The Lived Experience: Hirschsprung's Disease with Down syndrome

At Positive about Down syndrome we work hard to ensure parents have the knowledge and access to support that they need. It is estimated that around 10% of babies with Down syndrome are born with Hirschsprung's disease or another Anorectal malformations (ARMs).

Hirschsprung's disease occurs when the nerve cells within a section of the bowel wall fail to develop, impacting on the ability of the bowel to push the poo along – effectively causing a blockage. ARM's can include a range of problems when the opening to the bowel (anus or 'bum hole') fails to form correctly and is either absent or in the wrong place.

These bowel conditions usually require surgical intervention, either soon after birth or at a later stage. Some infants may also require a stoma until a complete repair can be carried out. While all this may sound very daunting and some parents may be concerned about future bowel control, experience has shown that most affected babies can go on to be toilet trained at the same age as their typically developing peers.

Just because some children can have quite complex problems and will require a number of surgical procedures until the bowel is finally 'sorted' so full bowel control could be delayed.

There is a lot of information and support available to help managed any problems and you will see from the following stories how well children with HD and ARM's have done.

Fiona Gamble mum to Walter

We found out that Walter would be born with Down syndrome and a heart condition when I was pregnant. Although a full AVSD repair would be needed (which would mean open heart surgery by the age of 6 months), we were reassured that it was likely to be one surgery. We were told that afterwards he would have a 'normal' life with no physical restrictions. The doctors were confident he could be born in a general hospital as he would not need special care straight after birth.

Walter was born as planned at 38 weeks. However, due to him constantly moving in and out of breach, it was in the end, by Caesarean section. Walter was born with low blood sugar and was taken to be monitored in the special care unit. After 24 hours the doctors started to get concerned that Walter had a distended abdomen and he hadn't passed meconium. He was transferred to our nearest specialist centre immediately. It was difficult going to see him as I was in a wheelchair, trying to walk the short distance to the lift was challenging and I found the heat of the unit overwhelming at times as I was still in recovery myself. I was trying to breastfeed/pump and although Walter had an older brother, the circumstances of his birth and feeding had been completely different. I found the amount of feeding 'advice' that I was given by the special care nurses/ midwives to be challenging. It was within a few days that the possibility of Hirschsprung's disease came up and the hospital staff had started to perform washouts and biopsies.

The diagnosis of Hirschsprung's disease was devastating. All of our concerns regarding our child's future, all of those things that we asked ourselves about Down syndrome, were now all repeated with double the worry! 'How will this affect the rest of his life?' - 'Will it debilitate him?' - 'Will he be able to have 'normal' relationships? - 'Will he be able to live independently?' And suddenly the thoughts became the dreaded 'Will he become incontinent?' It is one thing to worry about developmental delay

and its social impact, but we were now facing the possibility of a child who has 'accidents' when he is a teenager. Many of my family were focussed on Walter's heart condition, but for me, the Hirschsprung's was what kept me up at night.

There was also the complication of dealing with multiple consultants for each health condition. Walter needed a high calorie formula for the heart, but it was harder for him to process in the gut. Certain symptoms could also be blamed on the other conditions and communication between the departments could, at times, be tricky! For example, for the first surgery, even though it was for his bowel, we were placed on a cardiac ward where the nurses were inexperienced with his bowel condition.

It was decided that Walter would have a pull through surgery where they would first do biopsies to see the extent of how the bowel was affected. So we went home with Walter after approximately 3 weeks to do twice daily washouts, until the date for his surgery came about. At 6 weeks, the time came, although there was an issue in the fact that Walter's original biopsies had come back as one positive, one negative and one inconclusive! It was thought that he could have Ultrashort Hirschsprung's and we were told that there could be a possibility that rather than the pull through they might perform an ileostomy instead. So Walter was taken away. Would he return with a pull through or the dreaded stoma? To our disappointment, they decided to give Walter an ileostomy and wait to perform the pull through after Walter's heart surgery (due when he reached 5kg).

For us this was a real setback and we were worried about dealing with the stoma. However, we quickly became accustomed to managing it. One of our biggest challenges was that the bag could get filled with gas quite quickly so we had to make sure that it was emptied frequently otherwise it would pop or pull away from the skin.

Walter was eventually diagnosed with short-segment Hirschsprung's disease. He was however finding it difficult to gain weight. The extra strain from his heart condition meant that he had to work harder. So he had to go to half breast milk, half high fat formula, to then 100% full fat formula. He was also put on diuretics. He was feeding well by bottle but the calories that he was drinking were being consumed by the effort of drinking so he had to go onto a nasogastric tube. Eventually it got to the point where he was choking and the bolus feeds were going slower and slower so about a month before his heart surgery he ended up on a feeding pump,

all due to his heart condition.

Then came the time when he was able to have his AVSD repair – open heart surgery. This was a success and a few months later we managed to wean Walter off the feeding tube. He had pulmonary hypertension for a year and he was on medication for a while, but fortunately, we got the all clear and we were just starting to discuss his pull through surgery when the pandemic hit. So there was a delay of about 6 months, but the surgery happened in November 2020 and we kept the stoma for another few months to give the pull through time to heal.

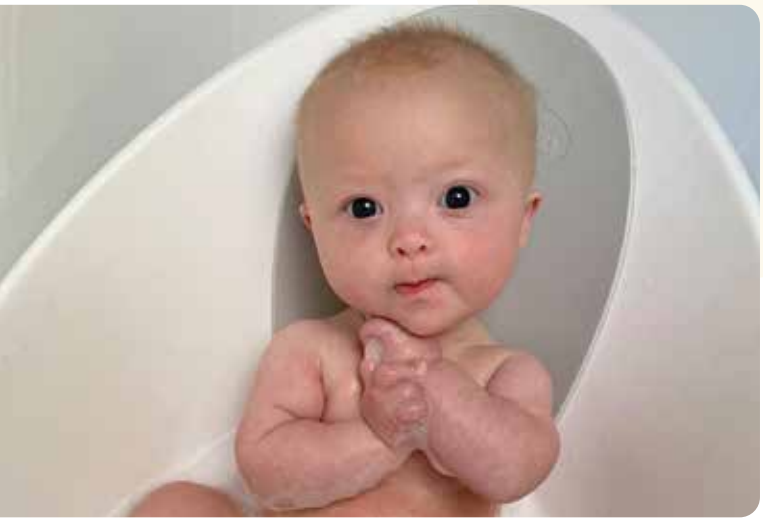
Fortunately, Walter's conditions gave us open access to our local hospital so we could go straight there if there were issues or concerns (his surgeries were performed by GOSH). The stoma surgery was performed using keyhole surgery but the Dhumal procedure (pull through) gave Walter a caesarean type scar. Walter was poorly throughout January 2021 with a blockage in the stoma. He then had a reversal and had a lot of trouble with his first bowel movements and the terrible nappy rash that comes with it! We tried all sorts of creams but he also ended up back in hospital because of an infection at the stoma site. All of the oral antibiotics gave him diarrhoea, exacerbating the condition of his bottom! He was pooing every 5 minutes so nothing was having time to work. We managed to get the antibiotic changed which did help and everything settled down after a few weeks.

At the moment, Walter is still in the bowel 'settling down' period which, I believe, can take 6 months. He is currently having issues with constipation and we are using laxatives and micro-enemas to manage this. It is possible that if Walter still continues to struggle, he may need another surgery, but we are hoping that we will get the right combination for him to settle down and have a comfortable life. He has, after all, only been pooing normally for a few months of his 2 years!

Walter is a real joy. He loves music and playing ball with his brother and we are very lucky to have such a strong loving boy. The pandemic has prolonged his situation and made it more difficult for us all.

Walter should very soon have a relatively 'normal' life but we are reminded by his surgeons that he still lives with the life-long risk of contracting enterocolitis because after all of his surgeries, he still has, and always will have Hirschsprung's disease.

Jenn Casper-Smith mum to Orla



It didn't come as a surprise that Orla had Down syndrome following a high chance NIPT result whilst I was pregnant. However, what did come as a surprise, was when she started showing signs of Hirschsprung's disease just three days after she was born.

Fortunately we were already in NICU as Fetal Cardiology scans had detected heart concerns which the doctors wanted to monitor as soon as she arrived, so I knew that open heart surgery was likely and had googled it to the nth degree! However, bowel related issues had never crossed my mind until after 48 hours when she still hadn't passed the usual meconium poo. I was reassured this wasn't too much of a worry, as due to her heart concerns, Orla was nil by mouth and was just being fed fluids through a drip. Fairly soon after that however, her tiny tummy started to balloon and doctors began using the term 'distended'.

The morning she turned 3 days old, I arrived on NICU, and was told Orla hadn't had a good night and the surgeons would be coming to speak to me soon due to concerns about how distended her bowels had become. She had also vomited in the night, which was very dark green, almost black in colour. That feeling was horrendous, as it was unexpected and I wasn't at all prepared for there to be problems with her bowels. I'd got my head around potential cardiac concerns (which turned out to be completely fine as her heart had actually repaired itself), but since when did I need to be worried about her bowels?!

When the doctor came during the ward rounds, he explained to me that he wanted to do a contrast scan, whereby dye is inserted into the bowels and Orla would then have an x-ray to show where the dye had travelled. The doctor told me that he was expecting to find a blockage in her bowel and he suspected that Orla had Hirschsprung's disease. He explained that this is where the nerves in the bowel haven't formed properly, meaning that the bowel may not contract

as it should to push the poo along, hence the swollen, loopy bowels, which were now visible. Hirschsprung's can affect part of the bowel or all of the bowel; it can cause sections to be permanently closed so a blockage builds up at that point, or become permanently open and unable to contract. The dye would show whether there were any blockages. This was fairly straightforward and didn't take long, plus there were no concerns regarding it being painful so I wasn't too worried about it.

The results of the scan were back the same day and the doctor showed me what was seen: no blockage and the dye moving from where it was inserted in the rectum all around her bowel. This was when a stoma was first mentioned, as the doctor suspected that her whole bowel may be affected. I immediately assumed that this would mean Orla would have a stoma for life, however was reassured that that absolutely would not be definitive because there were numerous surgical procedures which could be performed to overcome the affected bowel. But at that time, she was too small to do this. There was also a need to take biopsies to fully confirm whether it was Hirschsprung's disease.

