PWSAI Summer NEWSLETTER

Hi All

Welcome to our summer newsletter, a bit late I know, when I started writing this the sun had been shining for weeks yippee now it's raining but this is an Irish summer! This newsletter has some info on our 2013 AGM. The upcoming Family day is one not to be missed and is being organised by Margie Walsh she also gave us a lovely piece about her daughter being a rose of Tralee "Rose Bud" we have some lovely pictures of that!

We have some news regarding fundraising we are now the chosen charity of lake view medical more about that inside

A piece on the conference held in Dublin and some feedback we received.

I will also be sharing my one and believe me only marathon experience

We hope exams went well for all those that were doing them! and for all the parents who had to endure them! what`s a few more grey hairs!! .Best of luck with the results!!

Running for PWS

I decided that I would run a marathon for my son Oisin and PWS. A marathon being 26.2 miles!

One of those ideas and you think how hard can it be. Well it was hard but funnily enough the marathon was not hard it was the training before hand was both tough and time consuming. I would run 5 times a week, 3 evenings and 2 early mornings. Really it was my poor husband and the 5 kids that must have found it toughest because me and speed are not related so my long runs (anything over 7 miles is a long run to me) took a long time!!

I thought I would become a lean machine but unfortunately that was not the case as long runs made me very hungry which funnily enough often led me to train harder because it gave me a small insight how Oisin must feel on most days!!

When Marathon day came I was looking forward to it for 2 reasons really, the obvious one being taking part in a marathon but also knowing that this was my last big run and after this I need only run if I truly wanted to.

I completed the marathon in 4 hrs. 29 mins and 32sec and I was thrilled! There was many times during the 26.2 miles that I felt like stopping or even just to walk but I didn't I thought of all of us who work every day with pws and we don't have a choice to give up but only to keep on going. Running a marathon is very like living with someone with any special or additional needs, many stages my pace was good and steady and I felt confident and assured that I could do it and at other times my pace was slow, I struggled and I questioned myself and felt like just giving up, but I had my family and some fantastic friends to support me along the way and to shout me on!

When I got near the finish I saw my guys so I grabbed them over the railing and they crossed the finish line with me a great feeling!!I had a couple of teams in the relay section my 4 brothers had a team, my sister and my sister in law and 3 friends had a team, My niece and 4 of her friends also had a team and my great friend Regina Dorgan also put in a team. We had 2 half marathon runners Fiona O Driscoil and Thomas Barry, running for PWSAI. We had 4 full marathon runners, Ger Holland, Annemarie Buckley Paul Leahy, and myself running for PWSAI. I am hoping this will grow and become an annual event minus me of course, well minus me doing 26.2 miles anyway!!So if you know any runners or anyone looking for a challenge, ask them to do it for PWSAI.At present we have raised €6000 so I am really happy with that. This is a pic of me crossing the line I picked up my four best supporter's Tara Oisin Rory and Grainne, Ray and Fionn were waiting at the finish!!!

Committee Update

The period since our last newsletter has been a busy and productive time for PWSAI. We held committee meetings in January and March and our A.G.M in June. We worked on specific events and initiatives including the following:

1. PWSAI's conference on Behaviour and Mental Health in PWS held in April. More about this can be found elsewhere in this newsletter.

2. Formalising PWSAI Membership. Our membership system in now fully up and running. More about this can be found elsewhere in this newsletter.

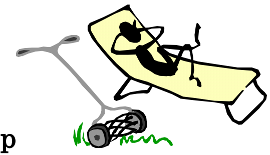
3. Research. PWSAI committee members held productive meetings with academics and medical personnel from TCD, Tallaght hospital and NUI, Galway with a view to encouraging research into PWS. One of the research projects stemming from these contacts is described in more detail elsewhere in this newsletter.

4. Formalising links with other organisations. PWSAI recently joined the Genetic and Rare Disorders Organisation (www.grdo.ie) and applied for membership of the Disability Federation of Ireland (http://www.disability-federation.ie/).

5. Supporting Fundraising. PWSAI was delighted to be named as one of Lake Region Medical's charities of the year for 2013 and to be nominated as the charity for the Ballycotton 10 Road Race. PWSAI also approached corporate sponsors and received one corporate grant of €3,000.

6. Governance. PWSAI continues to make progress in implementing in full the Governance Code for Community, Voluntary and Charitable Organisations in Ireland.

