WSAI News

Volume 20, Issue 1, Spring 2015

My Dream Come True

Ashleigh O'Hagan

Hi Everybody,

I am delighted to share my great news that I have been chosen to represent Ireland in the 2015 world games in Los Angles in July. I have been competing in rhythmic gymnastics for over seven years now with my club in Limerick. We compete at local level and some times we go to Cork to compete at national level. I have won quite a few medals in different categories.

Last summer I even surprised myself by winning two gold medals and one silver medal. Following on from the hard work in the all Ireland games, I then qualified to represent Ireland in the world games. I have to train quite hard, with the rest of the Irish team which means I have now made lots of new friends from all over Ireland. I train in my local club in Lisnagry, in Limerick twice a week. I also train in Dublin once a month with the full Irish team and all the athletes heading to LA this summer. The training in Dublin is great fun because we get to stay overnight in a lovely hotel and share a room with others in our group. No mammy's kisses or no Daddy smiles!

Ashleigh in Competition

Ashleigh O'Hagan

The organiser's get to find out who snores, who talks, or walks in their sleep. (Not me!) so we will all be comfortable with each other when we are ready for Los Angles.

All the local newspapers interviewed me over the last few weeks and I had lots of photographs taken with other athletes and famous faces. I was invited to the launch of the Great Limerick Run a few weeks back, which is a 10 Km run held in Limerick every year to raise funds for lots of different charities. I will keep you all posted on my progress in the games. Watch out for me in the coming months.

Love Ashleigh

From Limerick Leader on January 31 2015.

"This July, Ashleigh's long-held dream will come true, as she will be the only Limerick athlete competing in the world games of the Special Olympics in Los Angeles.

Ashleigh's speciality is rhythmic gymnastics and she has been in training in her field for the past 10 years in Lisnagry.

The Special Olympics World Games will take place in Los Angeles from July 25 to August 2. The Games will see 7,000 Special Olympics' athletes from 177 different countries compete in 25 sporting events over the duration of the Games and it is set to be the single biggest event in Los Angeles since the 1984 Olympic Games.

Team Ireland will include 88 Irish athletes, with some 13 from the Mid-West, 40 members of the management team, coaches and nearly 200 Irish volunteers "

Editors Note:

See a full version of this article on www.williamssyndrome.ie - latest news.

We will be following Ashleigh's exploits in the Special Olympics with keen interest throughout 2015. Watch this space for further updates.

Inside this issue:

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Update on Education

Fionnuala Tynan

A big thank you to all the families in the WSAI who helped me with the research I conducted on the education of children with WS. This was the first major piece of research on education of our children in the Irish context. Guidelines for teachers are now on the website. I'm delighted to say that since graduating in September I have been able to spread the word about WS at a number of different venues.

On 8 October I presented a paper at the Mayo Education and Teachers' Conference on how to access the voice of learners with WS or other special needs for research purposes and linked this to a good practice guide for teachers. The presentation was entitled 'From the Mouths of Babes: Accessing the learner voice'. Part of the presentation was giving information on WS. There was great interest among the teachers, lecturers and researchers there on the condition and many had questions to ask afterwards. The WS information leaflets were distributed among the participants and a batch were left in the Mayo Education Centre too so that teachers may pick one up when they do a course there.

On the 19 of November I attended the conference of the National Council for Special Education as a representative of the WSAI. I wasn't presenting, I was just in the audience but wearing a badge with my name and the 'Williams Syndrome Association of Ireland' meant that many people asked me about WS. This conference is attended by SENOs (special education needs organisers), educational psychologists, researchers, teachers, school inspectors, parents and lobby groups so it was good to get WS mentioned incidentally among such a group!

On 21 November to 23rd I attended the European Williams-Beuren Syndrome Conference in Budapest. I presented a poster at this conference on 'The Educational Experiences of Learners with Williams Syndrome'. There was quite a bit of interest shown in this as there are very few published papers on what learners with WS say about their own education. Thank you again to my friends in the WSAI who allowed me to interview them. This conference was of particular interest because it was the first major European conference on WS (or WBS as it's called in Europe!). Myself and Ann Breen have done a synopsis of the main contributions at the conference and they will be presented in issues of this newsletter during the year. There were representatives there from France, Hungary, Poland, UK, Romania, Slovakia, Sweden, Switzerland, Bulgaria, Germany, Czech Republic, Denmark, Italy, Netherlands and Belgium.