The doctor felt that we could probably manage the situation by doing washouts three times a day for the next few days, and then reduce this to just once a day and see how her bowels responded. I remember saying 'Washouts? What do you mean? Like colonic irrigation?' I felt totally clueless as to what washouts entailed or how on earth you did that to a baby! As usual, the staff at LGI were amazing and showed us exactly what to do. Despite my concern, Orla did not seem bothered at all by the washouts and it was clear they were needed; there was a lot of meconium coming out of her bowel when we did this and her stomach also deflated shortly afterwards. Unfortunately however, it didn't deflate enough and her bowels remained distended. So, on day five, I was informed she would go in for emergency bowel surgery later that day to create a stoma.

There was never any question in our minds as to whether this was the right

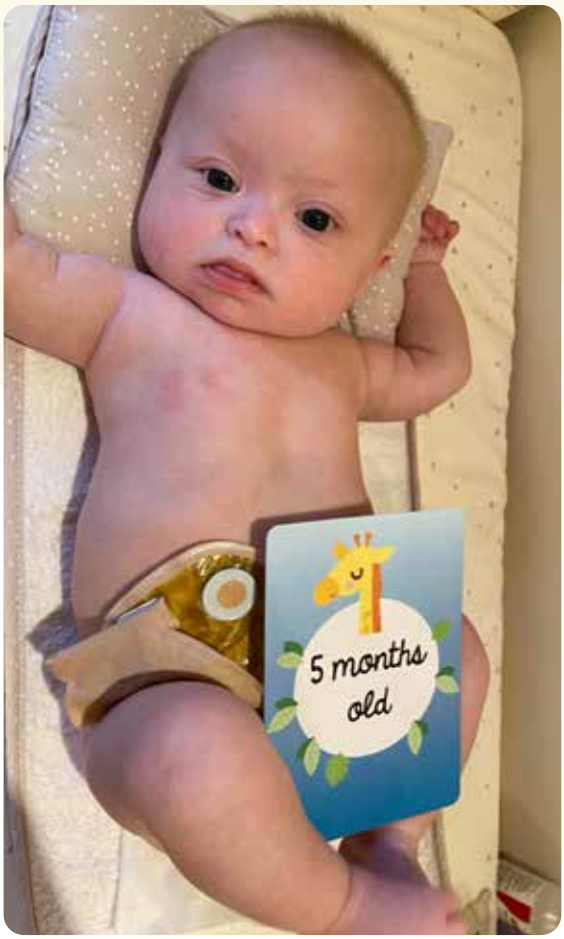
thing or not as both my husband and I had complete faith in the team. Strangely however, despite the procedure it had never occurred to me that Orla would require a general anaesthetic. It seemed so obvious afterwards: of course she would have to be put to sleep for the operation, but that was what unnerved me the most. I feel as though people talk about having to 'be put to sleep' for operations as though it's something which carries a high chance of not waking up. She seemed too small to be having to go through all of this, but within a few hours I was saying goodbye to her at the theatre doors. In all honesty that was probably the worst part in all of this and I felt completely bereft as I left her there.

The nurses advised us to keep ourselves busy during this time and that definitely helped as it was just over five hours later when our little girl came back to NICU. I don't really know how bad I thought it'd be when I saw her post-op, but it wasn't that bad. I feared she'd be visibly in pain; she wasn't, because the nurses had already sorted out her pain relief. I worried she'd have a big ugly stoma and lots of stitches all across her little tummy; she didn't. The stoma was neat and small with an even smaller mucus fistula alongside it, and yes there were stitches which made it look more unsightly and a little caesarean-section style cut where they had opened her stomach up. But it was all so tidy and looked healthier than I expected. Orla now had an ileostomy as the stoma was created from her small bowel.

Whilst operating, her surgeon had taken some biopsies from her bowel but these hadn't confirmed any areas of Hirschsprung's. This caused me worry; it felt like we could manage it if we knew what it was. Orla then had to have a rectal biopsy a few days later which took less than 20 minutes and performed under just a local anaesthetic. This confirmed that she had Hirschsprung's disease in her rectum, so she can have something called a Duhamel procedure where the healthy bowel above is pulled through and attached at the other side of the rectum to enable it to function as normal. Different hospitals seem to prefer different techniques, but pulling through the healthy bowel or bypassing the affected part is an operation which is done on a daily basis across the UK. Surgery on your baby is a huge thing for us as parents, but for the surgeon who is actually carrying out the procedure, it really isn't. These people are skilled perfectionists and that is evident whenever you speak to them.

Orla is now 5 months old and we are still waiting for this procedure due to Covid, however life with an ileostomy is really not as worrying or as stressful

as I initially thought it'd be. We were trained how to change the bag before we left the hospital, and shown how to empty it using a syringe. Due to the risk of enterocolitis (an infection in the bowel which can be dangerous if not detected), it's essential to monitor how much comes out of the stoma so you have to measure the output. This is something which soon becomes the norm despite it seeming fiddly and, in all honesty, a bit disgusting at first! I can honestly say that when I found out my baby had Down syndrome I never thought I would be syringing poo out of a stoma bag!! However, as anybody reading this will soon become aware, you genuinely would not change a thing. Orla is a happy, content baby who at present is none the wiser about Hirschsprung's disease.



All of the doctors and surgeons we have encountered have been amazing so I'm confident about the future. There are so many scary illnesses and debilitating things which can happen to any child, but I assure you that Hirschsprung's disease is not one of them.

Jess Dick mum to Lenny

When Lenny was born in June 2019, he had various health issues and was taken straight to NICU after birth. They very quickly noticed that he hadn't passed any poo and he got transferred from our local hospital to Manchester Children's Hospital at less than 24 hours old.

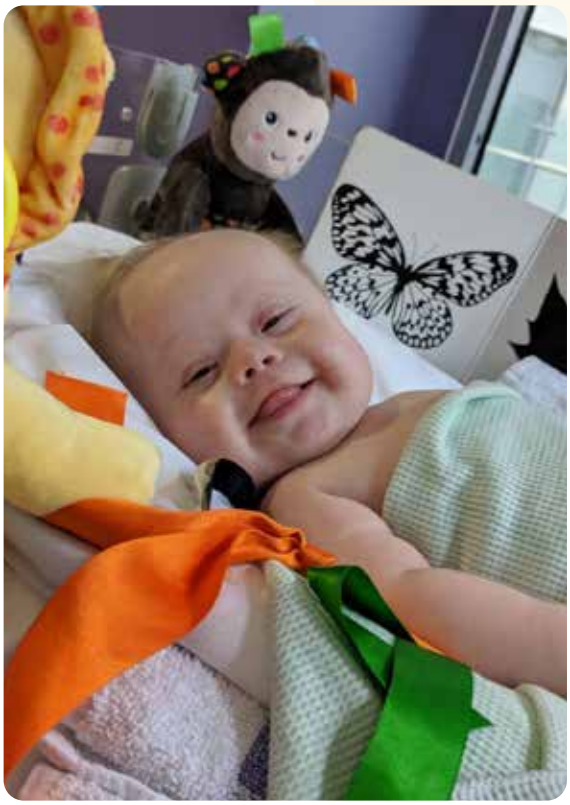
His tummy was distended and a surgeon came to perform a washout which got rid of a backlog of stools. They suspected Hirschsprung's but needed to carry out a biopsy in order to be sure. For a few days Lenny was nil by mouth and they carried on doing the washouts a few times a day.

The biopsy was really straightforward and we got the results back quite quickly. It confirmed he had Hirschsprung's as the ganglion cells were missing in the entrance to his rectum where they did the biopsy, but they wouldn't know how far up the cells were missing until they did the operation. An X-ray showed a tightening in Lenny's large intestines and they assumed this would be where the ganglion cells started.

When we were in the hospital, we were taught how to carry out the washouts on Lenny and we did them ourselves until he was discharged in order to get used to doing it. The washouts can be a bit fiddly - getting used to tubes and water etc. It's possible to do it on your own but we always did it with 2 people - one person holding his legs and the other person actually doing the washout. It's a strange experience and takes a bit of getting used to but I could tell Lenny felt relieved once we'd done it and it was so much better doing it ourselves rather than having to either stay in hospital or have community nurses come out to do it several times a day. It intrigued our 2.5 year old daughter so much that she did some washouts on her teddies!

Lenny was 4 months old when he went in for his pull through surgery, the Duhamel procedure. I was terrified before the operation, worrying about my little baby having such major surgery, but I knew it had to be done and that it would

hopefully improve Lenny's quality of life. The washouts we had been doing were always successful and he didn't have any complications such as enterocolitis so he didn't need to have a colostomy.



The surgery took about 7 hours and was successful. They found that he did have short-segment Hirschsprung's disease and removed a small section of his large intestine.

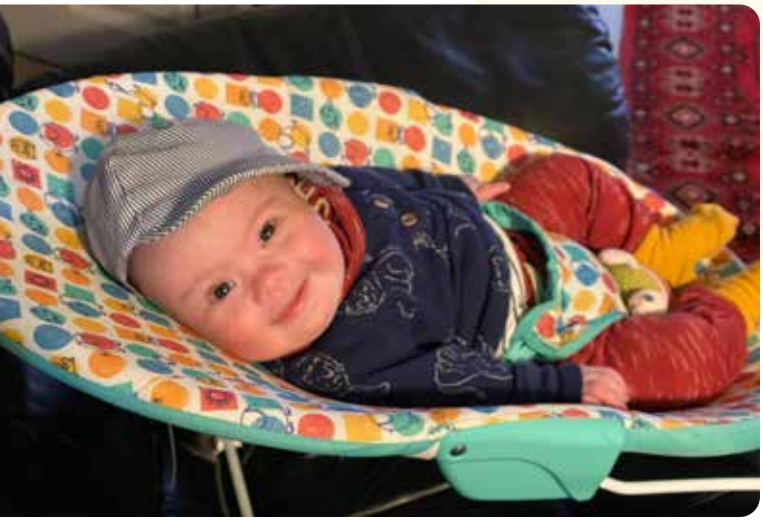
Unfortunately one of Lenny's lungs collapsed during the surgery so he had to spend a few days in intensive care and then was transferred to a ward to further recover, so we were in hospital for about 10 days in total.

