# Fragile X Syndrome: An introduction

Fragile X Society

# Introduction

This introduction to Fragile X Syndrome is based on previous editions and has been compiled by Jane Oliver, Senior Families and Professionals Advisor and Wendy Bowler (former Family Support Worker, retired) with information provided by the Society's Specialist Advisors. The Society particularly acknowledges the help and advice it received from:

Professor Jeremy Turk (Child and Adolescent Psychiatrist, retired) and Dr Angela Barnicoat (Consultant Clinical Geneticist, Great Ormond Street Hospital for Children, NHS Foundation Trust, London).

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Photography by kind permission of the individuals and families of those whose photographs appear. Thanks also go to the Edith Borthwick School and John Howden for their help with the photographs and to Change for the pictures.

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# Contents

# What is Fragile X ?

Fragile X Syndrome is the most common identifiable cause of inherited learning (sometimes referred to as intellectual) disability. It can cause a wide range of learning, social, emotional and behavioural challenges alongside problems with languages and maintenance of attention.

Fragile X is a family of genetic conditions which includes Fragile X Syndrome and carrier-associated conditions. Carrier-associated conditions do not affect all carriers and are collectively called FXPAC (Fragile X Premutation-Associated Conditions).

## Why is it called Fragile X ?

The gene that causes Fragile X Syndrome is found at the end of the X chromosome and shows as a 'fragile' site on the chromosome when viewed microscopically - it looks as though it is breaking off but is not quite separated. It is this 'fragile' site on the X chromosome that gives its name to the syndrome.

The appearance is, in fact, caused by an abnormally long repeating chemical sequence in the gene, which prevents normal coiling of the DNA in the chromosome. If the sequence is long enough, it interferes with the production of a protein called FMRP, which is important for normal brain development.

*The Fragile X chromosome, in the centre of the picture (left), shows the fragile site at the lower tip in contrast to the other typical chromosomes.*

# What causes Fragile X ?

In 1991 the gene which causes Fragile X was identified. This gene, called FMR1, is present in all of us and it must work properly for typical intellectual development to take place. In people affected by Fragile X, the cause of learning disability is an increase in the size of a length of DNA, otherwise known as an expansion of the CGG repeat, at the beginning of the FMR1 gene and associated chemical modification that prevents it from working properly. This change in the gene is called a mutation.

# What is meant by premutation and full mutation?

Men and women affected by Fragile X Syndrome usually have a large change, called a full mutation, in their FMR1 gene. It is the full mutation that is associated with the wide range of social, emotional, learning and language difficulties alongside problems with maintenance of attention (more detail can be found on pages 11 - 21).

Some people have a small change, called a premutation, in their gene. This small change usually does not stop the gene from working, but does make it 'unstable'. This means that the change is likely to increase in size when it is passed on from one generation to the next, although this only happens when it is passed from a woman to her children.

Males and females with premutations in the FMR1 gene do not usually experience intellectual disability. However, there has been research to show that some experience not only intellectual, or learning, disability but also other difficulties including social, language and attentional difficulties. The term 'carrier' is often used to describe people with a premutation on the gene. It is, also, sometimes used to describe females with full mutations, even though some may not show signs or experience Fragile X-related challenges (see page 22).

# Male and female premutation carriers

Women who are premutation carriers may experience primary ovarian insufficiency (known as FXPOI - Fragile X-Associated Primary Ovarian Insufficiency) and early menopause. Some women may experience menopause five years earlier than would normally be expected, with a small proportion of premutation carriers having their menopause in their 20s or 30s. This can make planning their families difficult and also brings with it the medical problems associated with the menopause. Premutation carrier women who are concerned they are having menopausal symptoms should seek medical advice.

