

Spring 2023



MESSAGE FROM THE CHAIRPERSON

We are thrilled that Fragile X International is now established, welcoming new members, and already making a difference. There is a lot of wonderful work being done in our member countries.

This newsletter will highlight some of those activities and hopefully inspire you with ideas for you to try in your own countries.

We all work to build awareness in our countries - so many have not yet heard of Fragile X Syndrome or Fragile X Premutation Associated Conditions.

One opportunity coming up to give a shout out for Fragile X is Rare Disease Day 2023. You can find more info on that at www.rarediseaseday.org

Many of us, when receiving the diagnosis of Fragile X Syndrome, felt bewildered and alone. Indeed, it is often difficult to get a diagnosis in the first place. I was recently interviewed in a podcast about the importance of early diagnosis for rare conditions, and highlighted Fragile X Syndrome. You can listen to that podcast via streaming links here, and a short clip via Twitter here.

Enjoy our first newsletter! And please keep sending in articles and stories so that we can celebrate and learn together!

Dr. Kirsten Johnson, FRSA, Chair, Board of Directors, Fragile X International aisbl

RARE DISEASE DAY 2023

Join Fragile X International on all our social media channels to support Rare Disease Day and raise awareness about Fragile X Syndrome. Rare Disease Day is a globally-coordinated movement on rare diseases.

Together we can influence and work towards equity in social opportunity, healthcare, and access to diagnosis and therapies for people living with a rare disease.



Working together as a unified voice we can influence sharing of international knowledge and expertise about these important issues related to Fragile X Syndrome.



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FRAGILE X INTERNATIONAL FIRST GENERAL ASSEMBLY

The first general assembly of FraXI took place in beautiful Lyon in November 2022.



Families, healthcare professionals, and Fragile X Organisations came together for an inspiring conference sharing knowledge and expertise with a series on best practices and future opportunities for international collaborations.



NEWBORN SCREENING

FraXi's Chairperson, Dr Kirsten Johnson speaks in this excellent podcast about the challenge a diagnosis poses for Fragile X families and the quality of life that Newborn Screening would provide to so many.

Listen here to the podcast and please email info@fraxi.org with any questions you have in relation to screening



NEWS FROM FRAXI MEMBERS AROUND THE WORLD

We are delighted to share news from our member organisations around the world in this newsletter and a huge thank you to all for your wonderful inspirational news, activities and stories.





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NEWS FROM THE NETHERLANDS

The Dutch Fragile X Association was very pleased to have a live Contactdag on 17 September 2022 after a long Covid-period with online encounters.

Around 120 persons attended and they were all very pleased to meet each other again. An extra special day because we celebrated that our Association was founded 15 years ago.



An extra extra special milestone was that our Chairman, Bas Douwes, who stood at the beginning of our Association, was knighted. He received a beautiful broach. It is a special procedure because a lot of

people have to give recommendations and then the King has to approve his knighthood.



Bas however has decided, already a while ago, that it was time to pass on his presidency to another board member. Lidwien Bernsen is our new president and she represented the Netherlands at the first FraXI General Assembly meeting in Lyon.

In non-Covid years we usually organise two days each year. This year we were able to have our Family Day live! on the 18th of June. On Family Day, families with their children meet each other and share experiences, in a relaxed environment with enough space for the children to play. Most of the times it takes place somewhere in the countryside such as a farm or an activity centre. This year we went to the Museum of Trains in Utrecht, on one of the hottest days of the year. But luckily the children could roam around in the whole museum, and outside too.

NEWS FROM SPAIN

Spain lit up brightly for X.X 2022, here are some of the lovely buildings lit up.





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NEWS FROM SPAIN

Females with Fragile X Syndrome

Since 2012, the Spanish Federation of Fragile X Syndrome, organizes meetings of women affected by the full mutation of Fragile X syndrome. Since then, 16 of these meetings have been held in different parts of Spain.

The philosophy behind this activity is to work on aspects related to interpersonal relationships, with special interest in those fears and psycho-social complexes that directly affect relationships with third parties. The aim was to provide strategies to remedy what was already demonstrated in the study "Girls and women with Fragile X Syndrome in Spain. Study on their needs and those of their families", where serious problems were detected in the social insertion of these young women due to lack of skills.

Based on these principles, at the end of 2022, a meeting was organized in Buitrago del Lozoya (Madrid), attended by nine young people from different communities in Spain. In order to promote personal autonomy, this meeting was held in a house.