He had to stay nil by mouth until he was out of intensive care and he did his first poo about 24 hours after he was allowed milk again. I'll never forget being so excited seeing my baby poo, something most people never even think about!! We went back into hospital a few weeks later for the surgeon to do a quick check under anesthetic to see that all was ok, which it was.

Since then it's been really successful. Lenny has continued to be able to poo on his own with 3ml of senokot in the morning to help him and we haven't had any further complications and no longer do any washouts. Lenny is nearly 2 now and isn't quite eating full meals as he still relies on a fair bit of milk. But as his diet has broadened, he has still been able to pass solids himself.

He has checks with the surgeon every few months and I'm hopeful that he won't need any further interventions. He isn't anywhere near potty training yet so that's the next hurdle in a few years time I think!

Jessica Savage mum to Albie



We were desperate for our baby to have a poo! Funny to think how a simple act was such a big deal...

Albie's journey was a rocky start... a beautiful planned home birth in the early hours of monday 23rd November 2020, the hospital to checkup for any chromosome markers for Down syndrome, to then be whisked away to be stabilised. From collapsed lungs, suspected heart issues, sepsis, blood pressure troubles and then suspected Hirschsprung's kept being mentioned. Down Syndrome barely crossed our thoughts, we just wanted our baby to live.

Google became our best friend because we had never heard of Hirschsprung's disease. The staff in NICU were concentrating on stabilising Albie so he could be transferred for exploratory surgery. He was too poorly for a dye test to look for bowel obstructions, and his X-rays looked like a mass of knotted intestine.

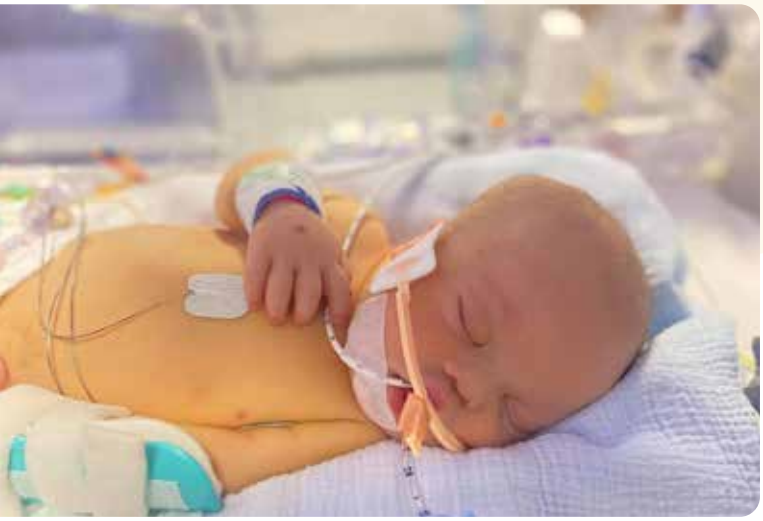
At around 3 days he started to poo! We were so excited, surely this meant it was ok? Sadly not. It was also here when we found out he was diagnosed with Down Syndrome, after a low chance screening.

At 7 days old, still intubated and wired I was finally able to hold Lenny once again. His greeting to me was green sick everywhere! We knew it wasn't right, so it was then that we went directly, via transfer, to Noah's Ark Children's Hospital in Cardiff, arriving around 11pm. Seeing the tiniest baby on an adult stretcher and not being able to go with him was so hard, but we were on autopilot.

It was scary! We hadn't had a chance to register Albie yet, so as his mum, I was the only one able to sign consent to surgery. I literally felt like I was signing life or death for my baby.

At 1.30am he had exploratory surgery with a suspected perforated bowel. When he came back out, he sported a stoma and a mucus fistula. Another new thing to learn, but even mid covid and

lockdown #3, we were allowed to stay together under special circumstances, due to him being so poorly. Meaning



our 3 year old was at home with grandparents.

We were situated in PICU for post surgery. It took them three days and a few 'mum tantrums' to get a much needed meeting and update with consultants. They told us that Albie had developed a kidney injury and they didn't have high hopes, even as far as his brain function, as he wasn't passing urine.

Our worlds shattered. None of this was even related to having Down syndrome. Of course, our little man being him, started to wee that afternoon! It was like riding an emotional wave, especially with a new ward and having different covid restrictions again, which was proving difficult.

For the duration of his stay in PICU, we were fortunate to have a room in The Ronald McDonald Charity House, which is an amazing space available to families with sick children.

Once Albie was strong enough, after about a week and half later, we were due to be transferred back to the original hospital. This isn't something I wanted to happen but because of shortage of space and the other hospital was in our catchment, it is just how it worked out.

So our learning of everything, including stoma bags and care, consisted of a 20 minute low-down and then we were off! There was a stoma nurse (mon-fri 9-5) who did end up showing me the ropes eventually but until then (5 days later) it was carnage!!

Due to being in a different hospital, the staff had to be on constant questioning to the surgical team at the previous hospital for ongoing problems due to the wrong handling of the stoma ...at one point I had 2 registrars, 2 nurses and a trainee watching me change the stoma bag - needless to say they were NOT prepared and I of course was 'Albie's Expert' *5 days into changing a stoma bag / 2 weeks of knowing what a stoma was!*

We stayed in the hospital room which was very much like a prison with no tv and we were not allowed out due to

covid restrictions. It was very lonely. The communication felt non existent due to the mistakes that kept happening.

Albie's fluid losses were always high, but when a consultant talked about a pathway to being discharged, it was me questioning his losses, not them. It was only here that they realised they had gone an entire week without replacement IV fluids! I had already been noting everything down, but from this onwards, I was like a woman on a mission. I would leave the door open and have to often correct or help calculate the day's losses!

At 4.5 weeks old, and with a huge progression of lowered stoma losses, we were allowed 'special leave' to go home Christmas day and Boxing day. I had made sure to do every single bit of care that was possible for us to do including the NG tube training, the stoma care, which was what I was doing anyway, to make sure the only possible reason to be in hospital was to do blood tests, blood pressure and obvious medical things I couldn't do at home.

We had one trip back to the hospital which was due to Albie's high stoma losses... we were very anal at keeping a measurement diary of his losses at home. We learnt the calculations to measure loss allowance, we even got to the point where he did not need replacement fluids by IV and we were able to do this at home via feeding.

Albie's medications were all done via NG tube until we slowly weaned him into having it orally, he really was a superstar! He had Loperamide as medication because his allowance is always higher and of course dehydration is a big no in terms of stoma care patients.

This is where we were, home for the foreseeable until surgery. The stoma just became part of our life, and in one way, lockdown made it easier as we were not able to go out and about during this time anyway.

Just as Albie turned 3 months, he had his pull through surgery. His surgeon, Mr Oliver Jackson, has been the most knowledgeable and helpful person through this experience. His attention to detail and care for his patients is beyond anything, and we really feel this shows in Albie and how he is.

Mr Jackson explained that he had a full team for the surgery. He explained it in a huge amount of detail, and of course we were armed with more questions. He had biopsies along the way to measure in full details where the Hirschsprung's diseased area was and whilst in surgery found that he didn't have long-segment Hirschsprung's Disease, but just past short-segment (we call this the Albie Segment, raising the bar!) Mr Jackson also used a brand new instrument for

the first time in his career with our permission. It was a smaller version designed specifically for children with HD.

Albie was in for a 9 hour surgery. Plan A was to leave the stoma, so his bowel would recover and then reverse it at a later date. Albie was a rockstar so Plan B happened and he came back with no more stoma!

We planned for the worst but hoped for the best. He had a bed waiting in HDU, and it was needed. He had an x-ray and some drugs to help his airways, but I genuinely expected him to be in PICU with his past issues and his sensitivities, but he was incredible...the happiest little boy.

His rash was unreal when he did start to poo, but I had my hospital head on and when creams were being washed away I was requesting more. I had a cocktail of creams on the go. I had read up on how severe it could be, and I was armed with how to prevent it. We found our favourite cocktail and his rash cleared up in a few weeks.

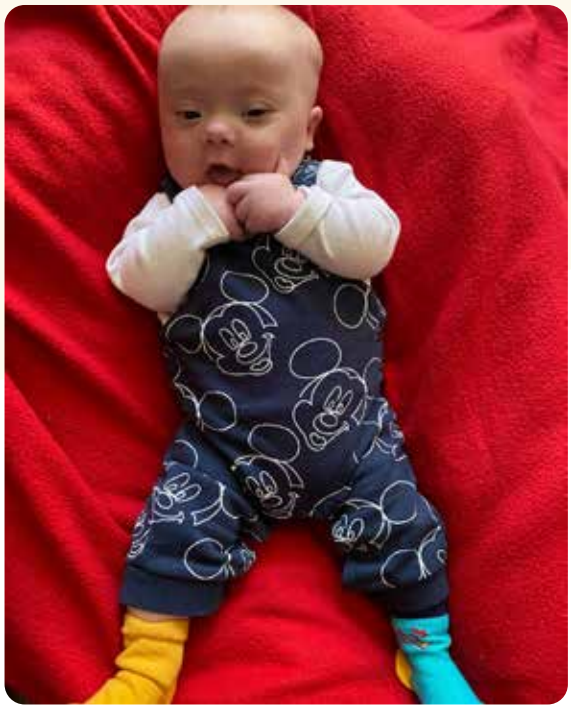
Albie is now nearly 6 months old. We have just started the weaning process which is interesting with how nutrition can affect the bowel, so we are just adding to our (daily rolling) diary and enjoying every second of our baby. He has been through more than many and he is the most content, happy baby I have ever known. We are so grateful for him, and we are very proud of our HD Warrior

Things I wish I had known:

- All hospital wards are different, even if they are within the same hospital. Rules and protocols change.
- Constant nagging and asking questions got Albie the best care possible, because without it we wouldn't have had an input into his care.
- Syringe and kwill to empty stoma bags and measure at the same time.
- Put the stoma bag and adhesive ring in your pocket or bra to warm up - it sticks better.
- Only use warm water and no perfumes on the stoma area.
- Make sure it's completely dry, stoma powder, adhesive ring, bag.
- Keep the plastic backing for a template for your next stoma bag - don't do too many at a time as it changes all the time though.
- Squash down the adhesive ring so its super thin around the stoma so it doesn't raise the bag.
- Orabase paste - which is for stomas - is an excellent barrier for nappy rash - metanium lathered over the top - only

remove cream on top layer and leave under layers - its looks yuck but as long as the dirty area is off and clean, it prevents the rubbing.

