

Newsletter

Spring 2022

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Welcome!

Dr Kirsten Johnson, Chair, Board of Directors

Welcome from the Board! Our Directors have been busy this year on many fronts, working with Pete and our wonderful staff team to raise awareness, support our members and encourage research into Fragile X.

Most of us have family with Fragile X Syndrome and quite a few of us are carriers. Our first-hand experience of living with Fragile X helps us shape the Society to serve our Fragile X community.



Getting to know each other is key - during the pandemic we've been able to hold online events, webinars and coffee mornings. Now we are moving to face-to-face meetings, and I am so pleased we are able to have our Family Conference at Thomley Activity Centre this year (see opposite). If you haven't been before, you are in for a real treat! Have a look on their website - it is a beautiful and safe environment where our children/adults can roam freely with plenty of breakout space for chatting and getting to know one another. Please do come!

I would like to say a big thank you to all of you who have been fundraising for us this year - we have come through the pandemic with our services intact, and we are grateful for everyone's support. If you aren't a Friend of Fragile X yet, please see p. 21 and think about becoming one, it would really help. We are keeping the conferences and newsletter free, so that everyone can access the information and talks, and meet other families, but if you are able to give a small monthly donation it would really help.

Pete Richardson, Managing Director



First of all, a huge thank you to every one of you, our members, for your continuing support to all of us here at the Fragile X Society and to one another during a very challenging year.

The Fragile X Society exists to support you our members to the best of our ability. So please do continue to update us, share your stories and in doing so help us to grow and develop public and professional knowledge and interest in all things Fragile X.

Despite all the difficulties we faced together, 2021 was a very successful year for the Fragile X Society. Against a background of a worldwide pandemic and the loss of income, badly affecting small charities in the UK, we grew and developed our services, upgraded our website, communications, and social media functions. We have also employed two new part-time staff to help upgrade the information we provide to you, secured additional grants, and continued to keep our helpline operating.

We are particularly grateful to those of you who took part in events and fundraised for us. Thank you! 2022 will focus upon consolidating our success, integrating new staff into their roles, and delivering a well-informed and up-to-date service to you, our members. This next year will undoubtedly bring about new challenges for us all. Collectively, we need to work together to raise awareness of Fragile X and in doing so improve services and facilities for you. This is a big task. We don't underestimate the amount of work that is needed. That said, I'm really encouraged by your support, and we all remain determined to change our world for the better to benefit everyone in the Fragile X community now and for years to come.

Thomley Family Weekend



Join us for our Family Weekend at Thomley on Saturday 18 and Sunday 19 June!

We look forward to welcoming you to our Family Weekend at Thomley! This fun event is a great place to meet and share experiences, as well as attend talks and workshops to learn about Fragile X and practical strategies to help in day-to-day life. This year, sessions will include: medication and trials with Dr Andrew Stanfield; managing anxiety with Dr Hayley Crawford; fathers and Fragile X; 'Freedom to Thrive' project - setting up an inclusive community. A full agenda will be available soon.



Thomley has something for all interests and preferences. We're already counting down the days and can't wait to meet up with other members and swap stories and share similar experiences.



The Thomley Centre is "a place for people of all abilities and disabilities. We provide an exciting site for disabled people, their families, carers and friends. We support people at any stage of their diagnostic journey, whatever their additional needs." You can find out more about Thomley at www.thomley.org.uk.

Spaces are limited and this event is likely to fill up quickly, so make sure to book soon if you wish to join us (we may need to run a waiting list). Find out more on our website at www.fragilex.org.uk/family-weekend-thomley-2022 and register for your free place/s.

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Please note that all information is correct at time of going to press, but may be subject to change

Annual General Meeting



Notice of AGM: Saturday 18 June 2022 at 12pm

The Annual General Meeting (AGM) is an important part of the Fragile X Society's year. At the AGM, we present the financial accounts from the previous year and discuss the recent achievements of the Fragile X Society.

The AGM is also an important opportunity to vote on key issues facing the Fragile X Society, and to find out more about the Society's future plans. An important part of the AGM is the election of Directors. As you may know, we can have up to 15 Directors to lead the Fragile X Society. The majority of our Directors have a personal or family connection to Fragile X. Each year a third resign by rotation and are eligible for re-election. The elections are for three-year terms. We are very grateful to our Directors for giving us their time and expertise to help lead the Society.

Our AGM will take place on Saturday 18 June 2022 at 12pm at Thomley. If you are unable to attend the meeting but would like someone to vote for you, then please complete the proxy form (below) so that someone can vote on your behalf. Please return this form by 16 June at the latest.

Agenda

1. Apologies for absence
2. Minutes of the Annual General Meeting held on 25 September 2021
3. Review of the year (Pete Richardson)
4. To receive the report of the Directors
5. To receive the Independently Examined accounts for the year ending 28 February 2022 and the Auditor's report.
6. To elect up to 5 members to the Board of Directors. One Director, Andy Clarke, resigned in January 2022 and we thank him for his service. Two Directors, Gaia Scerif and Lucia Elghali, have served 3 three-year terms and may not stand for re-election. Two Directors, Marie Williams and Kirsten Johnson, have served three-year terms and will stand for re-election.
7. To appoint auditors
8. Any other business (please contact us in advance of the meeting)



AGM papers

Scan the QR code above
to access the AGM
papers online

Proxy Form

Please complete, delete as appropriate and return by 16 June:

I/We (name/s)..... of (address).....

being a member/members of the above named company, hereby appoint:
(name)..... of (address).....

or failing him/her the Chair of the Meeting, as my/our proxy to vote in my/
our names and on my/our behalf at the Annual General Meeting of the
company to be held on the above date and at any adjournment thereof.

Signed:..... Date:.....