There, the participants were in charge of organizing everything related to living together. They also visited different places in Madrid, had lunch and dinner in restaurants and attended a musical show.



The main objectives are:

- 1 To enhance the use of cognitive and communication strategies, which can help them to implement problem-solving tactics
- 2 Increase self-esteem
- 3 Encourage autonomy
- 4 Learn to identify and express their feelings
- 5 Improve communication with the immediate environment.
- 6 To deepen in the expression of their needs in an appropriate way
- 7 To defend their ideas
- 8 Promote their social and labor integration

In addition, we work with families on the following aspects:

- 1 To know the behavior of their daughters
- 2 Detecting which are the main problems they detect in their daughters
- 3 How to face new challenges
- 4 Reflecting on whether they will be able to lead an independent and autonomous life



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NEWS FROM SPAIN

Study of incidence of Fragile X Syndrome in Spain

The Spanish Federation of Fragile X
Syndrome has undertaken a study of the incidence of Fragile X Syndrome in Spain using an online platform. The need for methodical and accurate information that impacts and facilitates health practice is recognized. This facilitates management, making it possible to direct and evaluate research and keep patients correctly informed. This function entails the use of appropriate diagnostic techniques, precise interventions and may enhance studies and clinical trials.

The main objective of the study is to provide useful information on prevalence, evolution, results and needs. The intended benefits of this study are as follows:

- Real knowledge of the prevalence of Fragile X Syndrome
- To have data by sex, age, population, social and family situation
- Knowledge of the treatments administered
- Knowledge of the centers that treat patients.
- Knowledge of the specialists who treat patients.
- To facilitate clinical practice
- To serve as a basis for research and clinical studies.
- To offer adequate knowledge of their problems
- To deepen knowledge of the problems of associated disorders: FXTAS and FPOI.

NEWS FROM CATALONIA

The Catalan Fragile X Association organises workshops aimed at family members of people with Fragile X, led by leading professionals in each field, here are just two of these excellent workshops:

For grandparents: Two monthly online sessions, where grandmothers and grandfathers of people with Fragile X find not only a space for reflection, but also support tools to accompany their grandsons and granddaughters and to be able to respond to each stage of the person's life. The sessions are led by Belén Solano, a psychologist trained in cognitive behavioral therapy.

Mutual Support Group: This support group is facilitated by Eduardo Brignani, psychologist and psychotherapist specializing in disability, families and minority diseases, these groups are ideal for sharing experiences, challenges and needs in relation to raising a person with Fragile X.

National Patient Meeting

Pilar Mas, vice president of the association, participated in the 6th National Patient Meeting organized by the Spanish Patient Forum. A unique event at European level to generate dialogue, debate and learning in relation to improving health and care for citizens.



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NEWS FROM CATALONIA

A Sant Jordi with Fragile X

The Diada de Sant Jordi (Saint George's Day, popularly known as Sant Jordi) is the day on which the death of Saint George is commemorated. On this day, April 23, lovers in Catalonia traditionally give each other a rose and a book.

What would happen if Sant Jordi and Princess Cleodolinda had the Fragile X? We explain it in this adaptation of the Legend, where through the two main characters we see drawn some of the most genuine characteristics of the syndrome. The release of this legend coincided with St. George's Day.

San Jorge,

San Jorge,

San Jorge,

la Princesa

y el dragón

Una leyenda de tierras catalanas

For more news from L'Associació Catalana de la Síndrome X fràgil you can visit their website https://www.xfragil.cat/

NEWS FROM SWITZERLAND

Although it is almost three years since the Covid pandemic began and changed our lives and the world quite dramatically, Kristin Mulcock, Co-Chair of the Swiss Fragile X association FRAXAS, wrote these beautiful words about the resilience that her two children showed and how our children with Fragile X can adjust to different situations.

My heroes in the corona crisis_

My two sons (20 and 14) have a genetic condition called Fragile X Syndrome. This genetic defect causes intellectual disability. I hate the word "defect", because from my point of view and from their own, they are not defective, they do not want to be different from what they are, and they do not suffer from Fragile X, but only from intolerance and lack of understanding of their situation in society. But that is another story...





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I have always marvelled at this, and also admired them for the fact that they both live only in the now, do not want to plan or control the future, and are in absolute confidence that everything is good - and if it is not good, that it will be good again. This basic trust in the goodness of life is so often interpreted as naivety, lack of intellect or lack of imagination.