- Write everything down, even if you think it is silly.
- If you don't get answers from doctors, ask them the question in a different way.
- You know your baby best, stand your ground!
- Know that you are amazing and so is your baby!



Kelly Porcas mum to Olly

Olly side-swiped us shortly after he was born with a diagnosis of Hirschsprung's disease alongside his other health issues. Hirschsprung's disease (HD) is a bowel condition where there are no nerves to part of the bowel, so passing stool as usual, is at best difficult, at worst impossible. Olly had been unable to pass meconium and quite quickly went downhill, which led to him being transferred from our local hospital to Birmingham Children's Hospital at 48 hours old. Unbeknownst to us, Olly was a very poorly boy and at 4 days old had emergency lifesaving surgery to remove a large section of his bowel and form a colostomy. After further bowel surgery and over a year later, his bowel finally stabilised. However, as a result of this and other health issues, we still have the colostomy in place 5 years later. This is a decision taken both by us and his bowel consultant. He is now a picture of health and developing beautifully, but it was a long road to get his health to this stage.

We'd been told all along not to expect much as far as toilet training was concerned as the lack of nerves and the pounding his digestive system had taken may well mean he would struggle with control. Alongside this, Olly has very delayed in speech development, so we held off on full potty training, believing we needed to wait until he was ready

From around 2.5 years old we had been consistently putting Olly on the potty at bath time, as well as having an open door policy when we went to the toilet. Around this time, I had signed up to attend potty-training training with our local support group. Unfortunately I was ill on the day of the training and could not attend, so as often happens, life got in the way and full potty-training kind of got forgotten about.

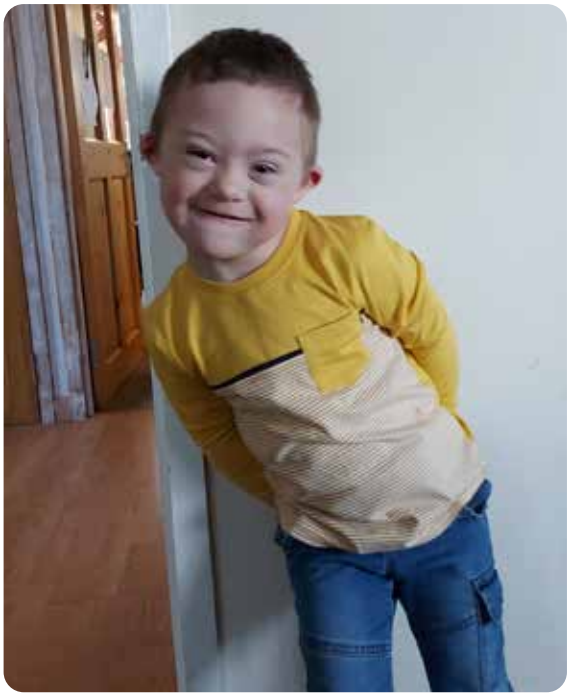
We started to notice that Olly was performing whenever we put him on the potty and would often be dry before we put him on. This pushed us to start thinking about full training again, which

we decided to try in the summer of 2020 when the weather was better. Then Covid-19 hit and I was furloughed at Easter. This seemed like the perfect time to get started. We joined PADS first potty training boot camp, did our baseline testing and couldn't find that much of a pattern to his weeing, but just went for it.

The first 3 weeks he went completely commando. After a few major accidents, Olly seemed to realise he didn't like the sensation of being wet. One incident in particular where he sprayed his bedroom like a jet washer, was particularly memorable! He started to take himself to the potty, which we always had in the corner of whatever room we were in. We then moved onto just pants on his bottom half, and moved to part toilet, part potty. It became obvious pretty quickly that Olly preferred the toilet to the potty, so we quickly dropped the potty. Throughout we were working on timed toileting as Olly still would not initiate letting us know when he needed to go. This continued for several months and well into when we eventually went back to having him fully dressed. Eventually it clicked... we now have a verbal cue from him, or a wee dance which tells us he needs to go but he still stubbornly refuses to sign for the toilet, despite knowing the sign!

Olly moved to pre-school that September and went from day 1 in pants and pre-school have completely supported our toilet training programme with him. He still has occasional accidents, especially when engrossed in something, but these are becoming much more infrequent.

All of Olly's colostomy care now takes place in the toilet, including colostomy bag empties and changes. This is much more appropriate and dignified for him as he no longer feels like a baby being laid down at a baby change station. It's also much more convenient for us, not having to find somewhere we can lie him down for a change or empty. It also means we can use 'normal' toilet cubicles when out and about, rather than me anxiously checking out the location of the baby change or accessible toilet everywhere we go! His additional

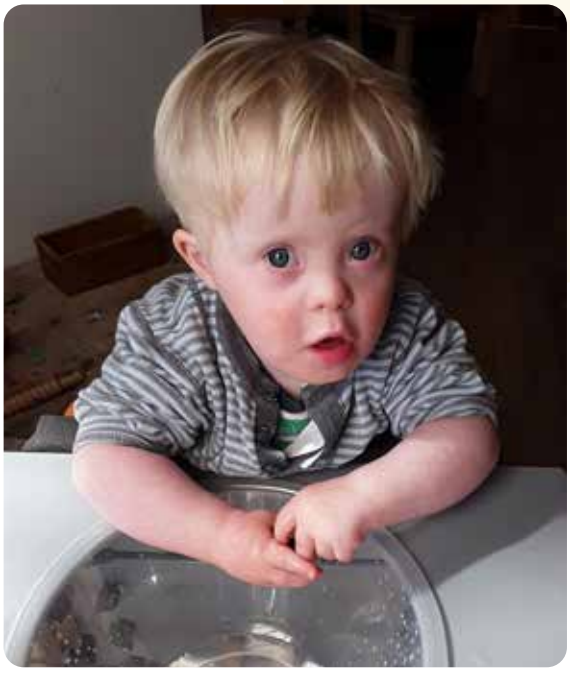


toileting needs are far less obvious to his peers at pre-school as they can be carried out in the same toilet cubicle as everyone else uses.

Olly's bowel consultant is significantly impressed with the progress he has made and the control that he is showing, and as a result has said it might be more likely in the future that a colostomy reversal might be possible. Toilet training has also improved the mucus stools that Olly passes every 6-8 weeks (this is like a smeary poo, as is his body clearing out the dead cells in the bowel and rectum). Prior to toilet training, passing these would often be a day or night long process that was painful and very distressing for him (and us). Since toilet training this seems to have become much easier for him to pass, and although he still becomes a little distressed, it certainly isn't the hours of upset we had from very early on. He now does these on the toilet, letting us know he needs to go via his usual cues.

For anyone questioning when to start potty training their little one with a bowel condition or similar, I would honestly say go for it! Follow the steps set out in the plan on the PADS site and just try it. The professionals team around us are great and highly supportive, but no-one really seemed to think we'd achieve having Olly well on the road to dry by the time he was 4. Olly starts school in September, and although we'll send plenty of spare clothes, this is less to do with accidents and more to do with his obsession with water (and consequently soaking himself and any bathroom)!

Kirsten Jack mum to Elliott



Our son Elliott was born 9 days overdue after a fairly uneventful pregnancy. Shortly after he was born he was transferred to the neonatal unit as he needed oxygen. A short while later the consultant came back to the ward to tell us he suspected Elliott had Down syndrome which came as a bit of a shock. We had screening during pregnancy but our chances of having a baby with Down syndrome were about 1 in 17,000!

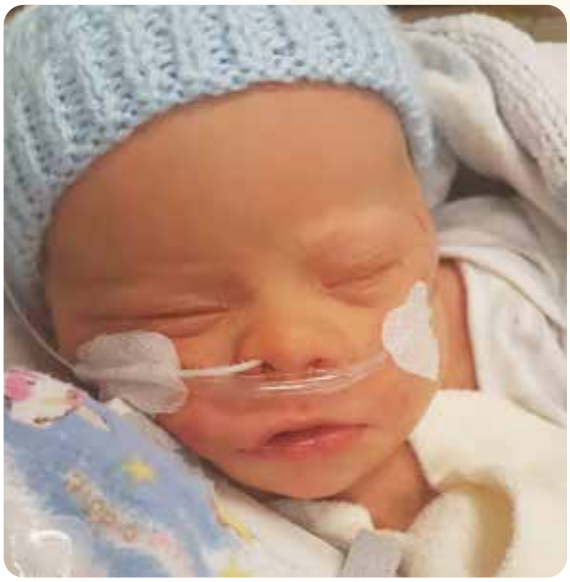
The next morning the consultant reviewed Elliott with a big beaming smile, he was so pleased with how Elliott was doing. Elliott was breastfeeding well and the consultant was confident his oxygen could be weaned down and he would be back on the postnatal ward that afternoon. Elliott however, had other ideas!

That afternoon Elliott's feeding tailed off and by evening he became unwell with a distended abdomen and bilious vomiting. He needed to be transferred to the RVI in Newcastle for a surgical opinion as the doctors suspected he had Hirschsprung's disease. Thankfully, at 24 hours post C-section, I was allowed to be discharged from the postnatal ward and could go with him.

The next few days are a bit of a blur. We had some doctors telling us it was unlikely that Elliott had Hirschsprung's disease, whilst the consultant surgeon was sure he did have it. It was a frustrating time watching Elliott make little progress with feeding yet not getting any answers. Everytime we introduced feeds he would become distended and need a bowel washout to help him poo. Eventually at a week old, the surgeons were able to take a biopsy to confirm whether or not he had Hirschsprungs. The biopsy itself was very straightforward, they were able to do it on the ward and Elliott didn't even flinch.

We were told it would take 48 hours for

the results. By this point, I was almost hopeful it would be Hirschsprungs, as something was clearly not right and



even though it meant he would need surgery, at least we would have a plan. I was quite shocked when an anaesthetist came to see us the next morning to prepare for Elliott going into theatre that afternoon.

That afternoon the surgeon came to say the biopsy confirmed a diagnosis of Hirschsprung's, this was only 24 hours after the biopsy had been taken. We didn't have much time to get our heads round the diagnosis or the fact Elliott was going to need surgery. The next thing I remember is signing the consent form and Elliott was whisked off for surgery. At that point, Elliott was only 8 days old.