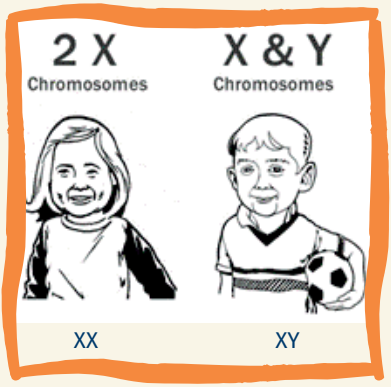
Some premutation carriers have been reported to develop a neurological condition in later years characterised by an intention tremor (a little like Parkinsonism) and balance problems. Later on, these symptoms can be accompanied by problems with working memory, planning skills, and psychological difficulties such as mood swings and anxiety. This is called FXTAS (Fragile X-associated Tremor/Ataxia Syndrome). It occurs more frequently and severely in male carriers with approximately 30 percent of male carriers developing the condition. Women are usually more mildly affected, with around 8 - 16 percent having a diagnosis. The risk of developing FXTAS increases with age, with the average onset of symptoms after the age of 50. Medical advice should be sought if carriers have any concerns.

There is now evidence emerging that some premutation carriers might be affected by other medical and psychological difficulties. Please see our website (www.fragilex.org.uk/carriers-fxpac-resources) for more information about Fragile X Premutation Associated Conditions.

# How is Fragile X passed on?

From our parents we each inherit 46 chromosomes, which decide a great deal about what sort of people we are. Chromosomes are made from DNA, which is organised into patterns, or codes, which we call genes. Genes determine whether we are short or tall, what colour hair we have and many other aspects of ourselves including, to an extent, our intelligence, temperament and behaviour.

The 46 chromosomes are divided into 23 pairs. One of each pair comes from the mother and one from the father. The paired chromosomes are all similar, except for the last pair, called the sex chromosomes. They determine whether a person is female or male. A female has two X chromosomes (XX) and a male has one Y and one X chromosome (XY).




A woman, who is a carrier of Fragile X, has one 'fragile' X and one unaffected X chromosome. She passes just one of her X chromosomes to each of her children. Whether this is the 'fragile' X or the unaffected X is completely random. The father of the child will pass on either an X or a Y chromosome. The child who receives his X chromosome will be a girl and the child who receives his Y chromosome will be a boy.

When the Fragile X gene is passed on from a woman, who has the small change (a premutation), the change in the gene may remain a premutation or may increase to a full mutation.

Therefore in each pregnancy, where a woman has a premutation in her Fragile X gene, the couple could have:

- a boy or a girl who is clear of Fragile X if they inherit her unaffected X chromosome
- a boy or a girl who has the premutation
- a boy who has the full mutation
- a girl who has the full mutation. See section on 'Girls and Women with Fragile X Syndrome' at the end of this book.

The chance of each of these possibilities depends on the exact change in the gene, which the woman carries.

When the Fragile X gene is passed on from a woman who has the large change (a full mutation) the children will inherit the full mutation.

Therefore in each pregnancy, where a woman has the full mutation in her Fragile X gene, the couple could have:

- a boy or a girl who is clear of Fragile X if they inherit her unaffected X chromosome
- a boy who has the full mutation
- a girl who has the full mutation. See section on 'Girls and Women with Fragile X Syndrome' at the end of this booklet.

A man with the premutation has one premutation 'fragile' X chromosome and one Y chromosome. He will pass on his premutation 'fragile' X chromosome to all his daughters and his Y chromosome to all his sons. His partner will pass on one of her two X chromosomes. When the Fragile X gene is passed on from a man, who has the small change (a premutation), it does not increase in size. As his sons will inherit his Y chromosome they cannot inherit Fragile X from him. However, his daughters will all inherit his X chromosome carrying the premutation.

Therefore in each pregnancy, where the father has the premutation in his Fragile X gene, the couple could have:

- a girl who has the premutation
- a boy who is clear of Fragile X.

At present there is little information available about men with Fragile X Syndrome having children. However, should they have a baby, all their sons would be clear of Fragile X Syndrome as they inherit their father's Y chromosome. Their daughters would inherit their father's affected X chromosome. There is some evidence that the change to the gene remains a premutation in the sperm which means all their daughters would be premutation carriers.

# Testing for Fragile X

The discovery of the FMR1 gene has led to the development of reliable DNA tests. These tests are done on a blood sample; they accurately diagnose those who have a full mutation and those who have a premutation.

