



Fragile X Syndrome

What Fragile X Syndrome?

Fragile X Syndrome is the most common identifiable inherited cause of intellectual disability (mental handicap). It occurs more in boys than in girls and is associated with varying degrees of intellectual disability.

As it is a genetically inherited condition, when one child in a family is diagnosed Fragile X, there are enormous implications for the parents and brothers and sisters of that child and indeed for many other relatives of the family. Both men and women can be carriers of the syndrome. Until quite recently little was known about Fragile X.

Why is it called Fragile X ?

In 1991 the gene which causes Fragile X was identified. This gene FMR-1 is found at the tip of the x chromosome and shows as a "fragile" site – it looks as though it is broken off but not quite separated. It is this "fragile" site on the x chromosome that gives its name to the Syndrome.

How is Fragile X caused?

The condition is transmitted on the x chromosome, one of the chains of genetic material controlling inherited characteristics. Under a microscope a fragile x chromosome has an abnormal appearance which occurs at the site of the gene FMR-1 which does not function as it should.

Why is it more common in boys than girls?

The x chromosome is one of a pair which decides a child's sex. A boy has one x and one y chromosome while a girl has two x chromosomes. If a girl has one "fragile" x chromosome and one "good" x chromosome, it seems that the "good" chromosome usually over-comes the effect of the "fragile" one. In this way a girl may unknowingly carry and pass fragile x on to the next generation.

A few men with fragile x do not show any problems, but will pass it on to their daughters. Such daughters will usually be of normal intelligence, but their children are at risk of being affected.

How does Fragile X affect a child?

Boys who are affected almost always have some learning difficulties to severe handicap. Girls are often of normal intelligence but up to a third have learning problems which may be mild or moderate, but are occasionally severe. The

behavioural features include short attention span, distractibility, impulsiveness and over-activity. Shyness and social withdrawal are often seen in girls who may have difficulty in making friends.

Many show autistic like features – a dislike of eye contact, difficulty in relaxing to other people, anxiety in social situations, and insistence on familiar routines. Speech and language is usually delayed. There are physical features associated with Fragile X – a long narrow with prominent jaw bones and ears but these are rarely obvious in young children and sometimes not present at all.

How is Fragile X identified?

Fragile X can be diagnosed by a simple blood test, which can be used to identify affected individuals and carriers as well as for pre-natal diagnosis. Families should seek a referral to the National Centre for Medical Genetics, Dublin, from their doctor or specialist.

The importance of diagnosis

The importance of diagnosing the fragile X child is two-fold. Firstly, there are many relatives who are at risk of having affected children who could benefit from genetic counseling, carrier testing and the offer of pre-natal diagnosis.

Secondly, the child concerned can receive specialist help in areas of education, speech and language development and behaviour. Adults with fragile x have specific problems too – the understanding and management of which can be helped once diagnosis is known.

Too few people have heard of fragile X and still many doctors know little about it. Parents, when told they have a fragile x child experience a sense of isolation and ignorance.

The Fragile X Society

The Irish Fragile X Society can offer support from others who know the problems and who have access to information about the syndrome.

For further information, please contact: -

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