But let us imagine that they are right in their assumption that everything is or will be good, and that it is important to live life only in the present without looking full of worries into the future. How many of us get anxious when we are not always in control, when we have not planned our lives in detail, when we have taken out insurance for everything even beyond our own death? I suspect we all feel at least some uneasiness about it.

And then suddenly there is an unprecedented pandemic on this scale, everything stops, becomes quiet, people close their doors to the outside world, stay at home, panic spreads, fears about the future, job losses, economic disasters are predicted down to the last detail in countless special programmes on television, all plans for the future can be buried for the time being, the supposed control is gone and so is the restful night's sleep. The entire planet seems to be reorganising itself, is in upheaval, chaos, question marks, inner unrest, nervousness, crisis ...



... and suddenly I watch my boys and I notice that they have remained the same, full of joy about every new day they can spend at home with their family, about the tasty food, a bike ride in beautiful weather, the weekly evening series, which thank God is still on TV, playing badminton in the garden, a visit to the neighbouring farm, where you can now buy not only eggs but also masks.

I realize that my two sons are my heroes, from whom one can learn so much, that they had and have the insight, not us! And I realize that the supposed control and planning of life was an illusion that made us feel safe. I notice that our minds have taken away our view of what is essential and really important. So I firmly resolve to take a good example from now on from their philosophy of life and understand that every single moment counts, not the prospect of tomorrow or afterwards.



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This brings to mind what my father, who died almost a year ago, often said to me:

"Life is made up of many small moments, and the more beautiful moments you had, the more beautiful your life was at the end."

If you look at it that way, it doesn't seem to be that difficult to achieve a fulfilled beautiful life.

NEWS FROM FRANCE

Picnics everywhere in France

For the 2nd consecutive year, the regional delegates of the Fragile X France association organized, each in their region, a day of meetings and discussions on September 18th. After the very good mobilization of 2021 (140 participants), we are proud to announce that in 2022, 11 regions participated and brought together 225 people throughout France. We hope that there will be even more of us in 2023.



Family weekend

After two years of waiting, the 2022 family weekend has filled up by bringing together 52 families from Port-Bail in Normandy. The participants were very satisfied with this high point in the life of the association, and the rating from the satisfaction questionnaires exceeds 9/10! The conferences and round tables were appreciated, as were more festive times such as the evening concert hosted by the music group Manata. Back in testimonials and images on this great event!



e-Workshop on Fragile X Syndrome

The e-workshop on Fragile X
Syndrome and associated diseases
organized by the association was a
real success. In numbers:
339 registrants from 31 different
countries including:

- 182 clinicians, genetic counselors
- 112 researchers
- 34 association representatives or members of the organizing committee
- 11 representatives of pharmaceutical companies



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Among the most represented countries, France comes first, followed by Italy, Poland, the United States, the United Kingdom, Belgium, Portugal, Spain, the Netherlands and finally Romania.

Satisfaction questionnaires were sent to participants at the end of the two days, and the average score awarded by professionals is 8.95/10!

The discussions were interesting, and several researchers wished to be put in contact with some of their colleagues present during the exchanges, in order to compare the results of their respective studies. The objective of the association is initially to link clinicians and researchers working on Fragile X Syndrome to consolidate or bring out new therapeutic avenues... the bet is won!

A scientific journalist, who attended all of the discussions, completed the drafting of a document which summarizes the main elements of each conference held during these two days.



NEWS FROM ITALY



Welcome 2023! We will be in the third year of our Erasmus Project +. New participants, new work and independent living experiences, new skills and more awareness.

People with Fragile X at the end of their secondary school studies will again have the chance to experience a one month stay in Malta, working for 4/5 hours a day at a restaurant or a hotel, taking care of their personal and home needs, supported by their educators. Everything is focused to improve self-esteem, awareness of one's abilities outside a family context so as to show everyone that inclusion, happiness and self-determination are truly possible. People with Fragile X can be a valuable resource to the community. We have to keep on creating opportunities for this!





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NEWS FROM ITALY

For this reason, our association has started its partnership with ANDEL, a national agency that will act as a bridge between possible employers and people with disability, who are currently being interviewed in order to gather information that will then match with companies and employers needs.