Family Story: Having Fragile X

by Katherine Zwink

I recently joined the Society's Board of Directors and they asked me to tell you a bit about myself.

I did not know I had Fragile X until I was 13. School was tough because I was hopeless at subjects like maths, geography, science and games. I also had lots of difficulties with the other students in my class. I was very shy and nervous with them. I did not understand most of what they were saying and I never knew what to say to them and when I did say something they would all look at me as though I was mad. They would say horrid things and sometimes I would try and stand up for myself and there would be big arguments, which made it all worse. The only person who did understand me was my one-to-one support teacher who was just lovely and she always tried to help me understand why things had gone wrong.



I went on to college and again I had problems with the other students on the foundation course for students with learning difficulties. Most of them did not want to do any work and I think they thought I was a pain because I did. They only wanted to talk about boyfriends. I then did an admin course which I passed except for the maths part but by the time this year ended I could not take any more college.

Then my mum found a different college which did a hairdressing and beauty therapy skills course, which was wonderful because I had always wanted to do beauty therapy but did not have the GCSEs to get on to the NVQ course. I trained as a junior to the hair stylists and learnt how to do manicures and pedicures. I liked the beauty salon best – it was quieter and calmer – and I loved the work. The other students were not all friendly but I was getting better at handling their remarks by ignoring them or walking away. There were also some mature students doing the beauty sessions and they were very friendly. The tutors were very supportive and when I passed this course they allowed me to go on to do the level 2 beauty course which I did and loved every minute. My tutor did not think I could manage level 3 and I agreed with her so I did part time courses in body massage, aromatherapy and Indian head massage instead. Then I tried to get a job in a beauty salon but they always wanted level 3 so I did beauty treatments at home and still have a few regular clients.

I then started to help as a volunteer at a local pre-school which I enjoyed. My supervisor encouraged me to get a qualification and I did the child care and education course. For a time I worked in two pre-schools but sadly, after changes in supervisors, these became unfriendly places to work and I left.



At 26, and at my fifth attempt, I passed my driving test. Yippee!



Family Story: Having Fragile X

Living in the country, as I do, you need a car. And with a car I was able to work as a carer/personal assistant for children with special needs, elderly people and adults with learning disabilities. I really enjoy this work but a big problem with working as a carer/PA is job insecurity – support funding can be suddenly cut by social services and you can find yourself out of work. I still do some caring/support work but a few years ago I started offering ironing and cleaning services to people living in villages near to me. So I am now a self-employed “Ironing Lady” – I collect and deliver back the ironing, clean our local church, village hall and a few homes in our village, and occasionally see beauty therapy clients.



The most important thing I do is keep fit and I love it. It keeps my morale up.



Going to the gym, doing fitness classes and swimming has been great for me. It keeps my morale up. After exercise I feel energized. I do it every day and I feel good. When lockdown began and the gym closed I got an exercise bike and haven't missed a day on it since - see photo! I would love to be a personal trainer and passed level 3 in personal training recently. I hope to find work in a local gym soon.

I live at home with my parents and brother who also has Fragile X, our ancient, rescued dog, Henri, and my cat, Billy.



I am going to the Society's Family Weekend at Thomley in June and it would be lovely to meet up with other girls with Fragile X there. I hope some of you can make it.



Summary of our year

Information and guidance

Supporting individuals and families through their lives

- We achieved our primary objectives for the year, continuing to provide support and information to families affected by Fragile X and professionals working in the field of Fragile X. Feedback on our support has continued to be extremely positive during 2021 - 2022.
- Online support and resources became increasingly important to our community during the Covid pandemic. This year we have continued to develop our online presence and we are proud to now have a large community in our Facebook Group (2732 members) and a steadily growing following on social media including 3008 followers on Twitter and 1513 followers on Instagram.
- Our website attracted just under 40,000 users, with the digital rebrand by Costello Medical occurring mid-way through the year. Whilst the most popular pages remain the introductory page for the syndrome, information about genetic testing, and the association between Fragile X and autism, our home page saw a significant increase in traffic (17%) when compared with the previous year.

Educate and inform

Teaching professionals and the wider public about Fragile X

- Professionals, too, want information so that they understand the needs of children and adults in their care and are informed of appropriate interventions, treatment and therapy. Via our helpline service, we have provided information and support in response to 88 enquiries (generating 322 contacts) from professionals, students and other organisations.
- In collaboration with Cornerstone, and thanks to the Scottish Autism Development Fund, we are able to train professionals via our e-learning course in Fragile X. This year (1 March 2021 - 28 February 2022), 92 professionals were trained via this course.

Improving support and understanding

Supporting and inputting into research

Fragile X Society members (with a direct link to Fragile X) have the option to agree to be contacted about research projects which may be relevant to them. In total, 1547 families (79% of our members) have agreed to support research. Our unique community in the UK is a vital resource for researchers, as there is no central database of individuals diagnosed in the UK.

We are proud to have supported recruitment to seven projects addressing key aspects of Fragile X Syndrome and Fragile X Premutation-Associated Conditions this year. These projects could not take place without the support and participation of our members.

Thank you for being so helpful. It's definitely nice to know there is someone I can discuss things with when experiencing difficulties who truly understands the disability.