The hardest part of Elliott's surgery was not knowing how much of his bowel was going to be affected so it was a very nerve wracking wait. I have some very surreal memories of trying to distract ourselves with shopping in Primark and Boots. Luckily the section of bowel affected was quite short so he was only in theatre for 4 hours and we were able to see him shortly afterwards.

In terms of recovery Elliott did brilliantly. He spent a night on PICU where he needed feeding via an NG tube but by the next morning he was managing full breastfeeds. He required no pain relief and was discharged three days later at 11 days old! Elliott initially lost weight when he came home but we persevered with breastfeeding and by three weeks old he was already back up to birth weight. You would never have known he'd had such major surgery to look at him.

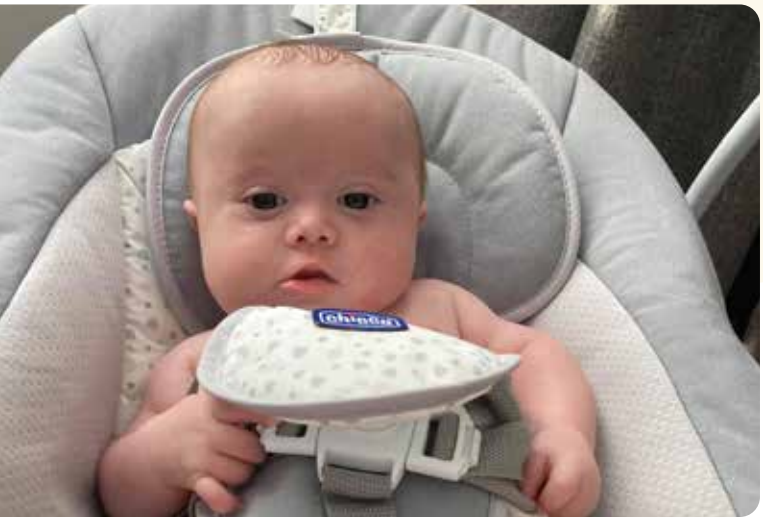
Whilst it was great that Elliott was able to have his pull through surgery so quickly and we didn't have to negotiate a stoma or bowel washouts at home, it wasn't without its challenges. We didn't have any time to prepare ourselves for what the recovery would really be like for Elliott. He could poo almost constantly day and night and even 6 months after surgery we would get through about 500 nappies a month! This also resulted in horrendous nappy rash. We were never told this was a common side effect of the

surgery, so we spent time using creams that were not very effective.

My advice would be to make sure you get some effective barrier cream on immediately after surgery. We found Ilex the most useful but different creams work for different people. I think part of Elliott's issue was that we weren't given anything to use in the hospital straight away, so his fragile newborn skin was damaged very quickly and took almost 9 months to completely recover. We also use reusable baby wipes as they are much gentler than wet wipes and we have certainly got our money's worth! Whilst it sounds like a very minor side effect, it did cause a lot of stress and anxiety, as it felt like it would never heal. Everytime we had a long car journey or a day out where I forgot his creams, we would be back to square one.

Elliott is now 2 years old and doing well. He has never developed any major complications as a result of Hirschsprung's disease. He still has very loose stools and poos frequently throughout the day. My biggest anxiety relating to Hirschsprung's is probably about managing toilet training as it's hard to know what the future holds with that. We are currently trying medication to make his bowels more manageable. Luckily there is a lot of support available through the PADS potty training group and it's lovely to see some success stories of children with Hirschsprung's being potty trained.

Leanne Constable mum to Parker



I knew Parker would have Down syndrome before he was born and due to my placenta failing, he was to be born at 36 weeks. I prepared myself that there could be some complications and Parker would need to go to special care. However, Parker was born, and he was doing brilliantly. I couldn't believe it!

We were on the maternity ward, and he was feeding lovely for the first 8 hours. Throughout the night he became more and more difficult to feed and the midwives kept saying "oh we will feed him" as if I was doing it wrong. Parker being my 4th child, I knew how to feed a baby. I kept telling the midwifes that he had not done is first poo, but they were not concerned.

By the next day I was becoming increasingly worried as he didn't wake, he didn't want to feed, he didn't cry, and he still hadn't done a poo. Again, nobody listened to my concerns. Luckily, late on the second night, the consultant came to complete Parker's heart scan. I told her that he had not passed the meconium. You could see she was concerned straight away. She completed her examination and said she suspected that he had Hirschsprung's... that was soon to be a name I would never forget!

Parker was transferred to NICU straight away and I was told he would be transferred to Cardiff the following day. Parker was put on nil by mouth and the nurses began bowel washouts. Parker was transferred to Cardiff NICU the next day. Watching him being taken in the ambulance was probably the most heart broken and scared I have ever been in my life.

Once we arrived at Cardiff it was an agonising few days before they could complete the biopsy which was required to confirm if it was the dreaded Hirschsprung's. The day finally came that the surgeon was happy to complete the biopsy. I was petrified of what my baby had to go through. In all honesty the surgeon and his team were amazing, and

it was completed very quickly. Parker as usual blew me out of the water with how tough he was. It took around 4 days to get the results back which confirmed that Parker had Hirschsprung's disease. He was missing the little ganglion nerve endings in part of his bowel. Getting the news was a shock as I had convinced myself that it was something much less complicated and all would be fine! It was my coping mechanism I suppose.

The surgeon at Cardiff Hospital was amazing and he explained everything in fine detail and ensured I understood the next steps. He didn't sugar coat anything and gave us the worst and the best possible outcomes. The plan was that I would be trained up to complete the bowel washouts at home and once Parker was a bit bigger and stronger, he would have Soave pull through surgery.

The nurses were great and before I knew it, we were at home, and I was a fully-fledged 'Bowel washer-outer' as I called myself. The first few were truly daunting. I gave myself a stress headache every time due to concentrating so hard, however I soon became confident, and it was like second nature. Family and friends were in amazement but when it comes to your children, us Mam's and Dad's will do anything.

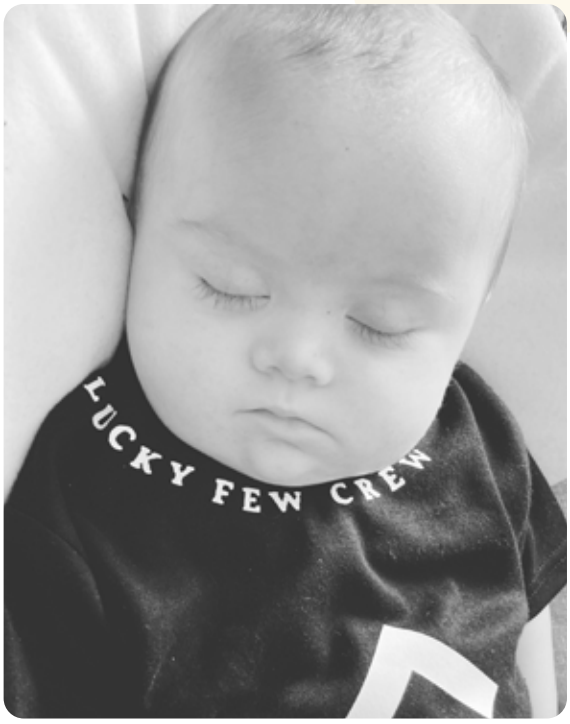
It was planned that Parker would have the surgery at 12 weeks old and the day had arrived. We were admitted the night before the operation so they could carry out bloods etc. I was petrified and I didn't sleep the night before the operation. I just wished I could take his place as I didn't want my tiny baby to have to go through this even though I knew it had to be done.

The morning of the operation, the surgeon came to speak with me and explained how the day would play out, and he really put me at ease and filled me with confidence that everything would be ok, and I thank him so much for that. The nurses came and we walked Parker to the theatre. The nurses did tell me not to be with him when he went to sleep as it can be traumatic to watch but I had to be there with him as much as I could.

He went to sleep, and I had to leave. That day was the longest 7 hours of my life. The hours seemed to drag but I got the call from the surgeon that the operation was complete, and that Parker would be back with me shortly. My goodness... I was so happy. Parker came back to the ward, and he looked so sleepy and was horrible seeing him hooked up once again to all the machines. Parker as usual was so amazing and by the next morning he was smiling and more back to his usual happy self.

The doctors came to see Parker that morning and I couldn't believe it when

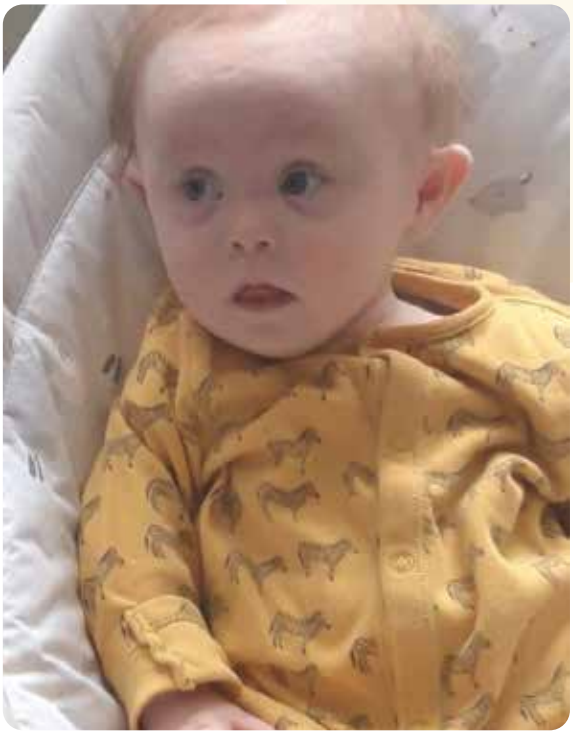
I opened his nappy, and he had done a poop!! I was so excited! Never did I think I would be so happy to see a dirty nappy in my life! The poops came thick and fast as Parker started to feed again and he went from strength to strength. We were discharged from hospital on the 5th day, and we have been home ever since.



Parker is doing so well but the nappy rash can be an issue as we are still waiting for the pooping to slow down and some days, I can change endless amounts of dirty nappies, but I am just so happy my boy can poop!!

We know Hirschsprung's is a lifelong condition, and we may hit some bumps along the road as Parker grows but we know we have a fantastic team behind us, and we are ready!!