We are also continuing to spread awareness about Fragile X to support families, schools and professionals with our books and through training. Our action focuses at both nationally and regionally, thanks to the activities promoted by the local sections of our association. The main target is to inform families about opportunities and entitlements they can claim from their local authorities. One of the key topics is the Personalized Life Plan, a resource, quaranteed by law, to getting to know people with disabilities, their dreams, skills and needs so as to organize effective supports and aids in order to allow them to realize their own life project and to be citizens in full.

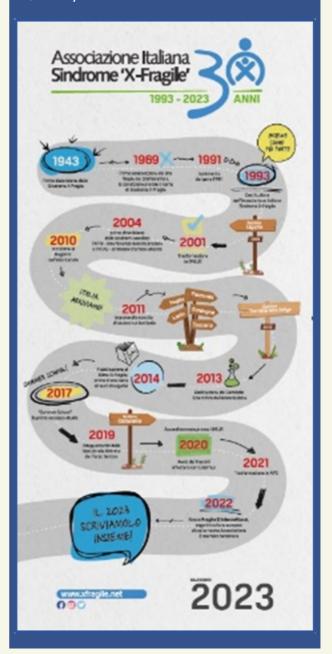








2023 is our 30th anniversary. It will be another year of challenges for our community and families. Some of them already known, others brand new. But we are not afraid. We are READY. Our calendar will be day by day to remind us how tough every challenge can be, but, also, how successfull can be. We can do it, always!





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NEWS FROM GERMANY

A major step in our German fragile-x community was taken in October last year. Our new executive director started her work: Nicole Schmidt. She has got a young daughter who is diagnosed with Fragile X Syndrome and knows all the tricky parts of being a parent with a special needs child. She has great ambition to change the way we obtain the authorities' support. We are very lucky to have found her – or do we better say, we are lucky she has found us.



NICOLE SCHMIDT, EXECUTIVE DIRECTOR

Nicole is the first official director to lead the association and we hope to be able to professionalize our way of work significantly; more so since the community grew to 630 members, each member being one family. We are very proud of this development and happy to be able to offer 14 seminars and workshops in 2023.

In addition, last year the three experts working for us started online seminars

for care workers of all institutions, teaching and sensitising the special and sometimes challenging behaviour of our younger children as well as the grownup children.

Many people living with the Fragile X Syndrome of all ages are being misunderstood, be it in day care or group homes. We are grateful about the ongoing demand on the seminars of "Fragile-X: Handle with care" and eager to help.



Friday, January 13 2023 was a lucky day for the Fragile x community. Our new heritage video clip was launched on Youtube, our website, and several social media platforms. The wonderful film agency "Sympathie-Film" in English something like "Good Chemistry-film" did a great job sketching the complicated matter into nice and easy to get images. The muted video is interesting, too. But since there are already similar videos in the English language available, there won't be a need for translation.



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NEWS FROM GERMANY

See the video on the following link: https://www.youtube.com/watch? v=tz5WvsAt1UY

The new video is a sequel of our first video, which shows the family life of two carrier sisters whose children are affected by the Fragile X Syndrome.



NEWS FROM MOROCCO

Who we are? A nonprofit association that is interested in people with Fragile X Syndrome and underlying diseases; The founding members are Adib's mom and brother, Adib's teacher and friends working in the health sector who have shown interest, understanding and motivation to help and to bring added value to the Moroccan Fragile X Association.



First General assembly at the Foundation Orient-Occident in Rabat, July 28,2022. Pictured from right: Syrine, Hanane, Zineb, Kawtar, Leila, Ahmad, Fatima Zahra

Mrs. Leila Roudani – Founder and president of the Moroccan Fragile X Association; Mrs. Syrine Hamada – vice president; Miss Zineb El Abdessalami – general secretary; Mr. Ahmad Wahid; Mrs. Fatima Zahra Allaoui; Miss Zineb El Abdessalami; Mrs. Kawtar Ainouch; To contact us phone +212661679138/+212670025632 or

+212661679138/+212670025632 0f mail: roudanileila@gmail.com /elabdessalamizineb@gmail.com

How was the association created?

The impact of the syndrome on the behavioral, health and educational levels of Adib's life and all the other members of his family have motivated Mrs. Leila Roudani to create the Moroccan Fragile X Association. The idea of creating this association existed for a long time and the meeting with beautiful souls made this project real.