2021 - 2022

Finances

Overview of income and expenditure for the year 1 March 2021 - 28 February 2022

Summary 2021 - 22

Total income
£205,521

Direct charitable expenditure
£117,273

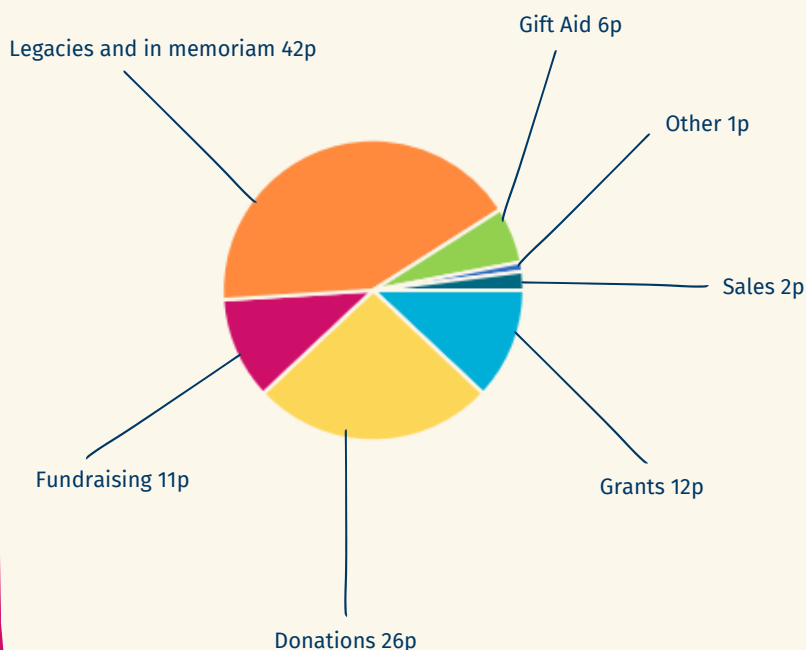
Total expenditure
£127,654

Balance of funds at
28 February 2022
£188,293

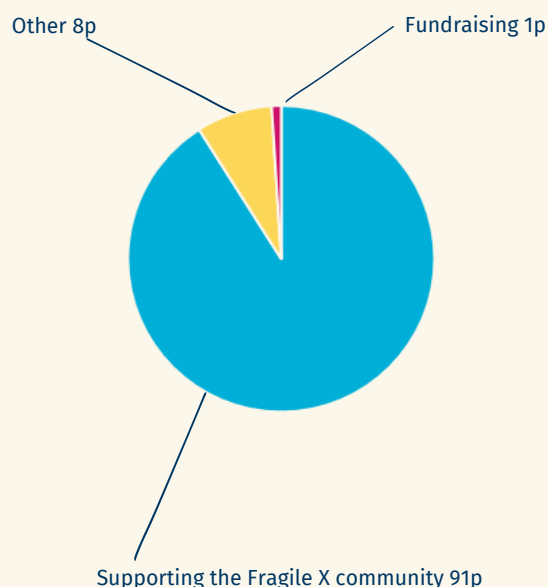
We are very grateful for the generous support of all our members, our donors and grant givers, as well as the Trusts and Foundations that have helped us record a small financial surplus during 2021 - 22.

This is an enormous achievement given the challenges we all faced within the charity sector. We are grateful for all who have supported the Society's 'Friends of Fragile X' and 100+ Club initiatives, which have provided vital funding for our work.

Where each £ came from



Where each £ went



Snippets of Advice: using visual aids

From Caroline Pringle, Families and Professionals Advisor (Child Enquiries)

Visual aids (also known as visual supports) can help your child and give predictability to their day. Your child's school or their speech and language therapist may help or provide you with these resources but as I have access to Widgit symbols I can also support you with making up resources to meet your child's needs. Many children with Fragile X struggle with transitions and due to the changes in their routine their anxiety levels may rise. It's not easy to process the spoken word when you are anxious and visual aids can build up their confidence. I trust the following tips will be useful.

Object of Reference

Objects of reference are used to motivate a child and represent a daily activity e.g. a plate to indicate a meal or a shoe to indicate going for a walk. However, this strategy can be beneficial to children who don't normally use this resource, as it's a method of explaining what's happening next. Putting out a wooden spoon and mixing bowl for cooking or trowel and seeds for gardening is a good motivator and they can take these items to complete the activity with you.



Using symbols

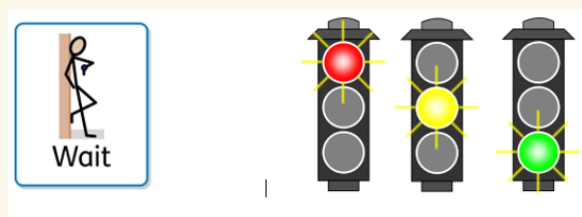


Some of you may have access to photos or symbols such as PECS (picture exchange communication system). You may feel able to draw pictures to help your child. I use Widgit symbols and have given you some examples in this article which may help your child. Some children may still be using first and next boards (also known as first and then boards) so simple symbols will be of help. Others will be able to cope with more details and their visual aids may be in the form of a planner or schedule.

They may be used to a planner at school to act as a timetable of their learning activities, it gives them certainty of the day ahead. Continuing to use a planner before and after the school day, at weekends and school holidays will really help to give them structure.

You may need to limit the number of items on a planner, otherwise it will be overwhelming. For some children just the choice of 2 things will be all they can cope with. Activities/tasks can be cut up separately and blu tack or Velcro put on them so that they can be taken off the list once completed and put into a box or bag.

When activities are going to change to something your child is excited about, I find that the wait sign is useful. Also, a traffic light system of coloured paper/card can be helpful when trying to explain to them an imminent change e.g., amber gives the warning that an activity is about to finish, red the activity has stopped and green for new activity.

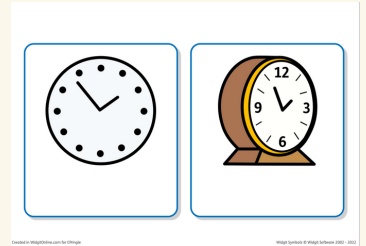


Visual prompts can be used for a variety of activities/reasons; perhaps the use of just one card such as the wait card (previously mentioned) or cards used to support a child to express they need help, or are experiencing sensory overload e.g., finding somewhere too noisy. In addition, a visual aid can be a reminder, such as for things to remember to pack in a school bag and how to stay calm in tricky situations.