Membership



Our survey late last year indicated a strong support among families for PWSAI to become a formal membership organisation. So we amended our governing documents and since April have been registering members. The cost to join is only €15 for 2013 and 2014 combined.

So, why join PWSAI?

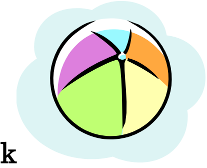
- To strengthen its voice and mandate in seeking to raise awareness and understanding of PWS and to improve the choice and quality of care, education and support for persons with PWS in Ireland.
- To ensure that you can influence the priorities of PWSAI.
- To be connected with others who have PWS, who have family members or friends with PWS or who have a professional interest in PWS.
- To ensure that you receive regular newsletters and other correspondence from PWSAI.
- To ensure that you are invited to PWSAI events.
- To contribute towards the financial costs involved in running the association.

So, if you have not yet joined PWSAI please consider either:

1. Completing the application form and paying for your membership subscription online at www.pwsai.ie OR

2. Completing the attached application form and posting it with payment by cheque to Prader Willi Syndrome Association Ireland, Carmichael Centre, Carmichael House, North Brunswick Street, Dublin 7. Cheques should be made out to Prader Willi Syndrome Association Ireland.

Thank you!



Facebook

Late last year PWSAI finally entered the crowded world of Facebook and since then we have been delighted to see people posting comments and sharing links on our page. We too have tried to use Facebook as a means of sharing information and resources from around the world. Since the beginning of May, for example, we have introduced a Wednesday webLINK feature whereby we direct people to at least one link each Wednesday.

If you are a Facebook user please check out (and 'like'!) our page. You can access it via our website - www.pwsai.ie

IPWSO

PWSAI is pleased to report that two of its current committee members, Marguerite Hughes and Don Tallon, have recently been elected to the board of the International Prader-Willi Syndrome Organisation (IPWSO). PWSAI looks forward to a strengthened relationship between PWSAI and IPWSO as a result of their involvement.

Stem Cell Research

Hello. This is Marguerite from Galway. I recently emailed those of you for whom I have current email addresses regarding a stem cell research project that people with PWS in Ireland are being invited to participate in. For those of you whom I didn't reach by email I would like to describe the research proposal here.

The research being considered would be conducted by Professor Sanbing Shen and his team at the Institute of Regenerative Medicine in NUI, Galway. Prof. Shen is currently developing iPS cells from individuals with conditions including autism and Fragile X. He has offered to extend his research to PWS if there are volunteers willing to participate.

So, what would be involved? Volunteers with PWS deletion or disomy (and one of their siblings who would act as a control) would need to give a blood and skin tissue sample for research purposes. Unfortunately, for licensing reasons, the giving of these samples would have to happen in Galway, but people's expenses in coming to Galway would be covered.

Giving these samples would not personally benefit participants. It could also be the case that nothing of use to people with PWS or any other condition would be found through the research. However, clearly the more research into this area that takes place then the more likely it is that ultimately something will be discovered.

If you're interested in finding out more about iPS cells in general I found the short video available at this link really informative and inspiring : http://www.youtube.com/watch?v=Q9-4SMGiKnE&feature=player_embedded

If you would like more detailed information about Prof. Shen's proposed PWS iPS cell research please contact me, Marguerite, at secretary@pwsai.ie or on 087 9621681 at any stage and I will happily pass on full details.

ELLEN FLAHAVAN SELECTED AS WATERFORD ROSE

On Friday night, Ellen Flahavan was selected as the Waterford candidate to go through to the next stage of the Rose of Tralee competition. The Waterford final was held at the Woodlands hotel, in front of a large crowd with sixteen girls taking part in the competition. Ellen is a 24 year old teacher and will now contend for a place in the final, at the 2013 Rose of Tralee Regional Festival, which will take place at the Portlaoise Heritage Hotel, Co Laois from 29th May to 2nd June 2013.

Some pictures below from last Friday night:

Ellen was supported and accompanied on the night by her six year old ‘Rose Bud’ - Mary Rose O’Connell (Margie Walsh’s daughter)!

The 2013 International Rose of Tralee Festival will take place from 14th to 20th August 2013 and will be hosted by Dáithí Ó Sé.

We all wish Ellen the best of luck in the June Regional contest and hope to see her in "the Dome" in Tralee in August!

