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Family History of Learning Disabilities and/or Mental Retardation

Mental retardation is thought to affect approximately 2-3% of the population. In many cases, the cause(s) of mental retardation remains unknown. Unless a specific diagnosis or cause for mental retardation in an individual is known, a precise recurrence risk for other family members is not possible. The best way to assess the risk of mental retardation for a pregnancy is through a thorough genetic evaluation of the individual with mental retardation.

There are various underlying causes of mental retardation and learning disabilities including specific genetic abnormalities and genetic syndromes, environmental causes, or multifactorial conditions (when multiple factors, both genetic and/or environmental, occur together and result in mental retardation).

Some environmental causes for mental retardation may include, but are not limited to the following: maternal infection during pregnancy, prenatal exposure to certain medications or drugs, prematurity, delivery trauma, and brain injury. These causes are not hereditary, and the recurrence risk for other relatives is not increased above the general population risk as long as these environmental triggers are not present.

Other forms of mental retardation are genetic. Chromosome abnormalities often lead to mental retardation, frequently in association with other physical abnormalities. Some chromosome abnormalities are sporadic, such as most cases of Down syndrome, while other types of chromosome anomalies are hereditary and can be inherited through an unaffected parent. A blood chromosome analysis is available if a hereditary chromosome abnormality is a known or likely cause for mental retardation in a family. The results from a chromosome analysis are available in approximately 10-14 days. A normal chromosome analysis will only rule out certain chromosome disorders and cannot guarantee a normal pregnancy outcome.

Another common cause of hereditary learning disabilities and mental retardation is fragile X syndrome. This syndrome is a genetic disorder that results in mental deficits, developmental and language delays, and behavioral problems. As many as 1 in 4000 males and 1 in 8000 females have this syndrome. In the general population, approximately 1 in 259 women are fragile X carriers. Preliminary studies indicate that in women with a positive family history of learning disabilities and mental retardation, approximately 1 in 40 have been found to be fragile X carriers. If fragile X syndrome is confirmed or suspected in a family, carrier testing is available and results take approximately two weeks. A normal fragile X carrier test will not rule out other possible causes of mental retardation.

There are numerous other genetic conditions associated with mental retardation which may carry an increased risk for mental retardation in family members. The specific recurrence risk for a related pregnancy depends on the diagnosis and inheritance pattern for that condition. Some genetic disorders, including chromosome abnormalities and fragile X syndrome, can be diagnosed prenatally; however, prenatal testing for these conditions is only recommended if such a genetic diagnosis has been confirmed in the family.

Other types of mental retardation are multifactorial, meaning that both genetic and environmental factors contribute to the condition. In these families the risk is greater for first degree relatives (parents, siblings and children) of the affected individual, and is lower for more distant relatives. This type of mental retardation cannot be diagnosed prenatally.

If you have additional questions regarding a family history of mental retardation or learning disabilities, please speak to a genetic counselor.