragged Red Fibers (synonyms- MERRF Syndrome and Fukuhara Syndrome

MERRF is a progressive multi-system syndrome usually beginning in childhood, but onset may occur in adulthood. The rate of progression varies widely. Onset and extent of symptoms can differ among affected siblings.

The classic features of MERRF include:

- Myoclonus (brief, sudden, twitching muscle spasms) – the most characteristic symptom
- Epileptic seizures
- Ataxia (impaired coordination)
- Ragged-red fibers (a characteristic microscopic abnormality observed in muscle biopsy of patients with MERRF and other mitochondrial disorders)

Additional symptoms may include: hearing loss, lactic acidosis (elevated lactic acid level in the blood), short stature, exercise intolerance, dementia, cardiac defects, eye abnormalities, and speech impairment.

Although a few cases of MERRF are sporadic, most cases are maternally inherited due to a mutation within the mitochondria. The most common MERRF mutation is A8344G, which accounted for over 80% of the cases (GeneReview article). Four other mitochondrial DNA mutations have been reported to cause MERRF. While a mother will transmit her MERRF mutation to all of her offspring, some may never display symptoms.

As with all mitochondrial disorders, there is no cure for MERRF. Therapies may include coenzyme Q10, L-carnitine, and various vitamins, often in a “cocktail” combination. Management of seizures usually requires anticonvulsant drugs. Medications for control of other symptoms may also be necessary.

The prognosis for MERRF varies widely depending on age of onset, type and severity of symptoms, organs involved, and other factors.

RESOURCES MERRF INFORMATION

MERRF by Salvatore DiMauro, MD, and Michio Hirano, MD, through *GENEReviews* <http://www.geneclinics.org/profiles/merrf/>

MERRF - online OMIM article by same title. Available through UMDF website www.umdf.org, then *Mito Info*, then *For patients and families*, then *disease descriptions*. Click on *MERRF* and then *links* for OMIM article

MERRF Syndrome – on WebMD

<http://www.webmd.com/hw/epilepsy/nord965.asp> or on NORD website http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=MERRF%20Syndrome



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8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239
Phone: 412-793-8077
www.umdf.org