The first constituent general meeting for the creation of the Moroccan Fragile



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X Association (FragilX.Maroc) was held on Thursday July 28, 2022 at 6:40 p.m. at the Orient-Occident Foundation in Rabat. The assembly took place in a serene atmosphere full of motivation and a lot of hope for families and children with Fragile X and it was honored by friends and Adib 's music teachers (Thanks a lot to: Hanane, Souad, Isaak and Horty).

What Are Fragil X.Ma objectives?
•Raise awareness and recognition of the Fragile X syndrome among Moroccan citizens and public authorities by providing information about this not yet very known syndrome

- ·Informing health professionals by organizing conferences scientific seminars about the topic
- ·Improve access to information by writing and distributing guides and materials relating to Fragile X Syndrome
- ·Create a partnership with other associations working on Fragile X Syndrome

The administrative procedure was ongoing to create our association, we considered it useful while waiting to make contact very early with people or associations interested in Fragile syndrome at national Alliance of rare disease or at the internationally by creating bridges with the International Alliance, moreover we had the very

great pleasure to attend it from the beginning through its first meeting in November 2022 at Lyon -France.
A lot of work and challenge certainly awaits us, but we believe that we have a lot of courage and the motivation to move forward.

Adib (in photo) celebrated his 20th birthday on Thursday January 05, 2023. Twenty years full of fear of the future and hardship, but also full of happiness, joy and love.



Last year he had succeeded in the baccalaureate (adapted program), that means a lot for him...for us...and the best is coming.



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NEWS FROM NORWAY



Greetings from FRAX-Norway. In 2022, we put the members best interest in focus. As we have not found an employee, the board has taken over the employee's duties. We used this opportunity to create more meeting points for the members.

The spring has been used to map work tasks and plan various events in the board and together with regional contacts. New families expressed a desire for more meeting points and we focused on this to make it happen.

In June we met at Eidene in Tønsberg - where we usually have a family weekend. New this year was that each regional contact was responsible for following up new families in their region. We got feedback from the members that this was a positive experience and the families felt well taken care of.

Around 80 members attended the family weekend, where also the annual meeting took place. This year we had a new activity, a disco on Saturday evening! On Sunday we went on an alpaca trip.





Later in June, we were at the Frambu Conference, Rarely Happy. Frambu is Norways largest competence centre for rare diseases. Frambu collects and develops knowledge about rare and little-known diseases and shares this with patients and their relatives, and professional communities. This is how they want to contribute to children, young people and adults with rare diseases being able to live a life in line with their own circumstances, wishes and needs.

Frambu is a self-owned private foundation, an interdisciplinary competence center for rare diagnoses, part of the specialist health service's national competence service for rare diagnoses, a statefunded supplement to the ordinary service apparatus, a meeting place for families and professionals, for both children and adults - from all over the country - throughout their lives



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NEWS FROM NORWAY

Competence development, knowledge dissemination and guidance: Frambu provides an interdisciplinary offer to children, young people and adults with over 400 different rare and little-known diagnoses and their families/relatives and service providers.

In September, we went to Kristiansand and visited the zoo and Captain Sabeltooth. He is a big attraction for many generations in Norway. We repeated the success from the previous year. 32 members from 10 different families came along and had a great time. We ate a pirate breakfast with the pirates and had a guided tour through the zoo where we had the pleasure of feeding some of the animals.







Later that year in September, we arranged a long-awaited summer camp trip to Rhodes in Greece. We had not arranged a summer camp since 2019, before covid.

Previous years we went to Mallorca. It was a super nice trip, and some of the new families joined too. We were lucky to have our own personal trainer with us who arranged two rounds of training. There was loud music and every



of training. There was loud music and everyone could participate.

In December we made a trip to Oslo, where we had lunch. There were 20 members (7 families) who participated, of which some were new to our association. We got feedback that this was a great success.

Greetings from the board in Norway



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NEWS FROM POLAND

We are a Polish charity organization "FraX Family" Foundation, which was established in 2015.



In 2021, we had the pleasure to organize in Poland in the city of Wroclaw during the pandemic the 11th international meeting of the European Network Fragile X (EFXN), which for obvious reasons could not take place in 2020.





The opportunity to host representatives from international fragile X organisations in Poland motivated us to take up challenges and activities for the year 2022.

The first of them was International Women's Day on March 8, this year the ladies met at the celebration by mothers from the FraX Family in the Portrait Restaurant at the Royal Castle joining a concert of film music.

