



## A note from our Chief Executive, Sorcha McPhillips

Hi to all our service users and friends.

It's been a while! I've only recently returned to work on a part-time basis. I'm going to share a personal story because you just never know who it might help.

On the 23rd of September 2022, I spent the evening with my two wee girls who were 18 months and 3 and a half at the time attending Culture Night events. We sang, danced, played drums, had a picnic and then went home. My husband was working overnight so it was just the three of us. I'd gone to bed with the baby around half ten and I started to get a severe headache in the base of my skull. I was prone to migraines so assumed this was a different type than usual and so just tried to sleep through it.

My husband returned from work at 10 am and I had to send my eldest daughter to get him as I was unable to sit up in the bed. My left arm and leg felt floppy, I had a pain in the base of my skull into the back of my neck and I just felt incredibly tired. I figured it was a 'hangover' from the migraine and I'd slept awkwardly. I spent another 24hrs in bed before going to A&E as the symptoms hadn't improved. The hospital ran some standard tests but didn't do any imaging of my brain and concluded I had a type of migraine, discharging me with tablets for dizziness.

After a few days, I felt things were not right, my left hand was awkward and I was totally wiped of energy and so dizzy. Eventually, I was referred for private scans of my neck and brain. In the meantime, I continued working and the last time many of you would have



seen me was at our family event on Oct 1st in the Armagh City Hotel. I got through the event as best I could but again was completely wiped of any energy. It was a very scary time as I went back and forth for more scans until eventually it was determined, a month later that I'd actually survived a stroke caused by a clot that got shunted through a hole in my heart into the base of my brain- the cerebellum which controls balance and co-ordination. This was a huge shock as we had no family history and I had no obvious risk factor and I was only 41.

I later learned that 1 in 4 strokes occur in people under the age of 65 and some like mine can be a freak event caused by an otherwise harmless hole in the heart that never sealed over after I was born. The good news was that I was recovering from a life-threatening event and it's scary to think of all of the other ways it could have gone for me. I consider myself incredibly fortunate and am so grateful to still

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have the chance to watch my girls grow. I've spent the last year learning about the brain, undergoing counselling, doing rehab which includes meditation, cycling, and yoga, and trying to better understand my own limitations and adapt to them. It is a constant learning curve but I am striving to find the right balance between self-care, family, and the work I am so passionate about.

I am delighted to be back at the helm of HDANI, albeit with a reduced workload, with the help and support of our team of staff and trustees and look forward to seeing you soon.

## Sorcha

### HDANI Staff team updates

There have been a number of changes in the staff team since our last Newsletter meaning we have been able to support our Service Users all over Northern Ireland. Sorcha our CEO who many of you will know has unfortunately had to take some personal time to deal with some health issues, we wish Sorcha all the very best and look forward to her being back to steer our ship with her hard work, determination and giving nature when she is back to full health, in the meantime I am sure you will join us in wishing Sorcha all the best for a speedy recovery back to full health. Ashley, our Youth Awareness and Events Officer finished as a full-time employee for personal reasons but keeps in contact and helps us when she can with different events for the younger HD community.

We have been joined this last year with three new Family Support workers. For those who aren't aware, the Family Support worker's role includes providing information, emotional and practical support to those affected by HD. If you have not used this service before please don't hesitate to contact us to see if we can support you in any way. We can offer support via telephone, video call or face to face.

Our new team has settled in very quickly and have been providing a vital service to our service users.

To learn more about stroke in younger people visit <https://differentstrokes.co.uk/>

Act **FAST** and call 999.



Facial  
weakness



Arm  
weakness



Speech  
problems



Time  
to call 999



### Goodbye to Zélie

We are delighted to congratulate Zélie on her recent graduation in Bachelor of Social Work. Unfortunately, this has meant Zélie will be moving on to pursue this avenue of work but we would like to thank her for all of her hard work and support over the last few years and we wish her all of the very best in her new career pathway.

**Zélie** As I reflect over the last five and a half years I feel honoured and privileged to have been able to support and get to know many individuals and families impacted by Huntington's Disease. Your endless strength and resilience is an inspiration to many and I thank you for allowing me to be part of your HD journey. As I move into the world of social work I will bring with me HD awareness and a hope for a cure one day. Lastly, I would like to wish HDANI all the very best for the future and thank Sorcha, all of my colleagues over the years and the Board for your support. Keep up the great work!