Áoife's Pre School Battle.

In New Ross we are in the lucky position of having Cunamh Mhuire Special Needs Pre School. This school is run by the Co. Wexford Community Workshop with the HSE providing therapists. Áoife has been on the list for Cunamah Mhuire from only a few months old. Unfortunately for her the HSE in an effort to to appear more equitable in allocating spaces to the Pre School introduced a Priority Rating form in 2013 and we were informed in February 2013 that Áoife would not be offered a place. We wanted Áoife to attend this school specifically as she would have regular access to Speech and Language Therapy, Occupational Therapy and Physiotherapy. Up to now, access to these services have been sporadic at best. The therapists themselves are excellent and do the best they can with the time allocated. Whilst I am aware some of these services don't even exist in some parts of the country I still don't think we should feel lucky that we had access to inadequate services.

I felt strongly after seeing a copy of the form used to prioritise Áoife that it was unjust and being used as a tool to prevent access. In New Ross we have 8 mainstream Pre Schools and I was asked by some if I would be able to sit with Áoife if she were to attend when I explained her needs. We were basically in a position where she couldn't attend a mainstream pre school and was being prevented access to the only special needs pre school in our area. The form provided boxes to be ticked to certain questions relating to need. Aoife doesn't really fit in a box and the questions asked didn't really cover what her needs were. The therapists involved in her care tried their best to put forward how signifcant they felt her needs were and how beneficial attending Cunamh Mhuire would be to her but their opinions didn't fit the boxes provided in the form.
On 4th March 2013 I wrote to the General Manager, Local Health Office, Wexford and copied the letter to the Manager for Disability Services in Wexford to highlight my concerns and

requesting them to reassess. I filled in the HSE complaints form, Your Service Your Say in an effort to get some sort of response. I enclosed copies of reports from Aoifes therapists, SLT, OT and Physio. I asked her creche to write a letter detailing how they find her and what her needs are when she is with them. I also contacted her Paediatric Consultant for a letter detailing her condition. I was so annoyed I also wrote to Enda Kenny and James Reilly highlighting the deplorable level of resources for disabled children in our area. I then contacted by letter various TD's looking for their advocacy on Aoife's behalf. All said they were delighted to help, some proved to better than others.

I spent a number of weeks playing phone and email tag with the HSE. I also made sure to follow up with the local representatives to find out how they were getting on. I kept a record of every phone call or contact I had.

I finally received a response from the General Manager, Wexford Local Health Office on 19th April 2013, some 7 weeks after writing to the HSE, to inform me that the form being used was equitable and had been designed by a multidisciplinary team. She said they regretted they could not meet Aoifes needs in a more comprehensive manner but were restricted by limited capacity. She suggested I contact the National Advocacy Unit if I wanted to appeal. She ignored all contacts by the various public representatives up to this date. I could not accept this. I contacted the Office of the Ombudsman for Children and filled in a complaints form. I also contacted the National Advocacy Unit requesting them to look into the issue. The National Advocacy Unit is part of the HSE so it is basically asking the HSE to investigate itself. I felt though that all procedures needed to be followed. I also wrote back to the manager and informed her that I didn't accept what she was saying and notifying her of my intention to speak to whoever I needed to in order to ensure Áoife secured a place at the Pre School.
The Ombudsman contacted me on 3rd May 2013 to say they would carry out a preliminary examination and had contacted the HSE looking for various information regarding access to Pre School and disability services. They particularly asked them to demonstrate how the form was equitable and what alternative provision was available for children who cannot secure a place in a playschool that is appropriate to their needs. They also encouraged the HSE to resolve the complaint locally.

I received a letter from the HSE on 27th May to tell me the Prioritsation form being used was being amended having taken my feedback into consideration. Another meeting took place to prioritise children and following this Aoife has been offered a place from September 2013. We are very happy and praying that the staffing levels aren't reduced even further or another family will be in our position next year.

The National Advocacy Unit wrote to advise me that due to backlog they still hadn't assigned anyone to look into my complaint. I contacted them and told them things had been resolved thanks to the intervention of the Ombudmans for Children.
From my perspective I felt it was important to follow procedures no matter how ridiculous I felt they were, ie contacting the National Advocacy Unit. I kept a note of every phone call I made and every letter I sent and received. I requested copies from all Aoife's therapists of her progress to date. I made certain that any statements I made were based on fact and not on emotion. I felt strong in my conviction and knew that the HSE were initially going to not do anything and hide behind the limited resources story.