For some children, they cope well with the written word, so you may only need to use the occasional symbol. They can use a desk diary, or calendar to write down key things for the day and they might like to tick them off when completed.

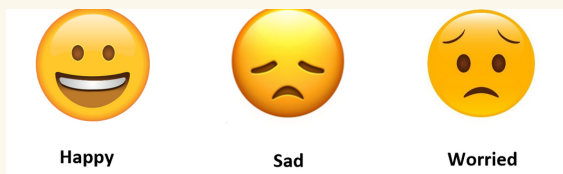
Time

Using symbols to represent the time or duration of an activity can be helpful so your child can see where an activity fits in their day. So often we say to a child in a minute, or later, and this can cause confusion so you might have to use visual representations of time. Perhaps they can cope with looking at a clock or you can use clock face symbols to indicate when a task will finish. A sand timer is another useful resource or showing them the digital time on an iPad, so they know when their screen time has to end.



Feelings and emotions

Children with Fragile X can experience high levels of anxiety and find it difficult to express how they feel. Visual resources such as emotion fans or basic emojis may help you start a conversation with them. You can put these emotions fans onto a stretchy key ring to attach to a belt loop or school bag.



Easy to read information

Easy to read information is a way of presenting material using simple words in short sentences, and with pictures. An example of this is our easy read 'I have Fragile X Syndrome', available on our website: www.fragilex.org.uk/general-resources. Books Beyond Words (booksbeyondwords.co.uk) is another good resource using wordless pictures to allow the reader to tell their own story about a situation. Some books are free to download and other books can be purchased to cover situations such as going to the dentist, making friends and a day on the beach.

Social stories

Social stories can be used to give clear information about people, events, and situations and to introduce a new experience. It puts your child in the situation rather than a general story and is a discussion tool to help them in everyday events. Social stories explain one situation at a time. For some children using photographs or symbols will give more meaning to the story and this will then progress on to them using a written social story, perhaps keeping some symbols. The concept of these stories and a specific format originally came from an educationalist in the States called Carol Gray and was used to help young people with ASD cope in social situations. Please see the two blogs that covers social stories in more detail, available on our website: www.fragilex.org.uk/snippets-of-advice-blog

Whatever visual aids you use, your children will still need time and space to process what is going on in their day so don't forget to have that quiet place for them to chill out in. I do hope the information has been of help and please contact me caroline@fragilex.org.uk if I can help you further, or for personalised visual aids to support your child.

Ten top tips for the transition to adult services

From Jane Oliver, Senior Families and Professionals Advisor (Adult Enquiries)

1 Start early

Many families say the earlier you and your son/daughter start to plan and think about the future the better. This is especially important if they might need support in their adult lives. Make sure your child's needs are assessed fairly and appropriately so any support in adult life can be planned effectively.

You can ask for a Child's Needs Assessment at a time of "significant benefit" when preparing for adulthood. This means that not all children will need to be assessed at a particular time - this can be during year 9 if you think that's appropriate.

2 Don't wait to be asked

Find out about Person Centred Plans. What process is used to put your son/daughter at the centre of their transition? Talk to family and friends and get their ideas about what is important for your son/daughter. Is there any information from the Local Authority (LA) about their transition process? Is there a transition team? How will they meet and get to understand the needs of this young person? Do they know anything about Fragile X and its effects? Think about ways of explaining some of the difficulties such as not able to manage change and transition well, the heightened anxiety that this can cause and the difficulty communicating your choices. Basically give you and your son/daughter a head start!

3 What things should you and your son/daughter be thinking about?

There will be lots of talk about "outcomes" and "goals" during this process. These will relate to:

- Further education and training opportunities
- Community inclusion and leisure/social pursuits
- Independent living or future living options
- Employment
- Keeping safe and well.

4 Talk to other families and contact organisations

Talk to other families about their experiences. There should be a “local offer” which is like a marketplace for what services are available in the local area. This should be on the LA website. Find out as much as you can about service provision from social enterprise schemes to day services, from friendship schemes to local clubs, from voluntary agencies to college courses. Contact your local carer centre for more help with this. They should have knowledge of the local Learning Disability Partnership Board and may have workers specializing in learning disability in your geographical area. They certainly will have knowledge of local support groups that might be helpful.



Think about ordinary life too such as: getting to the cinema, going shopping or using the local library, that kind of thing, how would these be accessed, would some 1:1 time or using a personal assistant be the right way.

5 What decisions are made in adult life?

Start to think about the kind of decisions that participation in adult life brings. For example, making decisions about money and where to live. Adult services will talk about these kinds of issues and will assess whether the young person is able to make these decisions. You will hear people talking about “capacity” and the Mental Capacity Act (England and Wales). In Scotland the legislation is Adults with Incapacity. This is legislation put in place to protect the interests of vulnerable people who are unable to make their own decision about a specific life choice - for example, where they want to live or how they want to spend their money. For England and Wales this may result in someone applying for Deputyship and in Scotland this is termed Guardianship. In Northern Ireland this is known as Controllership which comes under the Office of Care and Protection.

Please contact the office for more information if you feel capacity maybe an issue. Some young people with Fragile X will be able to make some decisions about their life and some will not be able to. Even if a person is unable to decide they should be included as much as possible, and the decision must be made with their best interests as the focus.

6 Have your say

As a parent you can be the advocate for your son/daughter. You know them after all and you should not feel excluded in the transition process or when adult services become involved. As a carer you should be listened to but if you do feel uncomfortable in any meetings take someone with you for support. Write down questions before the meeting and give yourself time in between meetings to look at and consider any options discussed.

