Condition: MERFF (myoclonic epilepsy with ragged-red fibers)

Inheritance:
Mitochondrial (maternal transmission).

Genetic etiology:
Mutation in *MT-TK* gene encoding mitochondrial tRNA lysine.

Frequency:
Approximately 1/400,000.

Clinical features:
MERRF is a highly variable disorder that affects energy metabolism in multiple tissues. Onset is most commonly in childhood, with myoclonus, generalized seizures, ataxia, weakness, and dementia.

Management:
Supportive care; administration of coenzyme Q10 and L-carnitine.

Genetic counseling:
Follows pattern of maternal transmission, but with widely variable expression due to heteroplasmy. Molecular genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.