I accepted that it was probably going to be a long fight but I was willing to keep going for as long as it took. In the end I was amazed it came to a good conclusion so quickly. I fully expected to be still going with this next year. When I felt myself getting irritated I stepped back bacause there were times I thought I would burst with frustration. Unfortunately though I know this was only one small battle and there is going to be so many more just to make sure that my child is given the same chance as every other child to reach her full potential. In Ireland children with disabilites are getting a raw deal. It is so bad here I have heard of one family from Peru moving back there as the services they require for their daughter is better over there. I worry that despite the moves forward in treatments and therapies for people with PWS, living in Ireland and the lack of services available to them here will negate any strives forward.

PWSAI Family Support Fun Weekend

Hope you are free Weekend of the 4th & 5th of October ????

Venue: 4 Star Newpark Hotel Kilkenny

Newpark Hotel
KILKENNY ★★★★
With escape Health Club & Spa
A FLYNN HOTEL

A Castlecomer Road, Kilkenny, Ireland
T 056- 7760500

Date Confirmed: Friday 4th to Sunday 6th October 2013

The Flynn hotel group have kindly offered the PWSAI generously reduced room rates as well as many other add-ons such as Crazy Cats Kids Club, private function rooms etc. for our annual PWSAI family gathering. All welcome siblings, grandparents, carers etc.

Special PWSAI Room Rate: €108 per room/night including breakfast. A family room can hold 2 adults, 2 children under 12 and a travel cot. Special 2 Night Rate for Fri & Sat €200.

For those with more than 2 kids: Rate for 2 Interconnecting Rooms: (2 adults & 4 Children under 12): €160/Night (€300 for 2 nights).

Please book your accommodation directly with our Event Organiser in the Newpark Marcella Kavanagh email: Mkavanagh@newparkhotel.com, Telephone 056-7760593 (Marcella works Tue-Fri). The hotel is currently holding rooms and we would really appreciate if you could book by 30th August if able to attend.

Other costs for Lunch on Saturday & Dinner on Saturday night will be confirmed nearer the time. *We are doing our best to keep costs to a minimum!*

We hope to send final details in September and we look forward to hopefully seeing you all in October.

Best Regards

Margie

The Plan

(Please note all provisional at this point, so please manage expectations as we do not want to disappoint anyone! **Also I am working on the plan to improve it for the older PWS people so happy to take your suggestions)**

Friday 4th October:

Check in at your leisure and enjoy hotel facilities.

We also hope to have PWSAI special SPA rates for those who fancy a treat! No doubt some of us will find the bar for our annual chats and if possible I will see if we can have some music for those who like to dance or a movie for others.

Saturday 5th October:

Breakfast at your leisure

10.30am Meet in the Lime & Larch Room

11.00am-1pm Crazy Cats Club (storytelling, disco, arts, crafts, toys & magician)

1.00pm Lunch Soup & Sandwiches, Tea & Coffee (Private area in Restaurant)

2.00-4.00pm PWS Swim Camp (Fun in the pool for our PWS people, if interested)

Free time for siblings or alternative activity TBC

4.00-6.30pm Free Time

6.30pm-late PWS Dinner Dance: Rossmore Suite

We hope to have a private function room for our first informal dinner dance kicking off about 6.30-7pm with a meal, followed by some awards for our PWS high achievers in 2013, followed by some music and fun. So get your glad rags ready and dust off your dancing shoes. (We hope to have the Crazy Cats Club from 7-10.30pm in room next door for light entertainment for those who might not want to sit through the meal!)

Sunday 6th October

Breakfast at your leisure

11am-1pm Final activity will be confirmed nearer the time if we have the funds!

The plan cannot be developed further without your help and we know you will have loads of questions especially on costs etc. but we can only answer them if you help us to get an idea of numbers. To this end can I ask you to complete the following form and return it via email or post by 30th August to Margie O Connell, Knocknagranagh, Dungarvan, Co, Waterford

Email: mwalsh@flahavans.com or text information to 086-3843840.

1 Name of Person with PWS:____

2 Age of Person with PWS:__________________ Date of Birth: ____________

3 Do you think you will attend the PWSAI weekend being organised for 4-6th October? Yes/No

If No you do not need to complete or return form unless you wish to make a suggestion for future events.