Mirrelle Kemp mum to Donnie



Donnie was born at 37 weeks at Queens hospital, but he was transferred to Royal London hospital at two days old as his belly was a lot bigger than usual. The day after they performed a full rectum biopsy. It was really worrying waiting for the results, not knowing what it could be.

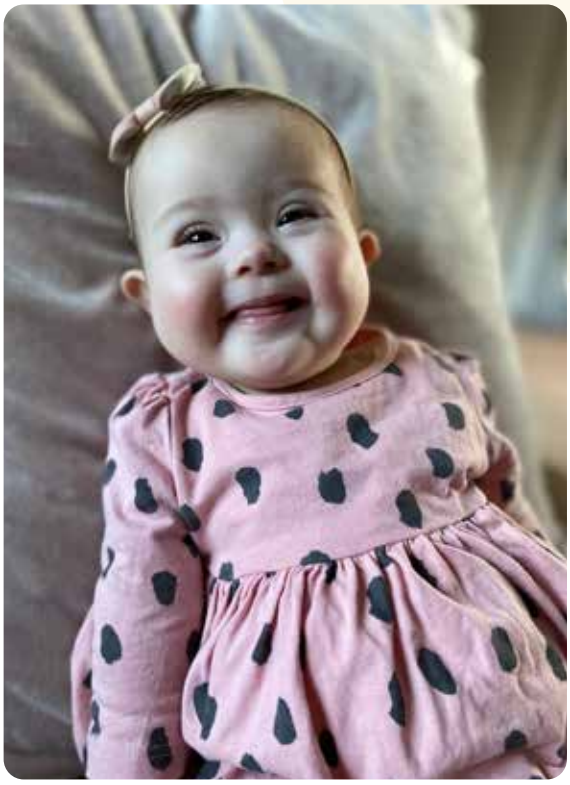
At 7 days old he got diagnosed with Hirschsprung's. The staff taught me how to do daily washouts. It took me about two to three days to be confident of doing them with the nurses help if I needed it.

In time, I became more confident and was able to do them on my own and then Donnie was discharged from hospital. He still had check ups with the surgeon to see how the washouts were coming along.

At 8 weeks old, Donnie returned to hospital to have the pull through surgery. It was scary knowing my baby was having surgery, but I knew he would be ok. The operation was on a Friday morning and he was later transferred to PICU for recovery. On the Sunday, he was transferred to a ward. Then on the Monday, he did his first poo! So on Tuesday he was discharged.

Donnie is now 8 months old and doing amazingly well! He does get constipated now and again, but he's on Movicol which really helps. He still has check ups every 2/3 months and the surgeon is very happy with how well he is doing.

Paula Sordini mum to Luna



We knew something wasn't right when our newborn baby didn't pass meconium in the first 48hs.

While Luna was in NICU at our local hospital, we were transferred to Evelina's Children Hospital to perform a biopsy. They told my husband to wait outside the room as it might be difficult to handle. "Too difficult for parents but babies wouldn't remember a thing". What was harder, was the fact that they had to perform a second one biopsy as the first one didn't have enough samples to confirm a diagnosis.

I was confident everything was going to be ok as she was able to pass stools on her own after the second day. But the results confirmed she had Hirschsprung's disease.

We left the hospital with that information but as she was still able to poop, we didn't think there was anymore to it.

I'm still baffled on how little information we were given and the lack of discussion on what to expect. So I hope now that our experience might help other parents in some way.

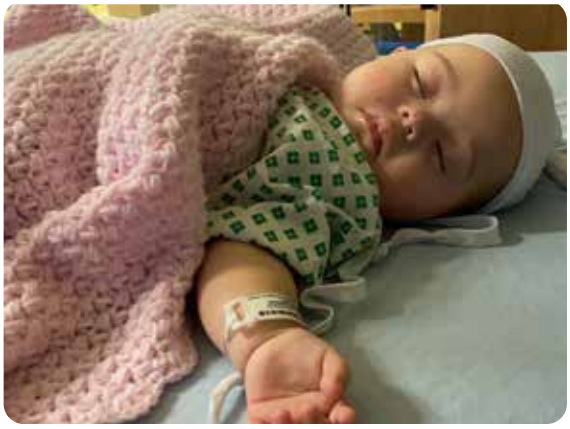
Less than five days after being home, we decided to go to A&E as Luna's tummy was slightly distended and she hadn't pooed in two days. It was then that they explained to us how to do washouts. Which we then started to do on a daily basis.

I remember how daunting the prospect of doing washouts was. How scared I was to harm my beautiful newborn baby. But, you soon learn to get on with things and make them part of your routine. I know this might sound odd, but to me, washouts gave me empowerment. It gave me the feeling of being able to help Luna without needing doctors or medicine. Doing this everyday, I was helping her to not need two operations and prevententing infections.

Every day as soon as she woke up I would warm some saline water and have the set up ready in the bathroom: easiest place to clean in case of accidents.

Changing mat on the floor with inco sheets, ensuring to cover the wall (once again in case of accidents).

My husband was responsible to keep her legs up and massage the tummy to help



movement. He was focused on her face. Talking to her, singing and making sure she was comfortable and having a good experience.

My focus was doing the job, quick but thorough. My routine:

- Insert the little finger first (with lube) for five seconds to stimulate
- Have the catheter ready with saline water and tube every time
- Pointy syringes are better to flush the water than the screw-head onesI would go from top to bottom. Going as far as possible so then it would address any blocks from the start
- If there was less water coming out than what went in, I would go with an empty catheter and move up and down slowly leaving time for the water to be release
- Once the liquid coming out was mainly just saline water, we would finish the session

After six months, Luna's pull-through operation was scheduled. Even though I knew it was necessary, I was still scared. We had two appointments with the surgeon before the procedure but I don't think anything can fully prepare you.

Luna was under general anaesthetic for six hours. She was having a PEG (Percutaneous Endoscopic Gastrostomy - this is when a feeding button/tube is place directly into the stomach (rather than down the nose) in the same operation so that added to the time. The surgeon explained that it was mainly them waiting on the lab results for the biopsies. They took a biopsy of three sections and removed the area without the relevant cells plus an additional six centimetres making it to a total of 20 centimetres in length.

When I went down to recovery to see her straight away. She was slightly awake but not fully, due to the morphine. She seemed to be in some pain but was stable, so we went up to the ward where she was monitored.

Less than 24hs after the operation Luna started to pass stools. It was mainly blood for the first few times which is to be expected.

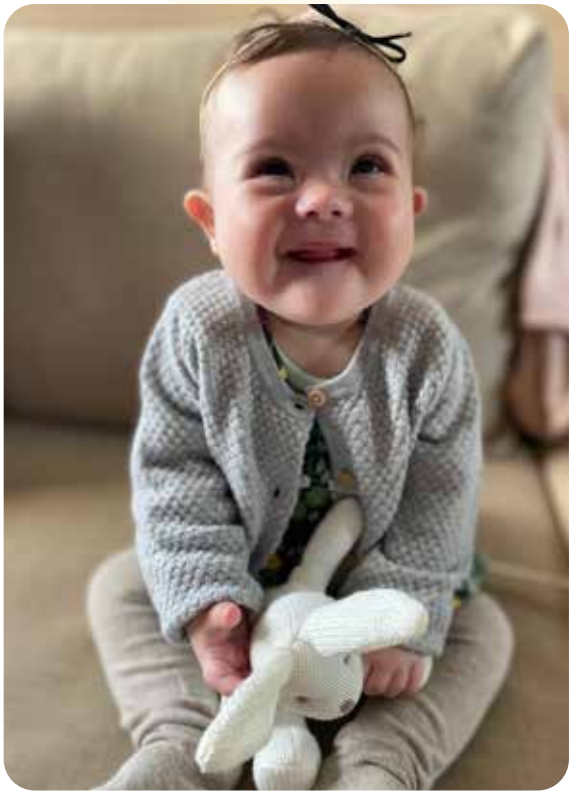
Things I wish I knew/was more prepared:

- It will be painful for the baby at the beginning. I know it's obvious, but I was so focused on the operation that I didn't think of the next stage
- Barrier cream! Have a good one on hand. Baby's bottom is going to need it.
- Nappies! Be ready. I know that like me, you'll be happy to change dirty nappies :)
- They might insert a catheter during the operation: which was later explained that is common practise when given morphine as this drug can make the bladder 'sleepy'
- Baby might develop high temperature. This can be caused by the aesthetic.

Luna ended up developing an infection so we had to stay in hospital for a few days extra, but everything quickly improved with antibiotics. I was surprised at how quickly Luna recovered. Once back home, it looked like it was painful for her when passing stools, to some degree, but eventually things got better.

I think the uncertainty on what was going to be the outcome of the operation was also a source of anxiety once Luna was recovering. Whether she might need more washouts or botox or stimulation.

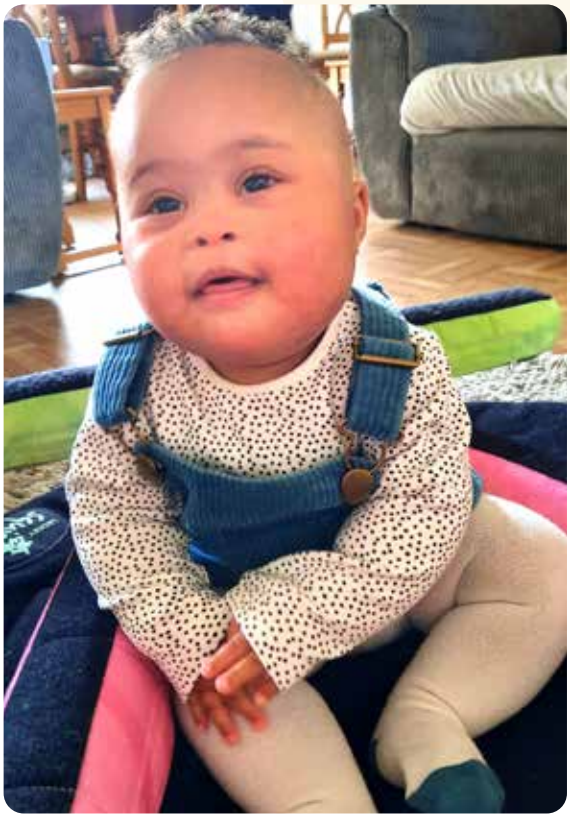
We are now five months after the operation and Luna has been going regularly with a little help from Movicol. Something so simple for many but tricky for our babies. Our strong and brave babies.