7

What resources might be helpful when thinking about the future?

There are a lot actually but be careful that you look at up to date ones. Refer to your child's Education, Health and Care plan (EHC Plans) which are used in England. In Wales this may still be a statement although there will be changes in the future to an Additional Learning Need. In Scotland your child may be seen as needing Additional Support for Learning. In Northern Ireland your child may have a statement. For more information about Wales, Scotland and Northern Ireland please go to the Contact website (www.contact.org.uk).

EHC plans in England can last until a young person is 25 but only if they remain in education or training. It's also worth saying that social care needs identified during a Child's Needs Assessment must be added to the EHC plan along with an indicative Personal Budget amount. This is supposed to minimize the whole assessment process.

"Thinking Ahead: a planning guide for families" from The Foundation for People with Learning Disabilities is very useful. You can download this from www.togethermatters.org.uk. Although it looks forward to a long-term future it has some very useful templates that help you think about any future support for your child in a practical everyday sense. This could be a starting point when considering a support plan for adulthood. For a resource that is more focused on the transition from children to adult services itself have a look at this website: www.preparingforadulthood.org.uk

8

What is a care or needs assessment?

In adult social care services this is what will determine the support your son/daughter will receive. The local authority have a duty to assess an individual, but, there is no automatic right to services or support because you have a learning disability or diagnosis. It is the impact your disability has on your independence and wellbeing that is assessed. This is done by identifying and evaluating the needs that they have and what would happen if the presenting needs were not met. If the outcome was of significant risk to their independence and wellbeing, then these needs would be seen as eligible unmet needs and the authority would have a duty to meet them.

Even if your level of need is not sufficient to generate support your son/daughter should be offered relevant information that might be helpful. Carers are also entitled to a Carers Assessment. This examines the impact of your caring role in your everyday life. It also focuses on wellbeing as does the persons needs assessment. There is a duty to assess you as a carer. If you need short breaks or respite emphasize this in your carers assessment as well. Please think carefully about the support you give on a daily basis. You will always be doing more than you think and don't be afraid to say what you are not going to do. This is very important. A carer's support is not supposed to be considered when the assessment is being completed but it will be when the support plan is being made.

The support from a carer has always had a significant impact in self directed support. This is when someone is allocated money through a Resource Allocation System to buy their own services. They may be allocated less as their carer is meeting some of their unmet eligible needs. Please think about this.

There is relevant legislation here to refer to. Please go to Carers UK (www.carersuk.org) and look for the acts of law for where you are in the UK. Carers UK also have very good information about assessments.

9 Think about the practicalities

The practicalities should never be ignored. For example, how would your son/daughter get to college, their work experience, go shopping etc. Could they travel independently or would they need help to learn this - do they need a taxi?

You will be asked about using a Personal Budget or accepting Direct Payments. Are you happy to become an employer and recruit personal assistants? Or do you want an agency to help with this? Or do you want the social worker to take more control over service provision? Don't be afraid to get help around these issues.



Do not sign any assessment or support plan unless you are happy and agree with it!



10 Charges for services

Your local authority will have information about charging for adult services. This is standard in adult services. Basically your son/daughter's finances will be assessed to see if they can make any contribution towards the costs of their support. It is not a financial assessment on the whole family's finances. Information will be online but you should have a visit from a financial officer who will explain it to you. Please think about all the extra expense that the person's disability incurs. This is Disability Related Expenditure.

For example, do they need a special diet, frequent clothes/bedding washed and replaced, aids/assisted technology. You need to mention anything like this as these should be considered when they figure out the contribution towards the costs of someone's care. Please note that across the UK there are some differences in how services are charged for. Please see this factsheet for more detailed information about charging for community care in your region of the UK: www.disabilityrightsuk.org/charging-community-care

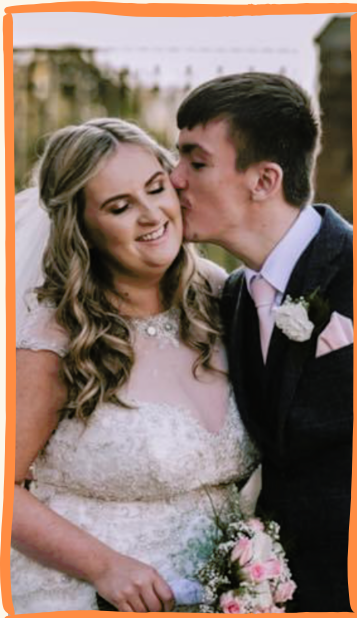
Inspiring Fundraising Stories

We are so grateful to everyone who is able to fundraise for us; no matter how big or how small the challenge, your kindness and efforts are heart-warming and we appreciate every penny you are able to raise. Here are some recent fantastic fundraising stories. If they've inspired you to undertake your own fundraising challenge, please do get in touch - we'd love to hear from you!

Dean Whitmore runs the Virgin Money London Marathon

Last October, Dean Whitmore ran the Virgin Money London Marathon on behalf of the Fragile X Society. Here, he tells us what inspired him to get involved.

"The reason I'm running and why this means so much to me... from growing up an only child after Kelly and Luke's parents passed away, and living with them both over the years, they became more like a brother and sister to me," Dean explains.



"Luke, (pictured left, with sister Kelly) also known as Lukey Loo, never stops smiling! Whatever happens, no matter if he's feeling poorly or down, he's always got a big, beaming smile for you. When we're out in public sometimes, Lukey can be quite loud and bring attention. I want everyone to know that although Luke has Fragile X, he has the biggest heart and always lights up every room he walks into with his amazing, unique personality."

"Lukey and his sister, Kelly, lost their parents at a young age and have been through so much together but I think you can see from the picture (left) that the bond they have is unbreakable."

