FACT SHEET

Fragile X

What is Fragile X?

Fragile X is a hereditary/genetic condition that can impact families in many ways. It includes Fragile X Syndrome (FXS), the most common cause of genetically-inherited cognitive impairment ranging from subtle learning disabilities and a normal IQ, to severe cognitive or intellectual challenges including autism or "autistic like" behaviour. Symptoms often include unique physical characteristics, behavioural deficits, and delays in speech and language development.

Fragile X also includes Fragile X-associated Tremor Ataxia Syndrome (FXTAS), a balance, tremor and memory condition that affects some older male carriers of the permutation. Fragile X can also include problems for female carriers such as early menopause, medically referred to as premature ovarian failure (POF).

How is it manifested?

Fragile X is the result of a genetic variation in the X chromosome. Specifically, there is a full mutation on one end of the FMR1 gene. This gene contains information about how to make the protein FMRP. Individuals with Fragile X do not make this protein, because the FMR1 gene is not expressed, due to its being replicated more than 200 times. Males with a full mutation experience severe symptoms of Fragile X. Females with a full mutation will show some symptoms of Fragile X, but will generally not be affected as severely. This gender difference is particularly evident in terms of intellectual impairment, but is less obvious in terms of behaviour and emotional difficulties.

Some individuals have a premutation of the gene, between 55 to 200 repeats, and show few or no symptoms of Fragile X.

Other people are carriers of Fragile X, but do not show any symptoms. In these cases, the FMR1 gene is replicated 45 to 55 times. A man who is a carrier will pass the premutation onto all female children; however male children will not be affected. A woman who is a carrier has a 50% chance of passing the premutated gene onto each child, male or female.

Fragile X can be passed down a family line, through carriers or premutation, for generations before symptoms actually appear in a child.

Common characteristics of Fragile X include:
- long face, large ears and flat feet
- hyperextensible joints
- learning disabilities and intellectual impairment
- decreased attention span
- hyperactivity
- anxiety
- moodiness
- seizures (25% of people with Fragile X experience seizure activity)
- behaviours commonly classified as autistic (difficulty with transitions, repetitive movements, sensitivity to overwhelming environmental stimuli, or self-injurious behaviour)

Who is affected?

Fragile X is the most common type of inherited developmental disorder in the world. Males are affected more frequently and typically more severely than females. Recent estimates indicate that for males with the full mutation the incidence rate is 1:3600 and for females with the full mutation, the incidence rate is 1:4000 to 1:6000.

How is it diagnosed or detected?

A simple genetic test or DNA analysis can determine the presence of the mutated gene responsible for Fragile X. It also provides extremely accurate detection of individuals who are carrying the gene, and can be performed before birth. This type of test is sufficient if testing specifically for Fragile X Syndrome. However, in cases where there is no history of Fragile X and the cause of intellectual impairment is unknown, a comprehensive genetic evaluation should be performed.

Due to the fact that Fragile X can vary greatly among individuals in terms of symptoms and severity, to the point of being very subtle in some females, most individuals with Fragile X are not correctly diagnosed.

Additional Resources:

FRAGILE X RESEARCH FOUNDATION OF CANADA (FXRFC) - www.fragile-x.ca
FXRFC is a national non-profit organization administered by volunteers, most of whom are parents and health professionals. It directly funds promising research aimed at treatment. FXRFC promotes awareness of Fragile X, and publishes a quarterly newsletter and information materials.

NATIONAL FRAGILE X FOUNDATION – www.fragilex.org
The National Fragile X Foundation unites the Fragile X community to enrich lives through educational and emotional support, promote public and professional awareness, and advance research toward improved treatments and a cure for Fragile X.