Prior to the early 1990s the tests that were used for Fragile X were not always accurate or reliable. Families, who are unsure which tests they had, should contact their clinical genetics centre who will be able to tell them if any family members need retesting.

# Genetic advice

Very rarely do parents know anything about Fragile X before they are told that their son or daughter has the syndrome. Once a diagnosis is made within a family, parents and the individual concerned should be referred to a genetics service, who will explain the genetic nature of the syndrome and discuss with them the implications of Fragile X for their immediate and extended family.

Understanding and acknowledging a family's concerns and worries at this time is very important, as is the continued support and care that families should receive from their genetics service.

Sharing with relatives the knowledge that an inherited condition has been found in the family can be a difficult experience. The genetics team can play an important role, looking at the extended family tree to work out which members are at risk of being carriers for Fragile X and discussing with the family how best to inform them.

Relatives are never contacted without the permission of the family and confidentiality is always respected. If you would like to see a geneticist, and have not had a referral, you should ask for one through your family doctor (GP). If you have difficulty in obtaining a referral, please contact the Fragile X Society.

Genetic advice is important for men and women who are carriers of Fragile X. If sought before pregnancy, couples can be informed about their chances of passing on the Fragile X gene and the chances of their child being affected. They will also have time to consider the different reproductive choices. A geneticist can give information about these, their availability and their benefits and limitations.

It is possible to have a test during pregnancy for Fragile X Syndrome. This is usually carried out from around 12 weeks of pregnancy and can pick up whether an embryo has a premutation, a full mutation or is clear of Fragile X. Preimplantation genetic diagnosis (PGD), which involves selecting a Fragile X-free embryo after in vitro fertilisation to plant in the womb of the mother, is also a possibility.

# What exactly does Fragile X Syndrome cause?

Please note: although we describe in this booklet a wide range of characteristics, which may be associated with Fragile X Syndrome, an individual person may only experience some of these.

## Physical characteristics

People with Fragile X Syndrome may have heads that are slightly larger than average, longish faces, largish jaws, large protruding ears, a high arched palate and dental overcrowding. However, these differences are seldom so marked as to give an unusual appearance, and may not be present at all. Most children do not show obvious outward features of Fragile X Syndrome, but subtle changes in appearance are reported, most of which are more obvious in older affected individuals. The most consistent feature seen in small children is a relatively larger head than might be expected; this can alert doctors to the possibility of Fragile X Syndrome in children with learning disabilities.

There is evidence to suggest that there may be a general problem with connective tissue in people with Fragile X Syndrome. This may account for the particular facial features and flat feet, hypotonia (floppy muscle tone), hypermobile ('bendy') joints and lax ligaments, especially in babies and younger boys. Fragile X Syndrome may also be associated with sucking problems in infancy, soft, velvety skin, occasional heart valve problems, 'double-jointedness' and loose bowel movements.

Problems of co-ordination (sometimes referred to as dyspraxia) may be helped by physiotherapy and occupational therapy.

In addition, there may be tendencies to short or long sightedness and squints, and to recurrent hearing difficulties and ear infections. It is important that any hearing or sight problems are treated as effectively and as early as possible, so that being unable to hear or see properly does not further complicate a child's learning disabilities.

In males beyond puberty, and in some younger boys, testicles may be larger than usual.

Some people with Fragile X Syndrome have epilepsy, which can usually be well controlled by medication.

People with Fragile X Syndrome usually enjoy normal general health and can expect to live as long as anybody else.

## Learning disabilities

Fragile X Syndrome results in a wide range of learning disabilities, from mild to, more rarely, severe. This means many children and adults with Fragile X Syndrome will need support to understand the world around them, to learn new skills, to complete some everyday activities and extra time to process information too. The amount of support an individual needs day-to-day depends on whether their learning disability is mild, moderate or severe. Some children will need special education, which could be provided in a mainstream school or at a school for children who have special educational needs and disabilities (SEND).