On 27 November I was invited to give a lecture in the Galway/Mayo Institute of Technology, to the final year students of Social Care. The lecture was entitled 'Promoting Best Practice for People with Williams Syndrome'. The students were particularly interested in the condition. WS leaflets were given out to all and some were also left in the college campus. Interestingly, myself and Jarlath were at a musical in Castlebar a few days later and I met one of the students. She made it her business to come over to introduce herself to Jarlath. That is exactly what we want to happen!

> *"This was the first major piece of research on education of our children in the Irish context. Guidelines for teachers are now on the website."*

I am half-way through the research on the use of Individual Education Plans for learners with WS of post-primary school age. This is being done in conjunction with St. Angela's College of Education in Sligo. Thank you to all the families who have agreed to participate. This will supplement the information we have on primary-school learners with WS..

At our last committee meeting it was also decided that as an organisation we would like to gather information on anxiety in our family members with WS. See the article on this in this newsletter.

Music Camp Details for 2015

Carmel Daly

Planning for the 2015 annual WSAI Music/ Activity Camp is now underway.

I will be writing to all families this month with full details. Keep an eye on the website www.williamssyndrome.ie and the Facebook page – Williams Syndrome Association of Ireland, for updates also.

Hope to see all our previous campers as well and some new faces there….

Sighile Hennessy On the Drums

Learning to Live with Diagnosis

Deirdre Graham

Our four year old boy, Ed, bounds around the kitchen table, curls bouncing on his head, his big blue eyes filled with smiles. Fleetingly I forget the reality of his situation. Chromosome 7 deletion. That August afternoon as I sat in the children's outpatients waiting room, I didn't realise the impact such a minute number of absent chromosomes would have and how our lives were about to be changed forever. Up to that day number 7 happened to be my lucky number. Now it meant deformity, devastation and disaster to our young family. I looked at my beautiful baby boy and the world literally shifted beneath my feet. The Paediatrician outlined what Williams Syndrome meant for Ed, health problems, growth issues, intellectual disability and a life time requiring care. My only question at that moment, would he be able to go to school? Up until now this was a given, but suddenly it appeared out of his reach. The doctor did not answer this question, wisely I suspect.

It is surprising what memories remain with me of the diagnosis day. I remember it was a beautiful summer's day. Ed was wearing a tiny shorts onesie, meant for a 6 month old, but obviously loose on my 14 month old monkey. A telling sign of his condition on reflection, but hindsight is always clear and obvious. The puzzle that had caused us so much worry and anguish was solved. The ongoing colic, the severe reflux, his failure to gain weight, failing to reach milestones, the delay in crawling and the sleepless nights…. (Oh my, the lack of sleep, how often I had imagined constructing a safe sound proof cradle to hang outside the bedroom window just to steal a few consecutive hours sleep on any night!!), and the reassurance of the odd beautiful open smile. Our son has Williams Syndrome. The diagnosis was a relief of sorts, following 14 months of self-doubt, speculation and increasing anxiety I realised that I wasn't crazy. I had always known that there was something wrong, however people kept reassuring us that he would be fine, that he would grow out of it, it was just colic, and "sure your brother was small". All of this advice was given with good intention as I believe that those around us loved us and probably didn't want to believe that anything could be wrong. The new reality of our situation came as a shock to all of the extended family, Ed being the pebble; he caused a massive ripple effect throughout all of our lives.

Ed and His Bedtime Teddy

The heartbreak of diagnosis is hard to describe. The heart wrenching utter sorrow on the loss of your "normal" child coupled with the immense guilt of feeling that sorrow because he is still the same baby you picked out of the cot that morning. His Dad describes the diagnosis day vividly. It was as if someone rolled in a trolley that afternoon and rolled out our normal baby boy replacing him with a child that looked like him, but with a completely different set of life goals and objectives. The potential of college, independent travel, marriage, children, mortgages were taken out on the first trolley, and an apparently empty trolley was presented. We had no expectations and very little hope that day leaving the doctor's office, but we still had our baby boy.