At the beginning of June, our 12th annual meeting of the FraX Family took place. We met in the Mountains in Karpacz (south-western Poland). We enjoyed beautiful views and fun, ranging from: trips to the Czech Prague and gondola rides to Sněžka, barbecues and games and activities for families. We had the pleasure to host a new family from Ukraine, dad, mother and 8-year-old son with Fragile X Syndrome. The family had to leave Ukraine due to the political situation.





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NEWS FROM POLAND



Next up was the X-Men Challenge and the Great Pendolino Super-Fast Train Journey into adulthood. In August, we announced a social media fundraiser for the X-Men tour.



Within 2 weeks we collected the right amount of money and had the opportunity to organize a trip into adulthood. The main goal was to make our adult X men independent Fra X men. A group of 16 participants and 3 coordinators (not parents), went on a trip to Krakow. For some, this was their first trip ever to do without parents. It was great. Activities included sailing a ship on the Vistula, the Castle of Polish Kings Seering, bagels...

baked bagel is a specialty similar to pretzels in the Polish region, selfpreparation of breakfast and dinner, an evening in pub for a beer, (some participants did not like the beer and it was confirmed that it was not a good drink!).



In October, as part of the XX 2022
International Fragile X Awareness
Day, our FRAXI Dwarf was officially
unveiled in front of the Diagnostic
and Therapeutic Center for Rare
Diseases in Wrocław, This is where
the previous year's EFXN meeting
took place. After the unveiling, there
was a health run for people with
Fragile X Syndrome, after which the
participants were awarded medals
and certificates. A group of 60 people
accompanied by the Police passed
through the streets of Wrocław
carrying the Fragile X logo.



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NEWS FROM POLAND

At the finish line there was a stadium for a family picnic for the residents of Wrocław. There were games, food, an orchestra and a music band that played on stage.

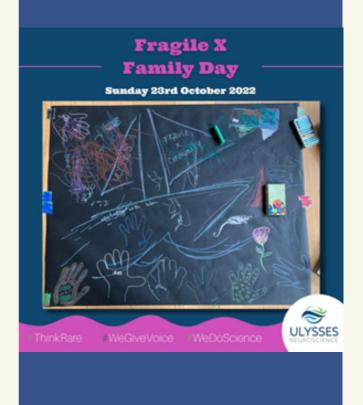




During the Picnic, our gentlemen collected money in cans for a summer camp. Our awareness day was featured on television and radio, On radio, there was the story of FRAXI Gnome. So far, we have recorded in Polish a story about how FraXI came to Wrocław and together with other dwarfs was looking for a diagnosis of who he is. The story describes the behavior of people with fragile X syndrome. We hope that Fra X is becoming more and more popular thanks to the fact that Frax consciousness will always continue. This year, we have been contacted by several young families with a diagnosis of Fragile X Syndrome.

NEWS FROM IRELAND

The Irish Fragile X Society held a family day in Maynooth University hosted by Ulysses Neuroscience. Ulysses Neuroscience ltd is a Research group based in Trinity College Dublin who are currently carrying out a study to assess cognitive abilities and develop composite biomarkers in Fragile X Premutation carriers. The research study is taking place to detect potential early signs of cognitive decline to assess the risk of developing early neurodegeneration in Fragile X Premutation Carriers. The family day was a lovely time for families to meet up, for children to play and to talk about the latest research taking place in Ireland on Fragile X Syndrome





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NEWS FROM BELGIUM

The Belgian association is holding a family weekend in March in a manor in the little town of Rochefort, in the heart of the Ardennes. The annual general meeting will be held on Sunday and a conference on post-school administrative procedures and opportunities will be presented.

A member of the Belgian association, Florinne, is presenting Spacio, a game she is playing with her 4-year old boy in order to get him familiar with animals, colours and space, by using silicone toys with a velvet touch. A card shows a particular placing of the animals, and the child has to place the toys in the same order on a plate, e.g. the elephant is in front of the giraffe, on the left of the hippopotamus and on the right of the monkey thus between them both. In a second step, animals are not presented with their face but on a side or backside. Each play lasts 10 to 15 minutes.



FRAXI COMMUNICATIONS COMMITTEE

We hope you have enjoyed reading the wonderful news from Fragile X International.

We would like to introduce our friendly communications committee and we welcome your news for the next edition of FraXI News!





Brigitte Hermans Anja Schweinberger





Brendan Weight

Please follow us on social media for the latest updates, events and news









Fragile X International welcomes donations. If you would like to donate please go to our website www.fraxi.org or click here

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