Dean had run the marathon before and keeps himself in good shape, but it was still a huge effort for him to both train (especially during last year's hot summer) and raise funds for the Fragile X community. Dean raised an incredible £2,348 for the Fragile X Society!



Dean adds, "Me trying to raise awareness for Fragile X is just my way of showing them all I'm always going to be here to support them whenever I can. When I was out fundraising, the first question was always, 'What is Fragile X?' I already feel like I'm raising awareness which is all I needed to keep my legs moving when they were aching after training!"



John Quaye runs the Boston Marathon

On 18 April, John Quaye, one of the Directors at the Society, is running the Boston Marathon on behalf of the Fragile X Society

John tells us, "Boston will be my 14th marathon and my 5th of 6 majors having run New York, London, Chicago and Berlin, leaving Tokyo to complete the World Marathon Majors series. I ran my first marathon in 2010 and my next in 2016 after which it got a bit serious and I run 2-3 marathons a year.

I've been trying to run Boston for a few years but between cancellations and Covid restrictions, the closest I've gotten was running the virtual Boston Marathon in October last year. The race takes place on Monday 18th April and so training is pretty intense at the moment and I'm typically running 70-80 miles a week.



My marathon PB (personal best) is 2:54:53 set in the Isle of Man in 2021 and I'm hoping to better this in Boston.

I have a lucky top which has all my times on the back from the majors. You'll also notice my nickname which everyone always asks about - it's not running related but a nickname I had since I was about 7 years old (because my hair was the colour of baked beans - the joys of childhood)."

To support John, please visit his JustGiving page:

www.justgiving.com/fundraising/john-quaye

Other Fundraising News

We are especially grateful for the continuing support of all our members over the last year as well as the Trusts and Foundations who have donated to us. These include the Rosalind Williams Discretionary Trust, the Chapman Family Trust, Dunmow Football Club, Salt Town Contractors Ltd, the National Lottery and the Pears Foundation.

In addition, volunteers with a particular expertise (such as media skills, journalism, and proof reading) save the Society money and improve the services that we provide. We would like to give our thanks to these individuals, to our Specialist Advisors, and to all the others who have volunteered their time raising funds and awareness of Fragile X. Thank you!



Looking ahead, in response to interest from our members, during 2022/23 we will launch a new Legacy Programme which will help us to secure our long term future and enable us to keep providing help, support and advice to our membership.

The Three Peaks - with a twist!

Chris Gibson, along with three friends, George, Barney and Sam, took on a remarkable fundraising (and fitness!) challenge last summer.

Chris explains, "Myself and three wonderful (foolhardy) friends, George, Barney and Sam climbed the three peaks. However, being the plucky (witless) souls that we are we decided to aggrandise the challenge to boost our ratings. While climbing the three peaks we carried with us a rowing machine (cumbersome) and a few weights (unwieldy). Not just for fun but to allow us, when we get to the top (breezy), to row the height of the hill (Stratospherically high mountain) and then (after a snack) also lift the height of the peak in KGs. Yes; climb, row, snack, lift, repeat. 3 times. One. After. The. Other. 36 hours.

All this of course, was for a cause dear to mine and my family's heart. Our son Harrison was diagnosed with Fragile X Syndrome at the age of two. Fragile X is the most common known cause of learning disability, leading to social, language, attentional, emotional, learning and behavioural difficulties.

Early diagnosis and befitting intervention are key to ensuring children and adults can reach their full potential and live their best life. A diagnosis like this can be overwhelming; the Fragile X Society provided us a support system. But not only support, a road map enabling us to navigate each milestone, advice equipping us to locate the best help available to him, education empowering us to foster the most nurturing environment for him. They were invaluable. They are the only charity of their kind in the UK. They have three primary aims; to provide information and practical guidance to individuals and families living with Fragile X; to educate and inform the public and professionals in order to raise awareness and understanding of the syndrome and to encourage and support research into all aspects of Fragile X."



I cannot put into words how essential the support of the Fragile X Society has been and continues to be to our family. I wanted to raise as much money as possible to help ensure that their services are always available to other affected families.



Chris (pictured above in Fragile X Society T-shirt) along with George, Barney and Sam at the summit of Mount Snowdon raised an amazing total of over £10,000 to support the work of the Fragile X Society. With sincere thanks to Chris and the team, and to all who sponsored their 'crazy, but worth it!' challenge.

We are so grateful to everyone who is able to fundraise for us; no matter how big or how small, your kindness and efforts are heart-warming and we appreciate and value every penny you are able to raise. Please do share you stories with us, and tag us on social media!

Fragile X International (FraXI)

Fragile X International (FraXI) is a new organisation for country family organisations who support those who live or work with Fragile X. FraXI's vision is to help those living with Fragile X Syndrome (FXS), Fragile X Premutation Associated Conditions (FXPAC) and their families anywhere in the world to enjoy a happy life.

FraXI was founded by a network of 17 country family organisations from the European Fragile X Network. It is a registered INPO/AISBL in Brussels, Belgium. Full membership is open to all Fragile X family organisations around the world. Associate membership is available to corporations and non-family organisations.



FraXI will work closely with all members to provide advice and support in setting up and maintaining country organisations and act as a conduit so that countries can share best practice with each other. This will involve proactive raising of awareness of FXS and FXPAC internationally; and promotion of social inclusion of FXS at all levels of society. FraXI will oversee communication of latest research and influence the direction of research internationally on FXS and FXPAC through work with their Board of Scientific and Clinical Advisors.

A family member said:



Having a child with a rare condition can feel so isolating but with FraXI, I now feel a presence of support and sharing of knowledge that is working for the best outcomes for my child with Fragile X.