4 If Yes how many people would be attending with the above person? No Adults: ______ No Children ______ (Under 12) Have you booked your accommodation with the hotel? Y N

5 Is the person with PWS interested in the Swimming Pool Activity on Saturday 5th October? Yes No ______

6 How many people would like **lunch (Soup & Sandwiches) on Saturday 5th** with their special person? **(Cost TBC)** No Adults: ______ No. Children: ______ Ages of Children ______________

7 How many people would like to attend the **Dinner Dance on the Saturday night** with their special person? **(Cost TBC)** No Adults: ______ No. Children: ______ Ages of Children ______________

8 Do you think your party will stay at the hotel for 1 night or both nights or not at all? 0 1 2

9 Please give us your special person's achievement of note over the past 12 months and ideally a picture/video clip that we can use on the night? Achievements can be of any nature e.g. Mary Rose's 1st Day in Mainstream School or Mary Rose learned to swim

____

____

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____

10 Can you consider any way of fund raising for PWSAI to reduce costs for all the attendees or come up with a good spot prize or 2 for the dinner dance? (No Pressure….no need to answer but all donations accepted!)

11 Any further Comments/Suggestions: ____

____

____

Name: ____

Relationship to PWS Person; ____

Contact Number ____

Email address ____

Home address ____

The Future is Bright: Parents Program (0 to 5 yrs group: Saturday)
IPWSO Conference 2013 – Emma Walsh

Our son Henry was diagnosed with Prader Willi Syndrome on the 25th July 2012. He was just two weeks old. Like all parents who receive a diagnosis, myself and Rory went through all the stages of grief; denial, anger, bargaining, sadness and acceptance. Once we had accepted that PWS was now a big part of our lives, we decided we were going to stay informed. When Rory first told me about the conference, I was a little hesitant in deciding to go. Even on the morning of the conference, I was anxious, nervous and feeling overwhelmed but within minutes of meeting other parents and hearing their stories, I knew we had made the right decision. We attended the Parents Program (0-5yrs) on the Saturday. We both left wishing we had been able to stay for the whole weekend. We're already planning for the next IPSWO conference in Canada in 2016.

Prof. Maithe Tauber (Paediatric Endocrinology team, France) shared a lot of information about the early days of feeding difficulties. Most children with PWS are tube fed and this can often lead to worry about missing out on the bonding that comes with feeding a baby. Prof. Tauber encourages plenty of face to face time, sitting with your baby on your lap, stroking their face and copying any noises that are made.

PWS requires multidisciplinary care right from the beginning:

- Physiotherapist
- Occupational therapist
- Speech and language therapist

- Dietician
- Paediatric endocrinologist.

Dr. Suzanne Cassidy (Clinical Professor of Paediatrics, USA) shared with us the importance of educating the people who are caring for your child. Most will have heard about PWS but few will have cared for a person with PWS.

Physiotherapy becomes a big part of day to day living with PWS. Motor milestones are delayed due to low muscle tone; babies will sit around 12 months and walk around 24months. Physiotherapy will make your child stronger. Dr. Harold van Bosse, an orthopaedic surgeon (USA) specialising in PWS spoke to us about Scoliosis. Early screening is very important. Once your baby can sit without support, he advised to start screening for Scoliosis. X-rays should then be taken every 6 to 12 months.

- If a curve of 10 degrees is detected, he advised non surgical treatment.
  - Treatment with brace
- If the curve is over 15 degrees, surgery is the best treatment.
- Treatment of Scoliosis is very very important as it can affect heart and lung development.
- He emphasized the importance of tummy time to help strengthen the muscles in the back.

Fallen arches (flat foot) are common in PWS. Dr. van Bosses advised the use of a small ankle brace. However he did say that the big clunky shoes that are often prescribed are not required. As long as the ankle is in a brace, a light canvas shoe (perhaps a half size bigger) is better.

Hip Dysplasia is also common in PWS due to low muscle tone. X-rays should also be taken as soon as possible. The main aim should be to get the child walking in any way possible. Alignment and posture can be looked at after.