I think we went into this with little knowledge so I hope that my experience can be useful to other parents going through this.

From a proud mama who is still happy to see dirty nappies.

Rachel Giegbefumwen mum to Etta



Down syndrome was first mentioned to us just after my 12 week screening tests as my results had come back as a 1:6 or 16% chance of Trisomy 21. We then decided to go for an NIPT test as we didn't want anything invasive and 16% didn't really mean anything to us. The NIPT came back as over 99% chance, so we continued the pregnancy pretty sure we would be blessed with an extra chromosome. We also found out we were having a girl and decided to name her Etta.

All through my pregnancy I was given extra scans to check the baby's heart and progress. At 28 weeks the scan showed a possible tiny hole in her heart, the cord pressure was increasing and the placenta had begun to fail. I was then scanned weekly to check Etta's growth, heart and my placenta. I was told if there was any change in her usual movements I must go straight to hospital. At 34 weeks our consultant decided Etta would be safer out rather than in, so a C-section was planned for two days later at Dorset County Hospital.

The day of the C-section arrived. We were very nervous about how "well" our baby would be, especially as she'd stopped growing over the last few weeks. Etta arrived screaming, tiny at 4lb 1oz but seemingly perfectly healthy. She didn't need help with breathing so she could stay in theatre with us until the end of my operation before heading to SCBU.

She had an ECG amongst other tests and it came back that her heart was absolutely fine. We were so relieved. A blood test confirmed Down Syndrome. She did have low blood sugar, high red blood cell count and jaundice, but that was all to be expected under the circumstances of her arrival.

We went to SCBU that evening to see her, she was just perfect!

The following morning we couldn't wait to go see her again, but as soon as we

got there, we were taken aside by a doctor, who told us Etta had vomited green bile overnight and had not passed the meconium yet. They thought she may have Duodenal Atresia and if that was the case, she would need an emergency operation. They had ordered an x-ray to be done as soon as possible and were in contact with NICU in Southampton. We went in to see Etta. She was having her stomach drained through an NG tube and had intravenous antibiotics and fluids set up. After waiting what seemed like hours, we were told an ambulance was on its way to take Etta to Southampton.

We drove to Southampton while Etta was in the ambulance. When we arrived, we had to wait around a long time while I was admitted to maternity, as I still hadn't been discharged after the C-section. Once that was completed, we could go to see Etta in the NICU. A doctor came and asked a few questions about our family history and then told us that the x-rays ruled out Duodenal Atresia. Again, we were very relieved. However, there were concerns she may have Cystic Fibrosis. A blood test ruled that out the following day. Etta was nil by mouth and was having twice daily bowel washouts while more tests were carried out.

After 3 days the doctors were beginning to think Etta had Hirschsprung's disease, a life threatening condition which is more common in babies with chromosome abnormalities. They continued with the washouts but Etta could start having milk. We tried breastfeeding but she just wasn't strong enough, so we introduced a bottle, but most of her feeds were through the NG tube. We stayed in Southampton NICU for 2 weeks where I was taught how to do her bowel washouts. It was decided we would continue to do the washouts until Etta was big enough for her biopsy to confirm Hirschsprung's and that way we would hopefully avoid an ileostomy.

We then returned to Dorset County Hospital for a week to establish feeding, and then we were discharged home

where I continued with the washouts.

Etta continued to grow and thrive, she started to take more milk orally and managed to ditch the NG tube completely by the time she was 6 weeks old.

A couple of months passed, then one morning Etta began vomiting before her feed. Her abdomen was swollen and washouts were not helping it to decrease in size. Her poo was also dark in colour and looked abnormal. We took Etta straight to DCH and they then sent her to Southampton. Etta had enterocolitis.

We were in Southampton for 9 days. Etta had antibiotics and her surgeon made the decision to give her an ileostomy and do the biopsy at the same time. The operation was scheduled for the following morning and I signed the consent forms.

On the day of the operation we waited, and waited and waited. An emergency had come in and taken priority. Eventually the surgeon came in and said that the operation would not go ahead but they would still do the biopsy. We would have to come back another day for the ileostomy. We were discharged the following day to wait for the results and continued life and washouts as before.

A week later the results came back negative for Hirschsprung's. I must say I had very mixed feelings about this. On one hand I was so pleased Etta didn't have this condition and possibly wouldn't need surgery. On the other hand we now had no idea why she didn't poo.

We went back to Southampton a month later for a planned admission where the washouts were stopped, to see what would happen without intervention. Etta didn't poo. Her abdomen just got bigger and bigger. After a week of tests and observations the surgeon found that Etta's sphincter muscle does not open on its own. Suppositories open it, so the washouts were replaced with a daily suppository, enemas when needed and washouts to only be done as a last resort.

Etta is now nearly a year old and is still on the bowel management plan. Weaning was a nightmare in the beginning, but slowly things seem to be settling back down. Our surgeon would like to try botox in the near future so we will see what happens.

Emily Mia mum to Hadleigh

I'm Emily, I'm 22 years old and a single parent to 3 beautiful children, Kennedy 7, Elodie 1 and Hadleigh 4 months. My daughter Hadleigh was born 20th January 2021 via emergency c-section at 30+1 weeks, weighing 3lb4oz and even more perfect than I could have ever imagined.

Hadleigh was born prematurely at 30 weeks. She had gone from strength to strength. She was off the ventilator, on low amounts of oxygen, she was showing feeding signs and doing really well for a 30 week-old baby already. But the consultant made me aware that Hadleigh had not passed meconium at all and that her belly was extremely distended and an x-ray had shown some suspicion. This is the first time I heard the term Hirschsprung's disease. He explained to me that with Hadleigh more than likely having T21, there was a greater chance of Hirschsprung's. I sat and listened and then he asked permission to take my tiny little baby to Alder Hey children's hospital, 50 miles from home and from me! Of course I said yes, but I was still being looked after in hospital so had to wait a gruelling 24 hours to join my pretty girl.

Once there, I met the surgeons and they performed a washout. This became the norm for me. I even learnt to do them myself. A washout is like colonic irrigation. You dilate and stimulate the bowel to clear it out. Usually a sign of Hirschsprung's disease is explosive poo's during a washout, and we had plenty of these... Usually 3 times a day! The surgeons let me know that they suspected Hirschsprung's, but I needed to wait and see. They started Hadleigh on antibiotics and oral feeds and monitored her closely. They asked for her to receive 3 washouts a day if she didn't have a bowel movement. Hadleigh has only ever had 2 bowel movements in her whole life.

After a couple of weeks, back and forth over whether I could go back to Stoke to wait for Hadleigh to be able to have the biopsy to diagnose her condition, it was decided that we had to stay in Liverpool and hit a 3kg target; she was only 1.4kg. Many local hospitals will not offer the life saving washouts that are extremely important and this is probably the hardest part of Hadleigh's care. Every time she has a problem with her bowels,I have to take her to Liverpool as my hospital refused to treat her.

Throughout Hadleigh's journey things were tough. Not only was it a battle of all things T21, but it was a battle of all things

preemie too! If she didn't have an illness due to her extra chromie, then it was an illness of prematurity. During our NICU stay we battled bradys and desats (heart rate and oxygen drops), oral feeding issues, weight gain problems, extremely low platelets, anaemia, TAMS scare, infection scares, back and forth oxygen requirements, failed washouts and the WORST...enterocolitis.

Enterocolitis is the BIGGEST thing to know about with Hirschsprung's disease. It is a potentially fatal bowel infection that happens when the bowel isn't being emptied effectively! When Hadleigh was found to have the start of enterocolitis, they started antibiotics immediately. It is always so important to remember to be safe, not sorry, and any little sign we should take seriously! It can be fatal and is so common with Hirschsprung's disease but unfortunately, even more so with Down syndrome.

Toni Holmes mum to Che

A few days after Che was born, doctors in the NICU became concerned about the size of his stomach and the fact that he had not had a bowel movement. He was transferred to BCH by land ambulance and on our arrival we were met by a doctor who told us that they suspected Che had Hirschsprung's disease.

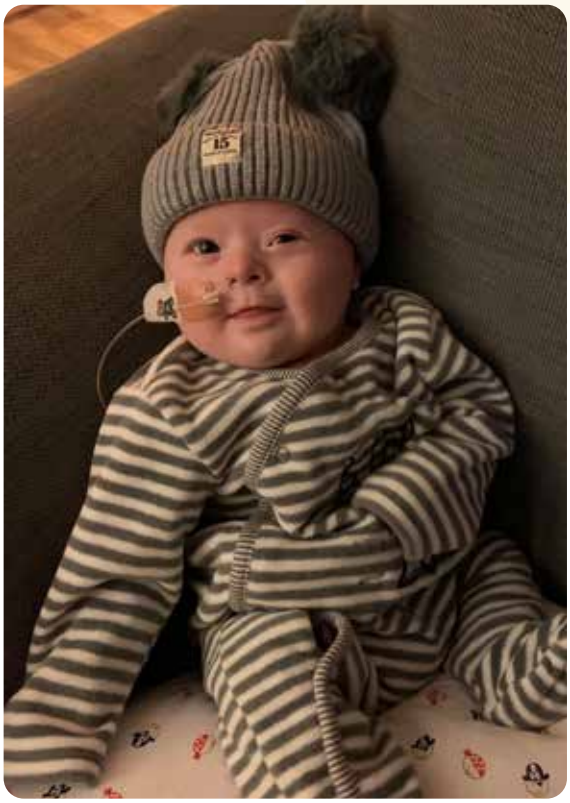
To confirm this, Che would need a biopsy taken. But until then, they would do regular washouts to see if this helped Che to empty his bowels. This involved inserting a small tube into Che's bottom and squirting in water. Washouts happened every few days and did not seem to bother Che at all. In fact, he seemed to almost enjoy them.

At around 8 days old, Che had his biopsy done. This involved taking little bits of Che's intestines to test to see if they had ganglion cells. Unfortunately, not long after the biopsy, at 10 days old, Che became very ill. The washouts were no longer working. He needed emergency surgery to create an ileostomy. This was one of the scariest experiences I have ever had and I remember standing by the door holding back the tears as a large group of doctors and nurses rushed around trying to get him ready for surgery.