FraXI will be a proactive organisation, with all countries working together to share best practices related to support and interventions. FraXI will have a real impact in all aspects of the lives of people with FXS and Fragile X Premutation Associated Conditions (FXPAC) internationally.

Dr Kirsten Johnson, Chair of FraXI's Board

International News: Ukraine

The invasion of Ukraine by Russia is a shocking event, which has upturned the lives of many millions of people in Ukraine and there have been huge concerns expressed that adults and children with disabilities have been particularly badly affected. Our Fragile X Society here in the UK is part of a growing network of countries dedicated to supporting the Fragile X Community and we thought that you would like to know how our colleagues in Poland are responding. Joanna Kulisiak-Kaźmierczak from the Polish Fragile X Society sent this update.

"We help Ukraine"... these words accompany every Pole on a daily basis. It is not only billboards in the streets or in the media, it is a heartfelt commitment from each of us. When we learned that the war had broken out in Ukraine, we were terrified. Ukrainians, women and children, travelled to the Polish border. Often they only had documents with them. They didn't have any things. They came without luggage. Luggage and suitcases were left on the road because they were too heavy. They walked over 35 km to the border with Poland. When they reached the border they were exhausted and cold and still had to wait from 12 to 30 hours for the border check. Polish aid to the Ukrainians was immediate and spontaneous. Poles themselves, among their friends, organized collections of food, clothes, mittens and other warm things. And they took them in private cars to the border, and on the way back they took Ukrainian families from the border and brought them to Polish cities.

Mum Renata and dad Grzegorz took part in such an initiative, they are parents of twins with Fragile X Syndrome, who are in our Foundation "Fra X Family". The "Potrafię Pomóc" Foundation was also involved in helping, at which the Center for Rare Diseases was established. Bartłomiej Skrzyński, helped in the Ukraine from the third day of the war. Their driver / volunteer, whose name is Maciek, transported people with disabilities and their relatives from Ukraine (Lviv, Tarnopol and Winnica).



In addition to this activity, in Poland, points for refugees were organized at railway stations, where they can register, receive the help of a language interpreter, medication, accommodation and food. There are also volunteers from various non-profit (charity) organizations and some who volunteered to help.

Not so long ago, there were many people and their pets in the buildings of railway stations. There were dogs, cats, guinea pigs and more. Now there are fewer and fewer of them, because some of these people continue to travel to Europe, some stay in Poland and live in hotels and rooms prepared for them. There is a lot of work in these hotels for refugees from Ukraine. They all work as a volunteer there. Lots of things have to be organized there. Clothes, food, toiletries and a place to sleep. People who feel sick are taken to a doctor or pharmacy. These people include pregnant women and young children, and most speak only Ukrainian. Often their illness is psychological, shock, stress.

Mother Joanna and father Maciej, parents of adult children with Fragile X Syndrome and their friends, got involved in helping in one of the hotels for refugees from Ukraine in Wrocław. As part of this aid for refugees, many donations in kind were collected also from donors from Germany and America. A playroom for children was created in the hotel, help during illness, transport or finding a new home, accommodation in Poland and abroad was provided. As part of the activities of our Foundation "Family Fra X", we are in contact with a Ukrainian family diagnosed with Fragile X Syndrome, who currently lives in Poland.

Research News



Research news, findings and updates from our Research Information Manager, Emma Mather-Pike. Please see our website www.fragilex.org.uk/research for further information.

Measuring electrical activity in the brain may help in the development of drug therapies for Fragile X Syndrome (FXS)

Electroencephalography (EEG) is a non-invasive way to monitor electrical activity in the brain. Researchers have recently used EEG to study children, adults, and laboratory animals with FXS, and their findings highlight that FXS causes detectable patterns of hypersensitivity to sound. These electrical brain patterns can be used as biological indicators of FXS, and such biomarkers can be monitored for improvements to assess the efficiency of new medicines and treatments for FXS. Various FXS clinical trials of drug called Minocycline show promising evidence that EEG is a reliable measure of therapeutic success in drug discovery process.

Barriers to participation in FXS clinical trials - what are they and how can they be addressed?

It is challenging for people with FXS to consent to and participate in clinical trials, as these individuals experience varying degrees of intellectual disability, autism, anxiety, hyperactivity, and hypersensitivity. Conducting such trials is, however, essential to meet the growing demands for new medications and treatments for FXS. The Patrick Wild Centre recently conducted a survey to better understand these challenges and how they can be attenuated. Providing abundant data on side-effects and safety, hosting information evenings, organising family-researcher meetings, providing transportation and financial assistance, allowing flexibility with clinical and remote consultations, and desensitising participants to trial procedures are several of the improvements people with FXS and their families suggested to enhance enrolment and improve experiences in clinical trials. This is highly valuable information not only for FXS-related clinical trials, but for any that hope to recruit individuals with intellectual disability or autism.

Whole genome sequencing (WGS) to detect repeat expansion disorders including FXS

Accurately diagnosing repeat expansion disorders like FXS, caused when short sequences of DNA are repeated many times over within specific genes, is a challenging, inefficient, and expensive process. Up to 70% of individuals showing symptoms of such disorders remain undiagnosed, as conventional genetic screening can only test one gene at a time. The Genomics England Research Consortium have for the first time trialled WGS as a single genetic test screening 13 disorder-associated genes (including FMR1 for FXS) in thousands of patients with neurological symptoms. Results showed that with manual correction, WGS can detect presence and absence of expansions with 99.1% and 100% accuracy. A total of 64 individuals received diagnoses due to these WGS methods, 10 of which were FXS diagnoses in young adults. These findings support the use of WGS as an accurate, rapid diagnostic test.

