Fragile X Syndrome

What is Fragile X Syndrome?

It is the most common form of inherited mental retardation and accounts for approximately 40% of cases with X-linked mental retardation. Clinical characteristics include mild learning disabilities to severe mental retardation. Approximately one-third of all children diagnosed with fragile X syndrome also have autism and hyperactivity. Almost all males with full mutations have developmental delay or mental retardation. Approximately 50% of females with a full mutation have IQs in the borderline or mentally retarded range; of the remaining 50%, half have learning disabilities.

Who should be tested?

It is recommended that any person with unexplained mental retardation, developmental delay or autism be tested. The American College of Medical Genetics also recommended carrier testing on the basis of a family history of unexplained mental retardation.

How common is Fragile X Syndrome?

The incidence is 1 in 4,000 males and 1 in 8,000 females. The carrier frequency is 1 in 260 and occurs in all ethnic backgrounds. If the test shows that you are a carrier of fragile X, your partner does not need testing because this disease is inherited only through the woman. If a mother is a carrier, there is a 50% chance to have a child with fragile X syndrome. Therefore, the next step is for you to consider diagnostic testing by amniocentesis or chorionic villi sampling (CVS) to determine if your baby is affected.

Where can I find out more information?

For more information see: www.fragilex.org/ or http://www.cdc.gov/genomics/hugenet/factsheets/FS_FragileX.htm

Spinal Muscular Atrophy (SMA)

What is Spinal Muscular Atrophy (SMA)?

SMA is an autosomal recessive condition that causes progressive degeneration of the lower motor neurons, muscle weakness and, in the most common type, respiratory failure by age two. Muscles responsible for crawling, walking, swallowing and head and neck control are the most severely affected. It is variable in severity and age of onset and does not affect intelligence. There is no cure or treatment.

What is the carrier frequency?

The frequency varies by ethnicity and ranges from 1 in 35 to 1 in 117 in the United States. The incidence is 1 in 6,000 to 10,000

What is the carrier detection rate?

Caucasian: 95%, Ashkenazi Jewish: 90%, African American: 71%, Hispanic: 91%, Asian: 93%.