So began our journey towards living with this new dynamic. We began slowly, deciding to gather relevant facts only about the condition in order to increase our own knowledge about his health needs. I was determined not to act true to form, and resisted submerging myself in literature and greater detail about the condition. I was afraid I would lose sight of my baby boy and his own unique Williams free individual personality. Post diagnosis life began with a succession of medical appointments, both local and in the "big" hospitals in Dublin. Then began the whirlwind of physiotherapy sessions, speech and language therapy, occupational therapy, dietician visits, orthotic fittings, liaison nurse visits, social work visits, psychology assessments and occupational therapy reviews. Our Ed had more professionals interested in him then a formula one car in a pit stop! We were fortunate that the team rallied around us quickly, providing a firm support to our shaky

Ed Graham

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Learning to Live with Diagnosis—Contd.

family, guiding our way as we slowly gained an understanding of the consequences of Ed's condition.

Each child is unique and individual as is every

Ed and Deirdre

child with Williams Syndrome. It is easy to lose sight of the child sometimes beneath the weight of the label, but we remind ourselves that Ed is half me, half his Dad minus a minute part of his 7th chromosome. Sometimes when he apparently is acting out or misbehaving in a certain way, the syndrome can't always be blamed as we recognise family traits in his behaviours! That is what makes him an individual with strengths and weaknesses like us all. In the early days we received some wise advice from an experienced woman. When we asked her what was the best thing we could do for Ed, she advised us to give him confidence, as you would with any child. However we feel he has given us confidence as we face the tough daily challenge of raising him. In turn we instil that confidence and other emotions such as joy and healthy fear in him. He will need it as he continues to face inevitable life challenges.

If someone had told me during those early months that my frail little boy would be walking at 20 months, kicking a ball at 2 years, singing rhymes at 3 years, cycling at 4 years and is currently preparing to start main stream school, I would have shook my head in disbelief, there is no way I would have allowed myself to dare believe them. My expectations were low at the beginning; they had to be to deal with the fundamental disappointment of losing my "normal" baby boy. But year by year our expectations are growing. We expect him to take his coat off, put away his shoes, set the table, tidy his toys, wash his hands – basic expectations of any 4 year old child, but laced with great pride in how hard he works to accomplish these seemingly menial tasks. From the day of his diagnosis our shattered hearts have battled to remain strong to provide him support, patience, care, love and of course confidence. Our hearts will never be the same having known the grief of a life changing diagnosis, but how can our hearts be the same when they swell to unimaginable proportions when he clearly articulates a full appropriate sentence, when he asks to do a "wee", when he takes his coat off without help, when he sings "jelly on the plate" solo at his Montessori Christmas concert! We have not accepted his diagnosis and likely never will, but we are learning to live with it a little more every day, and incidentally, 7 has regained its status as my lucky number!

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Blue Teapot Theatre Company

Alan Keady

I started in Blue Teapot Theatre Company in September 2013. I go there for 4 days a week from 10am to 3pm. for 3 years. There are 7 of us in group and we are called The Teacups and we all get on great together. I do something different every day.

I have learned to use a sewing machine and cut out a pattern, i made a cushion and waistcoat now i am making an Elf outfit for a show. I do costume design, drama, music, non verbal communications, film studies, creative writing, production and back stage and art. I never thought I could do these things but am very happy and can tell my friends what I do. I also do literacy and can use my tablet now to look up things on You Tube. I get to see shows in Galway and also go out to visit music shops to see the different instruments. I have small projects to do for each subject and do an exam each year. I also can get the bus to and from the venue so i am more independent. I really enjoy every day and have new friends.. I hope that there is another course when this one finishes as i love where i am. In February the group went to a theatre in Dublin to see the show Memory Box which was produced by Equinox Theatre in Co. Kilkenny.. I got to meet all the actors after the show and guess what Sighile Hennessy was one of them. It was a great show -Sighile played different parts as a soldier, Passport control Officer and an Official. We had a photo taken of the two of us.. Alan Keady.