### Henna Cooke

Henna joined us as a Full Time Family Support Worker in June 22. She said;

*"I am delighted to be joining HDANI and hope to provide practical and emotional support to families, carers and health professionals. I have a strong background with over 8 years experience of working with a variety of people with complex needs and their families, mental health and physical disabilities, acquired brain injury and autism. I have worked with families in challenging situations and made a positive difference through supporting them. Prior to working with HDANI, I have worked as a Lead Practitioner managing mental health and delivering behaviour interventions. I also have a Master's in Forensic Psychology. I enjoy volunteering and working alongside the Dogs Trust and tutoring horse riding lessons."*

Henna is currently on maternity leave with her new son William so we send her our congratulations.



Congratulations to Henna on the safe arrival of William Gordon Dillon born on 22/06/2023 at Antrim Area Hospital at 6.57pm weight 5lbs 5oz.



### Katie McClean

Katie joined HDANI in June 2022 as a Full Time Family Support Worker. Katie has a range of health and social care qualifications and over five years experience working closely with people with complex needs, physical disabilities, mental health issues and brain injury. She loves to meet new people and is always up for having some fun. She is really looking forward to getting to know everyone and learning from the organisation.

Hi everyone my name is Katie, I joined HDANI in June 2022 and by now have met quite a few people involved with the charity through support groups, events and youth days. I am originally from Carrickfergus but recently moved up to Belfast. I attended the Northern Regional College and qualified in Level 3 extended diploma in Health and Social Care, Level 4 Diploma in Adult Care and Level 5 Diploma in Leadership for Health and Social Care. As a teenager, I volunteered with Mencap and in a local youth club before beginning my first role as a support worker with Sense NI. I have greatly enjoyed my time so far with HDANI and have had the pleasure of meeting so many amazing people across NI who are touched by Huntington's Disease. I had the amazing opportunity to attend the Huntington's Disease Youth Organisation Youth Congress in March where I met people from all over the world who are affected by HD and professionals in the HD community. My favourite part of my role is being able to help people in any way I can, even if that may be through just offering a listening ear. In my free time, I enjoy spending time with family and friends and being outdoors.



## Mairead Mc Cann

My name is Mairead Mc Cann and I am from Dromore, Co Tyrone. At University I graduated with a BSc in Criminology and Criminal Justice and a Post grad Diploma in Counselling and Therapeutic Communication.

Six years ago, my partner and I rescued our dog Paudi after a road traffic accident, and he is our world. Paudi is full of energy and loves to go out walking with us at least three times a day. I enjoy spending time with my family, going out for walks with Paudi, going to as many beaches as possible and going to the gym.

I have over ten years of experience in the voluntary and community sector. I have been working as a Family Support Worker with HDANI since September 2022. For me, supporting people and families is very rewarding, as it allows me the opportunity to help to try and make a positive difference in people's lives who need that extra help and support.

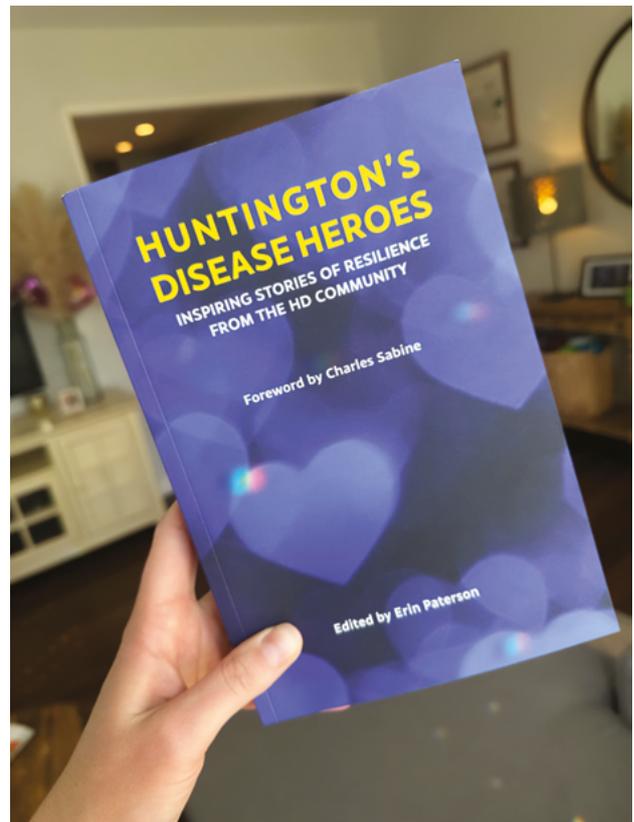
Working as a Family Support Worker can be challenging; however, it can also be very satisfying to see a positive impact on people's lives. As a Family Support Worker, my goal is to give the best version of myself to support each individual and their families, by always giving 100 per cent to each individual and their families I support.