Everyone who presented mentioned growth hormone therapy. However Dr. Shankar Kanumakala, a paediatrician in Brighton spoke in depth about it. The most difficult step all parents worldwide have is obtaining growth hormone for their child. He emphasized how important growth hormone therapy is but it is not the complete treatment (Phsyio, OT, and SLT). Initiation and monitoring of growth hormone therapy must be carried out by experienced professionals. A sleep study must be carried out to begin with and also close monitoring for Scoliosis throughout treatment. There are many advantages of growth hormone therapy in PWS:

- Promotes growth
- Enhances muscle mass
- Stabilises bones
- Improves cognitive/motor development

It is also important to be aware of when to stop growth hormone therapy.

- Linear growth plateaus
- Worsening Scoliosis
- Worsening morbid obesity
- Worsening Obstructive Sleep Apnea

As with any treatment, there are risks associated but with close supervision it is a safe treatment with many benefits. It is important in day to day life to promote physical activity, limit unnecessary calories, prevent excess weight gain and support development.

Other health issues to be aware of include;

- A high pain threshold
    - Make sure it's on your child's record if you notice that have a high pain threshold compared to other children.
- Poor temperature control.
    - It is a good idea to let your child's doctors know about this.
- Sticky Saliva weakens tooth enamel
    - Good dental care is advised from the 1st tooth appearing and as your child grows regular trips to the dentist will be important.
    - One tip is to rub the toothpaste in to the brush, to prevent eating of the tooth paste and also to be aware that bottled water does not contain fluoride which is important for healthy teeth.
- It is also advisable to have your child's eyes tested regularly for detection of a squint by a ophthalmologist from an early age
- Undescended testes

Dr Suzanne Cassidy and Dr Anne Livesey (Paediatrician, Brighton) both spoke on behaviour in PWS. With regard to food, the advice from everyone was the same: Consistency!

- Rules must be put in place and these rules must be the same for home, school, minders, and Granny's house; everywhere!
- There needs to be a consistent approach to food before food is an issue.
    - Put food controls in place early, rather than later
- Consistency lowers anxiety levels for the individual with PWS
- A routine around food is important
    - Set meal times
    - No 2nd helpings
    - Once you finish your plate, the meal is over
    - No food on the table, serve from kitchen
- No means No
    - Never give in to food requests.
    - Giving in, will teach the child that throwing a tantrum will get them what they want.
- You should request a Specialist Dietician referral from birth;
    - Needs to know about PWS
    - Regular reviews of meal plan
        - Filling in a food diary
        - Dietician can check child is getting everything that need nutritionally.
- Whole family approach
    - Healthy eating for all
    - Siblings should not ask for more in front of person with PWS.

- Don’t eat in front of the person with PWS, if it is not their scheduled meal time.

While the routine around food needs to be solid, we were advised that in other activities a more flexible routine is better. This will help the person with PWS be more accepting of change. As a parent it will be important to anticipate change and prepare your child for it.

Exercise is important for everyone but for someone with PWS it is crucial. It is important to have a positive approach to exercise, incorporate it into daily life. Swimming and horse riding are brilliant. Dr. van Bosse focused on the benefits of horse riding, as it’s great for back strengthening and balance.

One of the most important things we took away from the conference is education. People in your child’s life need to understand PWS to help understand your child. It is up to you to educate teachers, doctors, minders and family about PWS. Repetition is the key. You can’t tell people enough about the importance of a consistent routine around food. It’s essential to have high expectations for your child but to keep in mind their developmental age.

Dr. Tony Goldstone (Senior Clinician Scientist, UK) discussed drug and surgical treatments against hyperphagia (constant hunger) from the past, present and future. While there is still no cure there is some promising research being carried out, especially with regard to the gene in PWS that leads to increased appetite.

Dr. Suzanne Cassidy signed off with this.

The future is bright for PWS. So much has changed and so much will change. It is important not to worry about the future. The fact is that your child will have completely different teenage and adult years compared to people with PWS today, due to the increase in awareness, research and medical understanding of PWS.

Always one day at a time. Embrace every moment.

So that’s all we have guys thanks to all who contribute the newsletter, It won’t survive without you giving me info.MAKE SURE TO FILL OUT THE INFO FORM AND REPLY TO MARGIE REGARDING THE FAMILY SUPPORT DAY!

Best of luck to those going back to school or starting school or whatever new adventure starts for you in September

Remember the Association is here to help if you need info or think we could help pls contact us through Ann Grassick 0879354914

Thanks

Ann O Neill