Alan and Sighile Hennessy

European Williams Beuren Conference—Nov 2014

Fionnuala Tynan and Ann Breen

The European Williams-Beuren Syndrome Conference was held in Budapest in November 2014. Committee members Ann Breen and Fionnuala Tynan attended on behalf of the association. This conference was of particular interest because it was the first major European conference on Williams Syndrome. There were representatives from France, Hungary, Poland, UK, Romania, Slovakia, Sweden, Switzerland, Bulgaria, Germany, Czech Republic, Denmark, Italy, Netherlands and Belgium. Fionnuala presented a poster at this conference on 'The Educational Experiences of Learners with Williams Syndrome'. There was quite a bit of interest shown in this as there are very few published papers on what learners with WS say about their own education.

A summary of two of the papers presented are provided here and further Newsletters will contain summaries of the outstanding material from the conference.

If you interested in learning more about this material please contact Ann or Fionnuala for more details.

An Overview of the Genetic Aspects of WS and Research Approaches by Dr. Kay Metcalfe (UK)

In 1993 the genetic basis to WS was discovered. This confirmed that WS is caused by a deletion of material on chromosome 7 in the area 7q11.23 and that the elastin gene is affected. There are some cases that are caused not by a deletion of material but of translocation of the material (correct genetic material that has ended up in the wrong place) or by an inversion of the material. The latest developments in genetic testing such as microarray technology (which will tell exactly what genes are missing) has shown that 95% of individuals with WS have a 1.55 MB deletion of 26 genes and about 5% have a 1.8 MB deletion. The microarray analysis is like 80,000 FISH tests it is so accurate! Where there is a bigger deletion the WS condition can be more severe and is more likely to have features of autistic spectrum disorder.

Genetic testing and experiment continues on mouse models. Mice are genetically engineered to have WS so that new treatments can be developed for such aspects of the condition as vascular stenosis. This is also allowing geneticists to map the chromosomes and learn what each gene does. Stem cell research is also ongoing in WS. This raises very significant issues. If you found out your child could have a stem cell transplant would you change him or her?? There is currently no programme of screening a baby in utero for WS and it doesn't look like there will be due to the rarity of the condition and the fact that there are no specific risk factors.

EUROPEAN WILLIAMS-BEUREN SYNDROME CONFERENCE (EWSC)

21-23rd November 2014, Budapest, Hungary

WS_Conference

Oral Health (Care) and Williams-Beuren Syndrome: Challenges and Opportunities by Prof. Dr. Dominique Declerck (Belgium)

This was an excellent presentation that was full of practical advice. Dominique is a dentist and lecturer but is also the mother of young man with WS. Her clinic caters for clients with WS so she has built up great expertise on oral health care in WS.

Individuals with WS can have specific craniofacial and dental characteristics. About 85% of the WS population have fewer contact points between their upper teeth and lower teeth which impacts on their bite and many have spaces between their teeth. The teeth can appear different to those of the general population: they may be smaller with small roots, some teeth can be missing naturally (41% of individuals are missing one or more teeth, 12% are missing 6 or more teeth) and the enamel of the teeth can be thin and deficient (in 19-41% of individuals). They also have excess gingival tissue.

These features highlight the great importance of a rigorous oral hygiene routine and tooth-friendly dietary habits. Prevention is so much easier than cure! Limit sugar consumption between meals. Regular visits to a dentist should be started from an early age, even visits to get the child used to the dentist and to build up a relationship with him/her. You should inform your child's dentist about WS, and particularly your child's anxiety, and fear of loud/unusual noises if it exists. Declerk claims that in most patients with WS, regular dental care provision is possible, including orthodontic treatment. Should your child be too anxious for a dentist s/he may need inhalation sedation or may need to go under general anaesthetic for dental procedures. Be aware of the increased risks for individuals with WS who go under general anaesthetic. The risk is present where the individual has peripheral pulmonary stenosis. This should be weighed up with the dental procedures to be undertaken. You may wish to discuss general anaesthesia with your child's cardiologist or paediatrician. If your child is having an extraction, or any procedure where there will be bleeding an antibiotic should be taken beforehand. This will need to be discussed with your child's cardiologist, paediatrician

Weekend Break

Debbie Branningan

This years Weekend Break will be held on the 8th to the 10th of May in the Ardilaun Hotel, Galway. More details of the hotel are available on the hotel website: **www.theardilaunhotel.ie.** .