## Book Release

### Huntington's Disease Heroes

We have been delighted this year to be able to share with you the release of a book called "*Huntington's Disease Heroes*" which includes a contribution from our friend and colleague Ashley Clarke. This book contains stories from 26 authors who have been affected by HD including Ashley's own story among others to show the strength and resilience from the Huntington's disease (HD) community.

Huntington's Disease Heroes includes stories from a scientist who switched from cancer research to studying Huntington's after finding out her mother had tested positive for the disease, a young man whose biggest fear was testing gene negative and not inheriting the disease, and a woman who grew up in survival mode, unable to express her grief. We learn about the ways their lives are better off because of the hardships they have faced. By sharing these intimate parts of their lives, they are taking away the fear and stigma of living with Huntington's in their families. They are giving a voice to this unheard rare disease community and empowering others to speak up.



## Social events

### Reconnect and Recharge

We were delighted this year to be able to meet up face to face for a good chat, catch up and relaxation. In October 2022 we held our "Reconnect & Recharge" event in The Armagh City Hotel on Saturday 1 October. This was a relaxed event giving people the opportunity to try some 121 treatments such as massage and reflexology and learn about Mindfulness, Self reflexology and Creative Clay making during the day or if they preferred to enjoy the surroundings of the hotel, take a sauna, steam room and swim or just sit and have a natter and chat to friends and meet new people. The evening brought everyone together for dinner and dancing.

We are looking forward once again this October to meeting up, catching up, relaxing with a treatment, attending a workshop, just taking some time out and having a bit of craic at the dinner dance. Keep an eye on our Facebook page for some great photos of the day!



## Christmas Lunches

We held two Christmas parties in December, one in Cookstown and one in Belfast, these were well attended and good craic was had by all along with our delicious meal! It gave us the opportunity for staff, Board members and service users to all meet up ahead of the busy festive season in a supportive environment to discuss our plans for the upcoming season, what we still had to do beforehand and our expectations and hopes for the festive time.



# HDANI Support Groups

Our support groups have been able to recommence in Face to Face. Our support groups are open to patients, carers, partners, family members, those at risk of HD or those with a loved one impacted by the disease. Our groups are facilitated by a support worker and keep members up to date with our work, events and services and also act as a point of information, practical and emotional support for members.

The Support groups are run in Omagh, Newry, Maghera and Belfast generally on a monthly basis.

For further information on where your nearest group meets or when the next session is, please contact your Family Support Worker or check our Facebook page events page.

**There are no Support Groups for October 2023, due to the HDANI Social Event on 21st October at Armagh City Hotel.**

## **Omagh Support Group**

- 13th November 2023 - 5pm - 6pm at the WAVE Centre, 2 Retreat Close, Omagh BT790HW.