Learning (sometimes referred to as intellectual) disability occurs in almost all of the boys. Girls are, usually, but not always, less affected by Fragile X than boys, and some girls with a full mutation may be clinically unaffected. This is because a girl has two X chromosomes (one from her mother and one from her father) while a boy has only one (from his mother).

A girl who has a full mutation 'fragile' X, also has an unaffected X and her unaffected X can compensate, in varying degrees, for the effects of her 'fragile' X. This is not the case for a boy because his other sex chromosome is his Y. However, the full range of learning disability and social, language and attentional difficulties can be found in both girls and boys with Fragile X.

For many children, the early developmental milestones, for example, sitting up, crawling and walking, may be normal, although there may be early feeding and sleeping problems. However, some children with Fragile X may show late developmental milestones, for example, in language development or in areas of social development such as smiling and developing friendships late.

Individuals may experience particular problems with mathematics. There may also be difficulties with tasks which involved thinking in 3D, such as map work, finding your way around or following or giving directions. In addition, tasks involving the need to deal with sequences of information (sequential information processing) may present particular difficulties. This difficulty with sequential information processing explains the tendency for people with Fragile X to apparently lag further behind their peers intellectually as they approach adolescence. Difficulty with abstract concepts is also important in this respect.

Many people with Fragile X have difficulty concentrating on the task in hand. It is suggested that this may be because they cannot 'shut out' other things that are happening around them. If there is too much going on it may be confusing for them and provoke anxiety. There may also be problems with co-ordination, including hand-eye co-ordination.

Many individuals with learning disabilities experience these difficulties. On the other hand, research suggests that people with Fragile X have particular profiles of strengths and needs. They may have good long-term memories, especially for things they have seen, such as faces and places. Children and adults with Fragile X Syndrome are good visual learners and enjoy modelling their behaviour on other people.

# Speech and language

Speech and language delay is almost universal and may provide the first clue to diagnosis. The age at which children with Fragile X start to communicate and the rate at which their speech, language and communication skills develop varies considerably.

People with the syndrome sometimes repeat words or phrases rapidly with up and down swings of pitch (litany-like phraseology). They sometimes repeat the last word or phrase spoken to them many times (echolalia) or repeat the end of phrases they have themselves spoken over and over again (palilalia). There is a tendency to skip, rapidly and frequently, from topic to topic while talking. The term 'cluttering' is often used to describe their speech. This consists of the combined effects of speech that is too rapid with poor control of rhythm (dysrhythmia). Words tend to fall over each other with pauses, often in the wrong places, making speech difficult to understand. Individuals also tend to go on and on about one topic of conversation (perseveration).

However, one of the most unusual things about people with Fragile X is that their understanding of speech is often higher than one would expect, given their difficulties in speaking and being understood. Receptive language skills (comprehension) are, generally, better than expressive skills - unless there are difficulties that fall within what is known as the 'autism spectrum', as explained on pages 20 - 21.

# What can help?

An appropriate education is important for all individuals with Fragile X.

**More detail on how to work with them in an educational setting can be found in the Society's book 'An Introductory Guide to Educational Needs and how they can be met'.**

Speech and language therapy evaluation and input are invaluable for people with Fragile X of all ages. An occupational therapy assessment, particularly if there are difficulties managing sensory information (vision, hearing, smell, taste and touch) can also be very helpful.

Schemes such as 'Portage' (a detailed approach with pre-school children, which depends on parents and teachers working closely together) can be helpful in promoting young children's development. When children begin formal schooling, some may need additional support and differentiated work in the mainstream environment and others may do better in some form of special educational provision.

Many individuals with Fragile X are better at 'taking in' what they see rather than what they hear.

Learning to read may be easier for children with Fragile X, if they are helped to recognise whole words, rather than being expected to build them up from the various sounds the letters represent (phonics).

Demonstration rather than detailed spoken instruction may be helpful in other areas of learning, too, as may practical concrete teaching approaches which emphasise an overall view of the task. Visual input in terms of what is being taught as well as in breaking down the order of a task/letting an individual know what is expected of them can be useful.