Acronyms

FXS - Fragile X Syndrome

EEG - Electroencephalography

WGS - Whole Genome Sequencing

FMR1 - Fragile X messenger ribonucleoprotein 1 (gene)

Research News

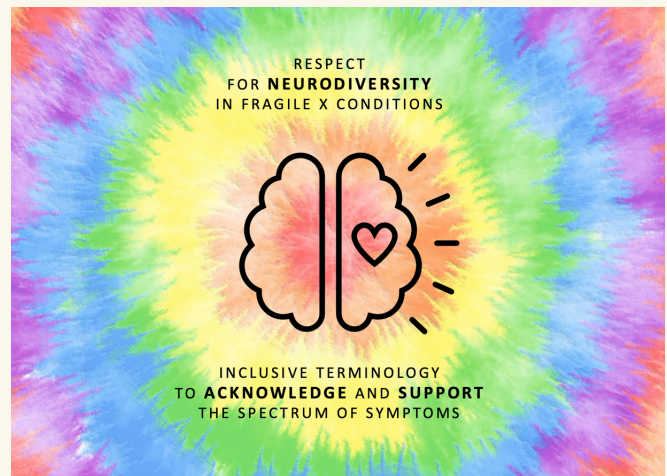


University of Edinburgh's Road to Recovery Project

Since the outbreak of COVID-19 and the enforcement of nationwide restrictions, caregivers of children with intellectual disabilities (ID) and the children themselves became increasingly vulnerable to mental health problems. To better understand these vulnerabilities, the University of Edinburgh began the Road to Recovery Project by interviewing carers, teachers, healthcare practitioners, and charity organisations (including the Fragile X Society) involved with children with ID before and during the pandemic. Carers reported elevated levels of stress which were attributable to feeling isolated and detached, losing enjoyable routines, and having restricted access to educational and respite services for their children. The distress further increased their child's problem behaviours, thus perpetuating a vicious cycle. Healthcare and educational workers felt similarly distressed due to being overworked and under supported, and charities lacked staffing and financial means to proactively support families. To restore mental wellbeing, governments are encouraged to provide target interventions to all levels of support which can withstand pandemic-related disruptions.

Eradication of 'mental retardation' from language describing Fragile X Conditions

Members of European and International Fragile X networks published an article and convened with the Gene Nomenclature Committee to advocate for removing 'mental retardation' from terminology describing Fragile X Syndrome (FXS) and Fragile X Premutation Associated Conditions (FXPAC). This pejorative phrase is outdated, offensive, and misleading, and people with FXS and FXPAC and their families are deeply upset by its usage in clinical and casual conversations surrounding their diagnoses.



FXS is indeed the leading inherited cause of intellectual disability, but the condition confers a wide range of other symptoms which manifest uniquely depending on the age, sex, and development of the individual. Moreover, FXPAC does not cause intellectual disability of any degree, so the usage of this label is inappropriate and reductionistic for both conditions. These justifications and many others were eloquently put forth and accepted, and 'mental retardation' has been successfully removed from terminology describing the genetic development of FXS and FXPAC. This achievement highlights a respect for neurodiversity, and it is hoped that the new nomenclature can be swiftly adopted in all fields.

Acronyms

ID - Intellectual Disability
FXS - Fragile X Syndrome
FXPAC - Fragile X Premutation Associated Conditions



Friends of Fragile X is a way you can support the Fragile X Society on a regular basis through monthly donations. We receive no government funding and are dependent on the kindness of others so we can continue to provide our services. This includes staffing, office costs, and maintaining our helpline, to deliver our objectives, which took over 967 calls last year.

You can become a Friend of Fragile X by pledging a monthly donation to the Fragile X Society. If you are signing up as an individual, you can join for a donation starting from £5 per month. If you are signing up on behalf of a family group, you can join for a donation starting from £10 per month. If you are in a position to do so, you may choose to give a higher amount. Your generous support provides a vital source of regular income to enable us to support the Fragile X community across the UK. Scan the QR code to see our 'Frequently Asked Questions', and to join the Friends of Fragile X scheme.



Friends of Fragile X



“

I cannot tell you of the weight that has been lifted from my shoulders knowing this information now...I cannot thank you enough.

”

The Fragile X Society



Pete Richardson, Managing Director

Pete holds responsibility for delivering the Fragile X Society's Charitable Objectives in accordance with best practice and charity law, ensuring that the organisation operates effectively raising funds and meeting the needs of its members. (pete@fragilex.org.uk)

Jane Oliver, Senior Families and Professionals Advisor (Adult Enquiries)

Jane is the Society's Senior Families and Professionals Advisor dealing with adult enquiries for individuals and families. (jane@fragilex.org.uk)

Caroline Pringle, Families and Professionals Advisor (Child Enquiries)

Caroline is the Society's Families and Professionals Advisor managing enquiries relating to children with Fragile X Syndrome. (caroline@fragilex.org.uk)

Kate Williams, Communications and Office Manager

Kate is responsible for supporting our office, our website, email communications and database, and our events. (kate@fragilex.org.uk)

Sharon Young, Administrative Assistant

We welcome Sharon, who has joined the team as Administrative Assistant, responsible for general office duties. (sharon@fragilex.org.uk)

For individuals and families

We provide the latest information, resources, literature and publications about Fragile X and enable peer to peer support networking through online forums, events and training, and our Family Weekends and conferences. We also offer support and opportunities to take part in research into Fragile X Syndrome and associated conditions.

For professionals

We offer helpline support, resources, e-learning to educate about Fragile X.

How to contact us

You can call our helpline on 01371 875100, available Tuesday and Thursday 9am - 5pm and Wednesday 1pm - 5pm; email us on info@fragilex.org.uk or write to us at: Suite 5, The Chestnuts, 4 Stortford Road, Great Dunmow, CM6 1DA.

Our website has lots of useful information: www.fragilex.org.uk
Follow our social media for more updates.





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