Genetic counseling for mitochondrial disorders

Mitochondrial disorders are being diagnosed wilh increased frequency, and present clinicians and genetic counselors wilh a multitude of issues to address. Mitochondrial disorders generally result in deficient energy production or availability. Mitochondrial disorders arc typically thought of as maternally inherited, but all modes of inheritance have been described and need to be considered during the genetic evaluation. The counseling issues involved with mitochondrial disorders thai are inherited in a Mendclian fashion are similar to non-mitochondrial Mendelian disorders. The maternally inherited mitochondrial disorders present the genetics team with a variety of unique issues to address. Mitochondrial disorders can affect virtually all organs systems. Most commonly the central nervous, cardiac, muscular and ophthalmologic systems are involved. When suspicious of a mitochondrial disorder it is necessary to determine the most likely mode(s) of inheritance, explain difficult genetic concepts and to provide psychological support. While these roles are typical for a genetic counselor, mitochondrial disorders do present a unique set of challenges. The genetic counselor must obtain a very targeted pedigree with special attention paid to the "soft signs" in family members, such as migraines, seizures, mental retardation, gastrointestinal complaints, chronic fatigue and weakness. Explanations of difficult concepts such as heteroplasmy, oxidative phosphorylation, nuclear DNA, mitochondrial DNA, and nuclcar-mitochondrial gene interactions are unique Providing psychological support can be a daunting task, even to an experienced learn. When one makes a diagnosis of a maternally inherited mitochondrial disease in a proband, one usually also makes the same molecular diagnosis in the mother and potentially other maternal-line relatives. In many cases prognosis varies considerably, thus counseling must by necessity be vague as to the prognosis Frequently there is no reliable prognostic indicator that can be used to predict future health concents Like with other genetic disorders the diagnosis is often accompanied by feelings of grief, despair and guilt. While those feelings are not specific to maternal line inhertance, the extremely high recurrence risk in maternal line disorders is specific to this type of inheritance. Reliable prenatal testing is not available for many mitochondrial disorders, and reproductive options and treatment are limited. In summary, mitochondrial disorders, especially those inherited maternally present many unique and difficult issues for the genetic counselor.

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