Teaching strategies which aim at strengthening the ability to focus and resist distractions will be beneficial. So, too, are strategies that avoid direct eye contact (which most people with Fragile X find unpleasant - so-called gaze aversion) and are aimed at reducing anxiety. It is helpful to provide a calm, structured, predictable and focused learning environment without too much going on to distract from the task in hand. It is also useful to have a structured, consistent and predictable routine where surprises and changes are kept to a minimum and to have some visual representation of this like a visual timetable.

Using computers is also helpful, partly because what is being taught can be seen, and partly because it avoids interaction with the teacher, which some pupils with Fragile X find difficult. Also, the individual can determine the rate at which the information is processed and the results of their actions are immediate.

Teaching strategies that focus on strengths tend to work better e.g. individuals with Fragile X Syndrome find processing sequences of information difficult and learn better when presented with the whole task.

> Adults continue to benefit from a predictable routine and strong structure. An environment that is too 'flexible' can cause anxiety so it is important that staff and work schedules remain consistent.

# Behavioural characteristics

Behavioural difficulties can often be an individual's communication of distress/not managing something. Sometimes this can be related to them feeling unwell or being in pain. As an initial response, it is vital to check there are no underlying physical problems causing someone to behave in a way that challenges the people around them.

How all individuals behave is determined not only by the sort of people they are, but also by what is happening around them. However, research shows that groups of individuals with certain syndromes/genetic conditions develop specific behavioural characteristics associated with that syndrome/condition.

People with Fragile X Syndrome are more prone to the following:

- Overactivity - always rushing about, never able to sit still.
- Impulsivity - not able to wait for anything, always having to do things straight away, tending to do things first and think later.
- Inattention - marked concentration problems, inability to stick to one activity or task for any length of time.
- Restlessness - always up and down, out of one's seat.
- Distractibility - inability to maintain concentration.
- Fidgetiness.

All of the above may improve as children grow up and individuals mature. When these tendencies are extreme and occur simultaneously in a variety of settings and with different people, they are called 'attention deficit hyperactivity disorder' (ADHD).

Other behavioural characteristics of those with Fragile X Syndrome include:

- Difficulties coping with changes to normal routines.
- Difficulties coping with things around them being changed (for example where they live, furniture, even their clothes).
- Difficulties coping with transitions (moving from one place or activity to another).
- High levels of anxiety. Anxiety and frustration are potent and common causes of challenging behaviour in people who have Fragile X. Thus in order to understand why an individual is behaving in a particular way, one must look at the underlying anxieties and frustrations and their causes.
- Self-injury - most commonly hand-biting at the base of the thumb or index finger. Often in response to anxiety, frustration, or excitement.
- Hand flapping - again often in response to excitement, anxiety, or frustration.
- People with Fragile X Syndrome often find it difficult to make sense of, and react appropriately to, the information coming to them via their senses. In busy environments, notably noisy/crowded places, they can be overwhelmed by the excessive stimulation; this may cause challenging behaviours, repetitive speech, repetitive behaviours or a 'fight, flight or fright' response. In addition, some individuals may show unusual fixations or intolerances with certain sights, sounds, tastes, smells or textures.

# What can help?

There is a need to understand the behaviours commonly associated with Fragile X Syndrome. Often what appears as challenging behaviour can be a person with Fragile X's way of communicating their worries, fears or concerns, particularly if their ability to communicate using language is limited.

It is important to understand why a person is behaving in a way that challenges and an assessment of the behaviour can help with this. This can be carried out by a learning disability nurse in adult settings or by a teacher (sometimes with help from an Educational Psychologist) in schools. Identifying what has immediately preceded the challenging behaviour (sometimes referred to as the 'antecedent') and what has followed the behaviour ('consequences') often helps to clarify what is triggering the behaviours and possibly maintaining them. However, triggers to behaviours are not always clear. There are occasions when incidents in the day gradually cause anxiety levels to rise and the challenging behaviour is triggered by something a person can usually manage. However, on that occasion, it just tips the balance of endurance levels.

Distraction techniques or having access to known calming activities are often good ways of managing rising anxiety levels.