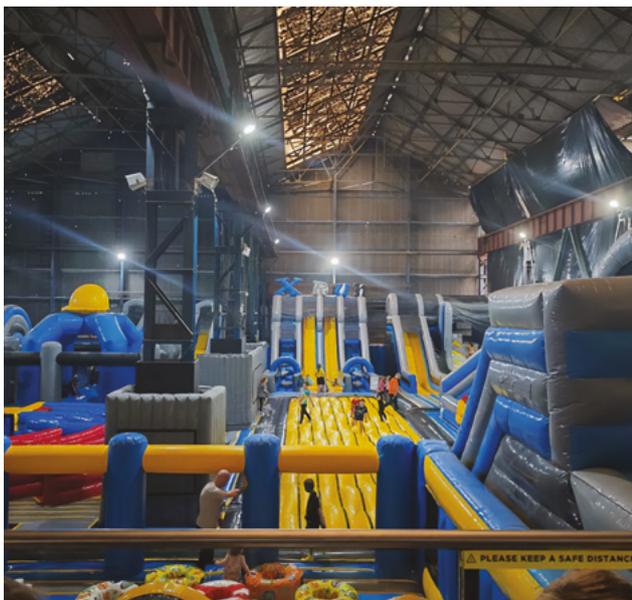
## **December 2023**

- Christmas lunch in December - Dates to be confirmed



## Youth events

We have had some great events with these youths who are affected by HD. These events are a great opportunity for the youths to relax and have some good fun whilst also providing them with a safe environment to get some further information/support for any worries/stresses they may have and learn about how HDANI can support them.



Over the last year or so we have visited **We are Vertigo**, had a great Halloween event at **the Ulster American Folk Park** and this year we were at **The Jungle NI** for a good bit of craic as you can see from the photos.



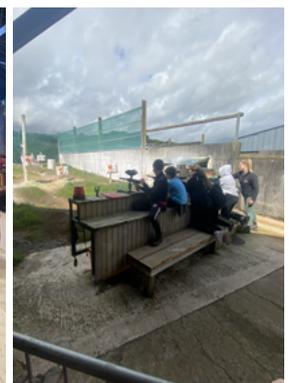
Our young people and parents experienced Halloween at the Ulster American folk park for a fun-filled day of exploring how people of rural Ulster lived 200 years ago. Ashley was delighted to see jiving in the barn and everyone joined her for a jive session! The rain didn't stop us from having a great time.



At The Jungle our activities included wall climbing, archery, rodeo bulls, bungee trampolines, pedal boats, quad train rides and more! We then had an icebreaker session as well as youth-appropriate HD education with Grant Walker, Youth Service Lead from the Scottish Huntington's Association.

Our young people had an adrenaline packed day at The Jungle NI, taking part in Zorbing, riding hoverboards, quad rides, paintball, rock climbing, archery, mechanical bull riding and trampolining followed by a much needed lunch break and some HD group work with our special guest Grant Walker from the Scottish Huntington's Association. Our young people worked brilliantly in teams and had some great talks about HD

At We Are Vertigo our young people had fun on the inflatable park, bouncing around and trying out the obstacle course. Ashley joined them in the inflatable park while Katie and Henna got to know some of the families after just beginning their family support worker roles in HDANI.



## HDYO Youth Congress

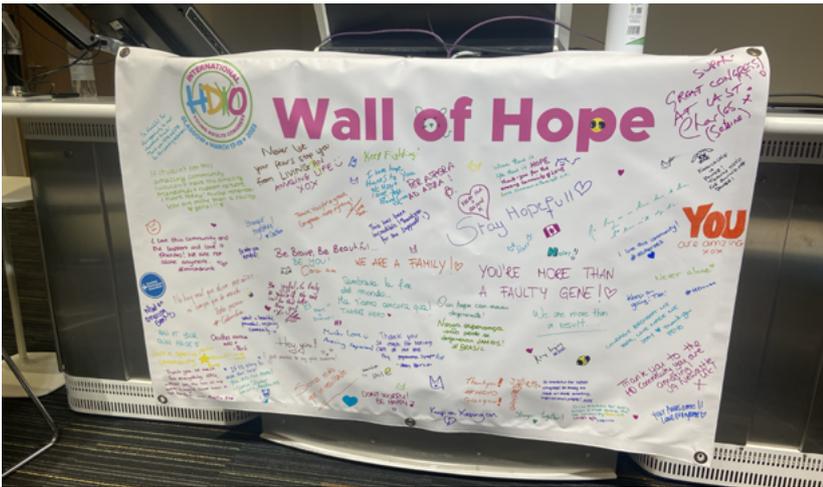
Henna, Katie and Ashley got to attend the HDYO Youth Congress in Glasgow on March 17th to 19th. This was HDYO's first in-person congress! They had the opportunity to meet people from all over the world who are touched by HD, as well as other professionals and even researchers. They attended sessions such as HD 101, scientific updates, talking to kids about HD and research updates. The Ask the Experts Panel was a highlight for everyone from mental health to personal experiences to the latest in science, they got to join HD's notable community members who answered questions about HD. Panellists included Dr Ed Wild, Dr Bonnie-Hennig-Trestman, Carly Evans, Matt Ellison, Dr Ferdinando Squitieri, Astri Arnesen, Dr Olivia Handley & Dr Selene Capodarca.

