Media Release **Nov. 2, 11**

Fragile X Awareness Day in Europe!

For many people, November 18th, is just another Friday on the calendar but for Fragile X families in Europe, this date has significantly more meaning.

November 18th is Fragile X Awareness Day in Europe and for Fragile X families it is an important opportunity to tell people about the syndrome that affects their children.

So what is Fragile X?

Fragile X Syndrome is the most common cause of inherited developmental disabilities worldwide.

It affects around 150,000 people in Europe – many of whom have not been diagnosed – and causes a range of intellectual disabilities and behaviours.

In addition, female carriers may experience infertility and early onset menopause (Fragile X premature ovarian insufficiency known as FXPOI), while some older male carriers may develop a neurological condition (Fragile X tremor/ataxia syndrome known as FXTAS) affecting balance, tremor and memory, similar to Parkinson's disease.

Although almost as widespread as cystic fibrosis and muscular dystrophy, Fragile X is greatly under-recognised and under-diagnosed in Europe.

To learn more about Fragile X, speak to the following people:

Available for interview:

Maria Panza Chair, Irish Fragile X Society

Fragile X Families:

Maria Mother of daughter affected by Fragile X, **Kildare**
Sheila Mother of son affected by with Fragile X, **Wexford**

For more information and to arrange interviews please contact:

Maria Panza, Irish Fragile X Society on 087 657 3089

See also: **Fragile X - The Facts**

Fragile X - The Facts

What is Fragile X?

- Fragile X syndrome is the most common inherited cause of intellectual impairment
- The gene that causes Fragile X is found at the tip of the X chromosome and appears as a 'fragile' site – hence the name Fragile X syndrome.
- Genetic testing is available to diagnose the condition and to identify carriers
- Early intervention by health and educational professionals can assist children affected by Fragile X reach their full potential.

How common is it?

- Fragile X is a greatly under-recognised and under-diagnosed condition as widespread as cystic fibrosis, and second only to Down syndrome in causing intellectual impairment.
- Fragile X affects one in 4,000 males and one in 6,000 females.
- One in 250 women and one in 700 men are carriers of Fragile X.
- Fragile X is found in all populations and ethnic groups.

What causes Fragile X?

- Fragile X syndrome is caused by a large change (full mutation) in the FMR1 gene which is on the X chromosome.
- The FMR1 gene shuts down and fails to produce a protein vital for brain development.
- People who have a small change in their FMR1 gene (called a premutation) are carriers of Fragile X. Men and women affected by Fragile X have a large change (a full mutation) in their FMR1 gene.
- Each child of a female carrier has a 50 per cent chance of inheriting their mother's Fragile X gene and either being a carrier or being affected by the syndrome.
- A father who carries the Fragile X gene will pass it on to all of his daughters, who will be carriers and at risk of having children affected by Fragile X, but to none of his sons.

What are the presenting features of Fragile X?

- Developmental delay/intellectual impairment, ranging from mild learning problems to severe intellectual disability.
- Short attention span, easily distracted, impulsive, often with hyperactivity.
- Behavioural and emotional problems including anxiety in social situations often leading to tantrums, poor eye contact, insistence on familiar routines, repetitive speech, hand flapping or hand biting.
- Speech and language is usually delayed with continuing speech difficulties.
- Around 30 per cent of people with Fragile X also have autism.

- Around 20 per cent of people with Fragile X have epilepsy.
- Physical features include a long narrow face and prominent ears which may not be obvious in young children. It is because of this lack of obvious physical features that diagnosis if often missed or delayed.

Fragile X Carriers

- Until recently, carriers of the Fragile X gene were thought to have no symptoms. Recent research reveals that they can be affected.
- Some female carriers have a condition called FXPOI (Fragile X-associated primary ovarian insufficiency) which affects ovarian function and can lead to infertility and early menopause – some as early as their 20s or 30s.
- Some male carriers, over 50 years of age, develop a condition called FXTAS (Fragile X tremor/ataxia syndrome) which causes tremors, balance and memory problems and is often mis-diagnosed as Parkinson's or dementia.

Testing for Fragile X

- The discovery of the Fragile X gene (FMR1) in 1991 led to the development of reliable DNA tests for diagnosis, carrier testing and prenatal diagnosis.
- As it is a genetically inherited condition, when one child in a family is diagnosed with Fragile X, there are enormous implications for the parents, brothers and sisters of the child and for other relatives.
- In some families, Fragile X has been the cause of intellectual disabilities in relatives through several generations, while in others it can cause problems in only one person.
- Fragile X testing should be considered for any individual with unexplained developmental delay, intellectual impairment or autism; and those with a family history of unexplained intellectual impairment.

Importance of Diagnosis

- Many Fragile X children and adults remain undiagnosed – a situation, which denies them access to the understanding and special care that should follow a diagnosis.
- Children and adults need the diagnosis so that appropriate medical, educational, psychological and social help can be given.
- And for the family – there may be many relatives, both male and female, who may be unknowing carriers of the syndrome and who should be offered genetic counselling.

Is Fragile X treatable?

- There is currently no cure for Fragile X, however there are ways to help with symptoms. The earlier Fragile X is diagnosed, the better the outcome.
- Early intervention therapies (such as speech, occupational, psychological and physiotherapy), appropriate educational strategies and understanding of their behavioural and social difficulties provide real benefits to children and adults with Fragile X.

- Worldwide research is regularly providing increased knowledge.
- Doctors can help diagnose, treat and manage the condition.

For more information about Fragile X, please contact Maria Panza, Irish Fragile X Society 087 657 3089