Individuals often manage change and transition better if they are prepared for them using a matter-of-fact, calm approach. The timing of this preparation can be crucial and will vary according to the individual. A visual representation of the change or transition, shown in advance of it happening, can be helpful.

Sensory overload or sensory difficulties may be helped by sensory integration therapy. Sensory integration programmes are usually devised and monitored by Occupational Therapists with specialist training. An Occupational Therapist with this training may prescribe a set of calming activities/exercises designed to be done on a regular basis. This is known as a sensory diet.

Some individuals may need a referral to a psychologist for help with behaviour management, or access to a Positive Behaviour Support Worker or team.

Professional advice is essential if you are thinking of any of these techniques. It is also important to work in conjunction with individuals' teachers and any other important people in their lives, such as relatives, carers, youth leaders and support workers, to ensure a consistent approach. Medication can be beneficial for some individuals with Fragile X Syndrome, but it is important to ensure that the person is being supported in other ways as well. A multi-professional or multi-disciplinary approach can give a good outcome.

# Social aspects

Most people with Fragile X relate well to others, although they may be shy, socially anxious and find large gatherings of people and noisy, crowded settings more difficult. Most will find direct eye contact very hard and will often avert their gaze, sometimes turning their head or their whole body away from the conversational partner. For people with Fragile X, difficulty in making and receiving eye contact is not a sign of social indifference. They find eye contact uncomfortable in social situations particularly when meeting new people, or when they feel they are the centre of attention. However, most individuals with Fragile X Syndrome enjoy the company of others and want to be part of whatever is going on.

There is a significant minority, who do have problems relating to other people. People affected in this way may:

- have difficulty letting others know what they want or how they feel. They may also have difficulties understanding what other people are trying to get across to them, whether they use words, gestures or other ways of communicating. They may have difficulty understanding other people's feelings and be unable to name or label emotional states.
- tend to be loners. They may find it difficult to get on with other people, prefer to be on their own or be extremely shy or anxious in company. However, some people with Fragile X Syndrome are keen to mix with others but are not aware of the rules around social engagement. This can make them stand out from others.
- be obsessional in their behaviour and insist on sticking to their own routines and habits.

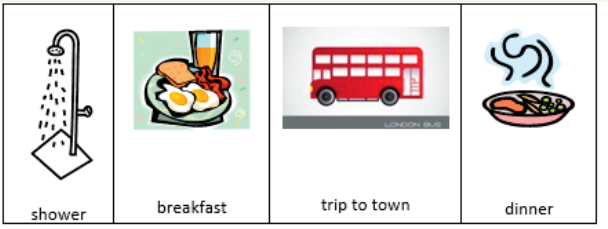
When these disabilities are severe and occur simultaneously, the individual may qualify for a dual diagnosis of having an 'autism spectrum disorder' (ASD) in addition to their having Fragile X Syndrome. This can be associated with any degree of intelligence. A substantial minority of individuals with Fragile X also have ASD. Many more have some of the above features. Particularly common in Fragile X (and much less so in other conditions) is to find someone who is socially aware but also socially anxious; a combination of a likeable, happy, friendly personality with a limited number of autistic-like features. These features include hand-flapping, hand-biting and finding direct eye contact unpleasant and anxiety-provoking. Other features include insistence on routine and a strong dislike of change alongside speech and language problems, including repetitive speech.

# What can help?

Care should be taken not to force eye contact, as this will simply exacerbate anxiety levels with increased risk of challenging behaviour. Standing or sitting alongside or behind the individual can avoid such problems.

The main way of tackling social difficulties involves special teaching approaches, with an emphasis on learning social skills. Role play can be useful as can participating in small social groups, which help to build up confidence and an ability to see things from other people's point of view.

Social stories and other visual structures can be useful aids in terms of preparing an individual for particular events or helping them to conceptualise how to manage a particular situation.



# Girls and women with Fragile X Syndrome

The situation with females is more complex. In varying degrees, the unaffected X chromosome can compensate for the fragile X chromosome. This means that some females with the full mutation have no learning disabilities; others have at least some degree of learning disability with a few experiencing severe difficulties. Typically, females who do experience learning disabilities are more mildly affected than males with Fragile X Syndrome.