The speakers for the event were amongst some of the most well-known names in the HD community. They came from a range of groups including young people, families, pharmaceutical companies, researchers, genetic counsellors, neurologists, psychologists, HD Associations and many more! Medical professionals, researchers and pharmaceutical companies updated the Congress on the very latest HD research and trials.



Those who attended had an amazing time while gaining an insight into the different ways people with HD experience life. All of the sessions were recorded and are available to watch online on the HDYO YouTube channel.







## Tipping the balance; new insights into HD genetic modifiers

A new study from researchers at Thomas Jefferson University delves into the details of how genetic modifiers of Huntington's disease work.

By Dr Rachel Harding August 31, 2023 Edited by Dr Jeff Carroll

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**G**enetic modifiers can influence when HD symptoms begin. Some of these genes encode for different types of molecular machines whose normal job is to repair our DNA when it is broken or damaged. A recently published study from scientists at Thomas Jefferson University uncovers details of how these molecular machines help repair damaged DNA structures that can occur in HD, revealing a complicated balancing act.

In this article, we explore what the scientists found, how this can help us understand how different modifiers work to alter the path of HD, and ways these new insights might guide development of new therapies.

### Genetic modifiers of HD change the age at which symptoms appear

Every case of HD is caused by the same genetic change, the extension of a long stretch of the letters "CAG" in the Huntingtin gene. An intriguing mystery in HD research has been the fact that folks with the exact same CAG number can often start to get symptoms at very different ages.

To better understand why this is the case, in a number of studies now, scientists looked at DNA samples from thousands of people with HD and looked to see what small letter changes in their DNA code tallied with symptoms starting earlier or later in life.

The genes they identified in these studies are called genetic modifiers as they modify the course of HD, from what we might expect based on the CAG number alone. Interestingly, many of the genes identified in these modifier studies encode molecular machines (proteins) whose normal role in the cell is to repair DNA when it is broken or damaged.



*Molecular machines involved in repairing damaged DNA play an important role in how HD works*

Two such modifiers are FAN1 and MSH3, which are the focus of this research study. However, MSH3 doesn't work on its own, it has to be together with another molecule called MSH2. One way to think about this is to consider how we make bread; yeast on its own is not enough to make the bread rise, it needs to be together with water and flour to be active and work properly. Similarly, MSH3 needs MSH2 to work, and the assembly they form together is called MutS Beta which is what Pluciennek and colleagues studied in their experiments.

## DNA repair is a double-edged sword

The huntingtin gene contains a long string of "C-A-G" DNA letters repeating over and over. In people without HD this CAG number is usually less than 35, but in people with HD, the CAG number is more than 35.

Long strings of the CAG letters in DNA code can make strange shapes and structures with mismatches in the DNA helix, some of which are called extrusions. DNA damage repair machines recognise and work on these mismatches and extrusions, to try and restore them back to regular looking DNA strands. If cells fail to repair their DNA correctly, a number of bad things can happen, including the development of cancer.

Sometimes, these molecular machines are rather sloppy and can actually make things worse, adding in more CAGs into the huntingtin gene, a process called somatic expansion. In particular, MutS Beta has been shown to jump onto CAG extrusions and can make long CAG repeats even longer over time. On the other hand, FAN1 does a much better job of chopping out the damaged bits of DNA and ensuring the DNA code is faithfully maintained with no additional CAGs.

## The battle of the molecular machines!

In this new study, Pluciennek and colleagues investigated how different molecular machines, FAN1 and MutS Beta, get recruited to these CAG extrusions and how they repair them.

