Position Statement on Carrier Testing for Canavan Disease

Canavan Disease is a serious incurable genetic disorder which causes mental retardation and is often fatal in childhood. It is cased by a genetic abnormality which produces a deficiency of the enzyme aspartoacylase and is inherited in an autosomal recessive pattern. Like Tay-Sachs Disease, Canavan Disease occurs at a higher frequency in individuals with Ashkenazi Jewish ancestry, estimated to be 1/40. The screening test to identify individuals who are carriers is a DNA-based test that is performed by only a few specialized laboratories.

If both reproductive partners have an Ashkenazi Jewish background, we recommend that carrier testing for Canavan Disease be offered before pregnancy. If only one partner is of Ashkenazi Jewish descent, she/he should be offered carrier testing and the couple should be counseled regarding the limitations and benefits of carrier testing and prenatal diagnosis for this situation.

If a family member is affected, the proband's mutation(s) should be defined and the relatives should be offered screening for this mutation(s).

This screening could be combined with screening for Tay-Sachs Disease as both disorders are more common in the same ethnic group.

This guideline is designed primarily as an educational resource for medical geneticists and other health care providers to help them provide quality medical genetic services. Adherence to this guideline does not necessarily assure a successful medical outcome. This guideline should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the geneticist should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from this guideline.

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