

Fragile X Syndrome

Fragile X Syndrome is now recognized as the most common inherited cause of mental retardation. The clinical and behavioral changes are not always obvious and can lead to delay in diagnosis. The most common findings are developmental delays, including speech problems and behavioral features such as hyperactivity and hand biting or hand flapping. Mental retardation can range from mild to severe and females tend to be more mildly affected than males. Physical signs in males include long faces, large prominent ears and large testes which are more apparent after childhood. Hagerman et al have put together a screening check list which can be used by the clinician considering referral to genetics for further evaluation. (See following table)

Diagnosis of Fragile X syndrome currently is made by a DNA test. Carrier testing and prenatal diagnosis from CVS is also available to help with genetic counseling.

Early identification of affected individuals can help them obtain optimal speech and language intervention, and behavioral management techniques utilizing positive reinforcement have been shown to diminish negative behaviors.

Genetic counseling and testing referrals can be made to:

Division of Genetics and Dysmorphology
 Department of Pediatrics
 University of New Mexico Health Sciences Center
 ACC 3
 Albuquerque, NM 87131-5311
 Telephone: 505-272-6631

They have outreach clinics in Carlsbad, Las Cruces, Roswell, Zuni, Gallup, Santa Fe, Taos, Shiprock, Farmington, Silver City, Las Vegas and Clovis which can be scheduled through the same number or through the local office of the New Mexico Children's Medical Services (CMS).

	Definitely Not present (score 0)	present in the past (score 1)	Borderline (score 2)
Mental retardation			
Hyperactivity			
Short attention span			
Tactilely defensive			
Hand flapping			
Hand biting			
Poor eye contact			
Perseverative speech			

Hyperextensible MP joints

Large or prominent ears

Large testicles

Simian crease or Sydney line

**Family history of
mental retardation**

Total score: _____

For scoring, consider each item separately and give:

2 points if definitely present

1 point if present in the past or questionable to borderline present

0 if never present

The maximum score is 26 points.

A score of 17 or above means that a Fragile X diagnosis is likely to be positive. A score of 10 or less means that the individual is unlikely to have Fragile X syndrome. Results with scores between 10 and 17 are mixed. Research also demonstrated that the large testicles and tactile defensiveness were the best independent predictors for membership in the Fragile X group.

Fragile X Checklist - Reprinted with permission from Hagerman, R.J. (1987): Fragile X Syndrome, Monograph.
Curr. Probl. Pediatrics 17:621-674.