This is a great social event where families take part in a number of events organised by the association or just relax and share experiences of living with Williams Syndrome. Closing date for participation has now passed but we do hope that you have booked your place. If not there is always next year.

If you have any queries about our Weekend Breaks please contact Committee Member Debbie Brannigan at 0851652641 or email deboraghbrannigan@hotmail.com

Ceramics Course and Other Things

Patricia Moylan

I go to a ceramics class every Friday from 10am to 12noon in the Stillorgan College of Further Education in Dublin.. I've made two decoration things but mainly what I do is bead and pendant making. Here is how I do the work:

First I make the clay into a pendant or bead. I can make them using which ever design I choose.

Then I put holes in them and wait for them to dry.

Once they are dry they go into a big oven thing called a kiln to be fired. I do not know the temperature.

After they are taken out and cooled I can then glaze them. This is like paint and it comes in many colours and has effects like crackle and there are speckled ones which dry on to the clay like a powder

Once that's done they go in the kiln again and the glaze comes out nice and shiny.

My teachers name is Carol and Thomas, Ciara, Susan, Marion and Caroline are also in my class.

I love Ceramics because making things is great fun. I like to get my hands dirty and I can make the clay into any shape I want. I also like beads and pendants because I like the look and feel of them. You can also make them into lovely jewellery.

A Sample of Patricia's Ceramics Work

I also buy a lot of jewellery. If you are wondering where I get my lovely jewellery from have a look at www.gemporia.com and there are other sites for you to check out such as www.jewellerymaker.com and www.gemcollector.com.

Anxiety in WS

Fionnuala Tynan

Following on from our last committee meeting in February it was decided that we should collate information on anxiety in our people with WS to acknowledge that parents have a huge amount of expertise dealing with anxiety in their child with WS. I am hoping to start this research project at the weekend away. I am currently working on an interview schedule and I plan to interview as many parents as possible at the weekend away. The interview will last between 20 and 30 minutes. Have a think about it before the time, you will not be pressured into participating. Anyone else who is interested can email me and I'll make arrangements to call to you to do an interview or to do it over the phone. Remember, the more people involved in the research the stronger our findings. This research could be a huge support to families who deal with anxiety. Please consider your participation.

2014 AGM

Tommy Moylan

We had a successful AGM for 2014 in November in the Lucan Spa Hotel in Dublin. The meeting reviewed the years activity and we were happy to report another year of solid achievements. Highlights of the year included the updating of our policy and guidelines for the Protection of Children and Vulnerable Adults to include provision for Garda Vetting. All our volunteers for 2014 underwent Garda Vetting and this will continue to be obligatory in future. Another major innovation was the work carried out on Education Guidelines for WS Children. This work was based on research carried out by Fionnuala Tynan with the help of many parents and WS people in the Association who kindly took part in the study. We are delighted to provide an update on Fionnuala's work in this Newsletter and to publish the Guidelines on our website. The meeting also reported on the successful running of our regular activity programme for the year including the Spring Weekend Break, The Picnic in the Park in mid summer and the Annual Music Camp in August. Thanks again to all who helped make these events successful including our many volunteers who give of their time so freely.

Nuala Keady presented the financial results for the year and the meeting expressed thanks to all who helped in fundraising for the Association during the year. The meeting was followed by a presentation on Fionnuala's work on Education Guidelines and a talk by Nurse Wendy, a specialist nurse, who supports families living with WS in the UK. We also arranged for our WS people to participate in a music session and at the end of the day they all got on stage to entertain us. We all enjoyed the special impromptu performance and this was a nice way to close the day. A big thank you to Carmel Daly who organised the event and the volunteers who helped on the day.

Special AGM Musical Performance