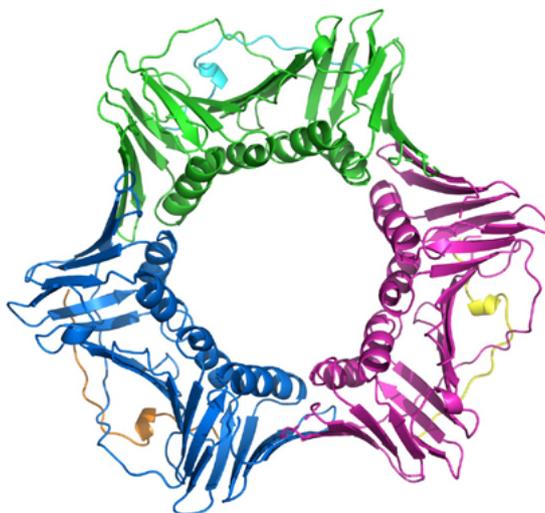
First, the team showed that FAN1 can work on the CAG extrusions, but not on its own; other DNA repair proteins need to be present too and the chemical conditions have to be just right. One of the most important partners for FAN1 is a cool looking star-shaped protein called PCNA which clamps onto the DNA strand and helps other proteins, like FAN1, climb on too.

Next, the scientists showed that MutS Beta can push FAN1 off the DNA extrusions and stop it from working properly. Interestingly, the team found that the precise balance of MutS Beta and FAN1 was very important as to which molecular machine got to work on the extrusions. If there is more FAN1 than MutS Beta, the FAN1 wins and can get to work to start repairing damage on the DNA.

## But what does this mean for HD research?

While understanding the precise minutia of how these molecular machines work may seem a million miles away from finding a cure for HD, the impact of this type of science can be very important for drug discovery.

The identification of genetic modifiers of HD gives scientists some of the best clues for how to make new medicines. These gene lists provide crucial insight about which proteins could be switched on or off, in the hope of delaying HD symptoms.



*PCNA is a star-shaped protein which helps FAN1 repair DNA*

It's because of thousands of HD patients and their families that donated DNA to research efforts that scientists were able to discover that both FAN1 and MutS Beta can influence the age of onset of HD. This new paper by Pluciennek and colleagues shines a light on some of the cool details of two of these modifiers, and the delicate balancing act between FAN1 and MutS Beta during repair of CAG extrusions.

Studies like this will in turn help drug hunters focused on these pathways to conduct better experiments as they attempt to refine and develop new drugs for HD.

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*The authors have no conflicts of interest to declare*[For more information about our disclosure policy see our FAQ...](#)

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### GLOSSARY

**somatic** relating to the body

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HUNTINGTON'S  
DISEASE ASSOCIATION  
NORTHERN IRELAND



*Families at the heart of all we do*

# FUNDED THERAPY PROGRAMME

**A time to destress ~ A time for you**

A FULLY FUNDED THERAPY  
PROGRAMME OF UP TO SIX  
SESSIONS FOR SERVICE USERS  
WITH EVIDENCED NEED

Counselling ~ Massage ~ Aromatherapy ~ Reflexology ~ Hand Reflexology ~ Ear  
Reflexology ~ Indian Head Massage ~ Reiki ~ Music Therapy ~ Art Therapy

## Funded Counselling and Therapeutic Service

We launched our Funded Counselling and Therapeutic Service in September 2020. It was impacted by COVID in that face to face was not possible a lot of this time so since things have eased with the end of the pandemic we have helped over 40 Service Users access services to help them with a variety of needs. This programme has been made possible through funding that has been secured, for which we are very thankful.

This service offers service users the opportunity to avail of six sessions of counselling or holistic therapies which they would like. Counselling can be delivered Face to Face, over the phone or via video sessions according to the service user's location, preference, mobility or availability. We have even been delivering Art Therapy to a service user in their home.

Our Holistic Therapy has again been more accessible to service users as restrictions have ended and we have been able to provide therapies such as massage and reflexology for those who would like a bit of relaxation and ease for their symptoms and to help with emotional stress. Again this has been offered in the service users' homes or at the therapist's treatment rooms (again this can be dependent on the service users' location).

This programme is designed as a time to destress, time out and time for themselves. Those with evidenced need can be referred into this by their Family Support Worker and we will do our very best to get a service according to their needs.

\*We are continually developing our pool of therapists. Should we not have a therapist in your area we will attempt to source someone for you or offer you an alternative.



## In loving memory



Very sadly some people lost their fight against this dreadful disease and we would like to take a moment to remember them and think of their family, friends and loved ones.

### **Dessie Clark**

Clarke, August 31st, 2022, (peacefully) at Greenpark Nursing Home, William Desmond (Dessie), late of Fellows Hall Road, Killylea, much loved father of Ryan and Ashley and a dear brother of Noelle and the late Alistair.