Even if they are more mildly affected, girls and women may still have large discrepancies across different subject areas and may show concentration problems with poor working memory, impulsiveness, distractibility and difficulty sticking to tasks. There can be difficulty in organising thoughts, planning ahead, shifting readily from one topic of thought to another and deciding how to tackle a task (sometimes referred to as 'executive function skills'). Some girls and women may also experience problems with co-ordination or over-activity.

Maths and numeracy generally can be particularly difficult subject areas for women and girls with Fragile X Syndrome. Teachers and support workers need to be aware of this.

It is very likely that girls and women with Fragile X Syndrome will have social, emotional and communication difficulties related to:

- Extreme shyness and anxiety in social situations.
- Oversensitivity to perceived rejection or criticism.
- Difficulties picking up social signals (verbal or otherwise) when trying to 'join in', making it difficult for them to make friends despite an eagerness to do so.

For girls and women with Fragile X Syndrome, their learning disabilities/ difficulties and social, emotional and communication difficulties need to be recognised and acknowledged so that appropriate support can be offered. Otherwise, repeated failure to achieve may perpetuate and further increase their social anxiety and low self-esteem. These difficulties may be subtle and therefore often missed or neglected because of their not being so often associated with challenging behaviours.

# Conclusions

Finally, people with Fragile X Syndrome will experience happiness and anger, like everyone else. Many people with Fragile X Syndrome want to be a part of things, as many people do. They will also experience grief and loss. They can be charming, kind and considerate.

Despite any challenges they may experience in expressing their own emotions, people with Fragile X Syndrome are very intuitive about other people's emotions and are extremely likeable with a fantastic sense of humour.

Clearly, people with Fragile X Syndrome have their own strengths, as we all do, and are unique individuals, like everyone else. However, we hope this introductory guide is helpful for families and professionals.

For further information about Fragile X Syndrome , please contact the Fragile X Society. Our contact information can be found on the back cover of this booklet.

# The Society's publications

- Fragile X Syndrome: An Introduction for Families and Healthcare Providers (leaflet)
- An Easy Read Guide to Fragile X Syndrome for Parents
- Fragile X Syndrome: An Introduction (this booklet)
- Fragile X Syndrome: An Introductory Guide to Educational Needs and how they can be met
- FXPOI: Fragile X-Associated Primary Ovarian Insufficiency
- FXTAS: Fragile X-Associated Tremor/Ataxia Syndrome

# Further information about Fragile X

We have a range of information available free to families affected by Fragile X, carers and individual professional enquirers. These include information on topics such as:

- Information for carriers
- Genetics
- Behaviour
- Communication
- Sensory issues
- Girls and women
- Guides to disability benefits
- Adult services and education

We have two accessible information publications:

- An Easy-Read Guide to Fragile X Syndrome for Parents; and
- 'I have Fragile X Syndrome': an easy-read guide.

At the Society, we have two Families and Professionals Advisors, covering adults' and children's enquiries. We would like to encourage families, professionals, carers and people supporting someone with Fragile X Syndrome to get in contact for individual support. Details are on the back of this booklet.

# The Fragile X Society

The Fragile X Society was formed in 1990 and our aims are:

To provide information and practical guidance to support and empower individuals and families living with Fragile X Syndrome and Fragile X Premutation Associated Conditions.

To educate and inform the public and professionals about Fragile X, raising awareness and understanding and improving support for all individuals affected by Fragile X.

To encourage research into all aspects of Fragile X through working with researchers, encouraging the participation of our family members in Fragile X studies and publicising the results.

## Membership of the Society

Family Membership is available to Fragile X families and carers and we welcome, as Associate Members, those with a professional interest in Fragile X.

Membership of the Society is free of charge, and enables you to receive information about our events and our regular e-newsletters. Importantly, becoming a member demonstrates that you are part of our community and therefore helping us to highlight the need for our services and that greater awareness and support for Fragile X Syndrome are required.

You may also wish to follow our social media for regular updates and news; details are on the back cover of this booklet.

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