### **Rosetta Donnelly**

Donnelly Rosetta, Mountfield, Co Tyrone, (nee Farry) formerly of 9 Forest View, Mountfield BT79 7XS, died peacefully in Hillcrest Nursing Home, Omagh, on Tuesday 13th June 2023 surrounded by her loving family. Treasured mother of Danielle Allen (Mark) and Megan (Danny). Loving and devoted grandmother of Tadhg, Lana, Fiadh and Jasper. Cherished sister of Pat-Joe, James Henry, Marie Devane (Richard), Dessie (Greta), Dympna, Stephen (Bernie), Ursula and the late Catherine McCann (Mickey) and Noreen McCann (late Sean).

### **Charlie Keenan**

Peacefully at home 11th May in the loving care of his family.

Dearly loved husband of the late Bridie nee Green

Much loved father of Tracy Corrigan (Kevin), Shane (Ann-Marie), Kera Teague (Fergal), Melissa (Ray), Gary (Orla) and Darren (Aisling)  
Loved and cherished by all grandchildren

Dear brother of Vera Grey, Mena Pugh, Rosie Keenan, Francis Traynor, Gaby, Martin, Malachy and the late Vincent.

### **Paul Baggaley**

Passed away peacefully in hospital on 19/03/2023.

Loving father of Christopher and Curtis

Devoted Grandfather.

Will be sadly missed and lovingly remembered also by Maureen, Amy, Owen, Ellie-May and Amelia-Rose.

### **HDANI - In loving memory.**

HDANI have a space for those who wish to share photos and memories of those they have lost through HD on Facebook, album, "In Loving Memory." You can also include birthdays/anniversary dates you would like us to remember.

We want to ensure everyone is given the opportunity to share their own memories. I understand this can be difficult and don't want to upset anyone. It may not be something you would like to do so publicly and we respect that, please get in touch with any questions.

If you would like to share memories, please get in touch via email [info@hdani.org.uk](mailto:info@hdani.org.uk) or via Facebook

[www.facebook.com/HDANorthernIreland](https://www.facebook.com/HDANorthernIreland) with photos, messages, dates and we can add these to the album but it won't be shared as a public post on the newsfeed.

If you require any support from HDANI please do not hesitate to get in touch.

# Fundraising heroes

Thanks as always to those who take the time and energy to create fundraising events and or take on challenges to raise much needed funds for HDANI. Please see below some of those who have helped us this year. We are always happy to share your event with our followers and love to be able to let everyone know how people have got on so please do let us know if you are having an event / taking on a challenge.

Mr Farry from Trillick, Omagh for his regular donations from his shop collection boxes and thank you to all those who donate Mr Farry's collection boxes! It is much appreciated!

Mosseley Hockey Club Golf Society who held a fundraiser at their club



ReGen Waste Ltd who chose HDANI as the recipient of their Corporate fundraising.



Knit and Natter group who meet at Newtownstewart Library on a Thursday who raised money for HDANI.



Lough Erne Integrated College who raised money through various events and got friends and family to save used stamps for us to exchange for money. This was led by one of their students Abigail who led the fundraising efforts.



North Monaghan District MCC who chose HDANI as their charity for their yearly classic Run



Killyleagh District No 7 who cycled approximately 100 miles in memory of Desmond Clarke for their Coronation Cycle.



Amy and Chris who chose to support HDANI rather than wedding favours and encouraged their friends and family to support a cause close to their hearts.



Arlene Jenkins and her crew who very kindly chose HDANI as their charity for their Belfast Marathon entry



Oliver and family from SPAR Killyleagh who have continued to host collection boxes for their customers to donate their spare change to.



Glen Spence and friends held a spin class fundraiser.



Christopher and Amy Dalglish who are running the Bangor 10K in September 2023 in memory of his father Paul Baggaley

Thank you to all others - some prefer not to be on social media / named directly so:

**our heartfelt thanks to everyone who has participated in events/ arranged events, donated to events in honour of all those who live with HD, it is very much appreciated.**

**There may be no cure yet but**

**There is help**

**There is hope**

**There is HDANI**

**& we need  
HD heroes like you**

**Contact Us:**

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