As the results for the previously carried out biopsy were not yet ready, Che had another biopsy done while in surgery, which meant that the surgery took a little longer than expected at around 7 hours. The wait felt like forever. Not knowing what was going on and what was taking so long added to the anxiety. Che came straight back to the ward after the surgery and we got to see his stoma bag for the first time. He was so little and the stoma bag almost covered all of his stomach. At this point, it was slightly upsetting to see, but I knew that this operation had saved his life.

At first, it is common for the stoma to pour and it took a team of doctors and nutritionists a little while to play around with foods and medications to get the

perfect balance of input and output to make sure that Che would gain weight. During this time, we were taught by the nurses how to look after Che's stoma



and how to empty and change the bags. It was very scary at first as we were worried we would do it wrong and hurt him. After a while it got easier and soon became the norm.

At six weeks of age, Che was finally ready to go home. The hospital sorted out plenty of stoma equipment for us to take with us and made sure our monthly supply delivery was set up and ready to go. Once home, we set up a stoma drawer and got ourselves organised. Within a couple of weeks, we had the stoma bag change down to just a few minutes. Taking Che out was a little bit daunting but we soon got used to emptying the stoma bag on the go.

Che is now 18 months old and for us, his stoma is just part of who he is! Sometimes we almost forget that it's not the norm. We always have lots of fun trying to empty the bag of a rolling, mischievous, hands everywhere toddler and have more poo disaster stories than you can ever imagine! Poo colour, texture and amount are often our main topic of conversation!

We have a monthly chat with Che's stoma nurse who helps us with any problems we have such as leaking bags or sore skin. We also talk regularly with his surgeon as Che will need further surgery to potentially have his stoma reversed. This has all been delayed because of covid.

Although having a stoma at first was scary and daunting, it is now just part of Che. Emptying the bag and changing it is now just part of our daily routine! It's going to be very strange when we have to deal with our first poo nappy!!

Rachael Forkan mum to Ollie

Goodness...so who'd ever heard of Hirschsprung disease before??

So there we were, on 28th October 2010, with much loved baby boy number three. Reeling from being told they highly suspected Ollie had Down syndrome, then having waited two hours while he was taken away for heart tests (yes he had AVSD as well) for someone to ask us (we were almost packed to leave)...

"Has your baby had a poo?"

"No, but that's not unusual is it? He's not even a day old."

Another four hours later and Ollie was moved into NICU at Redhill Hospital.

"We suspect Ollie is having a problem with his intestines not being able to push the poo along and out." a kind elderly doctor told us. The same doctor, who a few hours before, had tactfully taken my husband aside and asked him if he knew much about Down syndrome.

"He will have a good life, but he might only work in a supermarket... but then plenty of normal people work in supermarkets I suppose." Thanks for that!

So... Ollie spent twenty odd hours in a little see-through box whilst Redhill doctors desperately tried to get rid of him to a more specialised hospital. Great Ormond Street wouldn't take him as he wasn't poorly enough, rightly so, and then the Trevor Mann Baby Unit at the Royal Alexandra Hospital in Brighton said he could go there. We were so relieved but not as relieved as the Redhill team!

We were told that it was very concerning that Ollie couldn't poo independently and that it could be very serious if "things got complicated". (We really didn't think about a bowel perforation at that point and the desperate consequences of that).

I didn't sleep well that night. I spent most of it holding his little hand through the window in the little see-through box. Very much woe is me!

Fortunately, the next day, an ambulance took him to Brighton. I can't tell you how surreal that was, following an ambulance with my two day old son inside, driving down the A23 to Brighton. My husband and I were in total shock I think. Once there, he was settled in and all manner of tests and scans began in earnest.

After another day and sleepless night back in our bed at home, we were told about Hirschsprung's disease. We were told that the plan was for Ollie to have major bowel surgery in about three months. They explained that they thought the last three to four inches of

his intestine did not have the nerves that pushed his poo along and they would cut that part out and pull it down and reattach it. A pull through op! All pretty simple they said. That made complete sense to us and we prepared ourselves for what we would need to do in the meantime.

The nurses were amazing and I will never forget watching my first washout. The first week they must have done washouts when we weren't there (so as not to scare us I imagine) but the second week we watched. My husband could not believe the length of the washout tube that was fed up inside my son's bottom. He winced and closed his eyes as the lovely nurse fed the tube up and up and up his bottom. We were taught to warm up the saline first and then gently inject it into the tube, wiggle the lubricated tube about, massage Ollie's lower tummy on his left side (where his intestines curled around) and then remove the tube slowly and wait for the floodgates to open... and that first time they truly did!! Ollie's poo exploded out all over the nurses apron, very loose of course, as he was only drinking my expressed breast milk via an NG tube. (Another thing I mastered!) Wow! Okay I can do that, I thought! My husband did not think the same thoughts!

We knew Ollie couldn't come home until we could master the aforementioned NG tube and the washouts. I was told community nurses were in my neighbourhood to help us but I didn't want to rely on anyone else. I could do this and I practiced and I did. I owned a big blue bowl from Lakeland, which is designed to wash your oven grills in and decided Ollie would fit in there perfectly with disposable nappy sheets underneath him and the sides would contain the runny poo. Mmmm, mostly it did!

I still have that bowl which I now use to stand my plant pots in and I think back to ten years ago. Goodness. I was amazing!

After about six months, longer than planned, (emergency AVSD repair happened at two months, followed by double pneumonia at four months and a couple of weeks with Ollie on life support); his bowel surgery was scheduled.

I quite proudly will tell you that during the time Ollie was in hospital with his heart surgery and on life support, washouts still had to be done twice a day and I trained over a dozen nurses in how to do it well!!

Ollie had his surgery at the Evelina Hospital in London, where he had had heart surgery and been cared for amazingly well with his double pneumonia. Mrs Richards, (who

has been seeing us for follow up appointments for the last ten years) explained that she was going to do the pull through, but because we had been prodding and poking Ollie's bottom for the last six months instead of three, his insides were likely to be inflamed and irritated. So she wanted them to have a couple of months to settle down before he pushed poo all the way out of them. We nodded at her concurring, not realising that we then had, well I had, yet another new skill to master. That of a colostomy bag!!

This surgery took longer than the open heart surgery. He was gone for eight hours. Such a long time for us to wait. So many cups of tea later, there he was with his "cherry" as the nurses affectionately called his stoma.

I have seen some odd stuff but honestly, watching poo oozing out of your baby's hole in a "cherry" on his lower body was blooming odd! It was proper poo now of course, very soft but lots of it. It was mind blowing.

A wonderful stoma nurse came to meet me and with no messing about, told me I needed to learn how to take care of Ollie, put on a colostomy bag and manage his needs for the next few months. It was overwhelming, but I was determined not to let Ollie down. I could do this. It's not forever! So after some trial and much error I got quite adept at putting the ring (flange) on around the "cherry", being careful to cut my hole as snug as I could to avoid leakages. When I had a good template I kept it and cut out lots more to save time. Goodness it was fiddly. Luckily, with Ollie being little still and also having Down syndrome, he was not rolling about on the floor after his bath but lay quite content as I sang to him mastering the ring and then the bag and the adhesive. It was very tricky but I got quicker and more efficient. I used good old sudocrem cream all around his stoma area as the skin got quite dry and a little sore from being messed about with and Ollie spent maybe thirty minutes a day naked with just the cream on.

MY BIGGEST TIP which I learnt from a fellow mum, as you do and will continue to do along the way is to, wait for it, rub some of their poo from their stoma all around their bottom area as if it had been in a dirty nappy. I did that at least every other day. Reason being, that if the bottom area has not had the joy of being smeared with poo, then when it does, it won't be used to it and I was told that Ollie would get so sore he would cry. It worked!! When he pooed ten weeks later after the colostomy was reversed and the colon was all joined up, no untoward sore bottom in sight!! So when Ollie first pooed out of his bottom, we danced and shrieked and the nurses did too... and the other five mums who knew what I

was in hospital for! It was amazing!

I was also truly humbled in hospital, as on the day we were leaving, a mum and a six year old boy checked into the bed next door. The mum told me that her son had been born without a bottom. Without a bottom!! Imagine that!! Well, that put my woes right into perspective. I often think of that little boy. He will be 16 now!

Right...On to poo then...Honestly, the first couple of years were tricky. Ollie got constipated a lot! We used bucketfuls of Lactulose; a spoonful everyday, upping the dose as he got bigger. I was taught to lubricate my little finger and gently push it up his bottom to help him push. This was usually very successful and a quick result too.

Ollie is ten now and to be honest I had to do this about a month ago, with my index finger, to help him push stuck poo. I hadn't had to do that in a very long time but he was so upset trying to push and I knew he needed help. He knew I would make it better and it worked well.

When he got up on his feet at 2 years old, it all got better. He began sitting on the toilet a lot. He was too big for a potty. Gravity is amazing and I fed him so much fruit and lots of weetabix to keep his poo moving! However, we also had lots of loose days too. No temperature or sign of a cold but his poo smelt and it was very loose. I can't remember what we used back then to block him up, but it quite often meant that he was constipated afterwards for a few days until it settled down again.

This cycle of a few good weeks, a few weeks needing lactulose and a few weeks a bit loose, continued for a few years and made it very difficult to potty train. Ollie didn't have a particular time of day that he pooed either, until he was about five, and at that point most poos were done on the toilet. Hooray!!' I know five years is a long time but all our kids are different.

I think we were very lucky that our surgeon got Ollie's op just about right. She told us it was a very fine line from sewing his intestine back too tightly and then not too loosely either (layman's terms). We were told that there was a chance that Ollie wouldn't have much control of his bowels. His Hirschsprung's is in his sphincter, which can't be transplanted for a new one unfortunately. (Your sphincter is the muscle right at the end of your bottom that you tell to hold tight of your poo when you do need to go but you are in a meeting and it will have to wait. And then when you get to the loo you tell your Sphincter to relax and open up. You probably knew that, sorry).

We were lucky that Ollie never pooed in his sleep. A great omen for the future we were told and that has remained the

case. Ollie poo's on the toilet most days. Proper "big nice poos", his words!, which he loves to show us. Poo has been such an important thing in his and our lives. "Has he pooed today?'' My husband and I ask each other most days and school always tell me if he has. It's still a relief that he has had one that day. I know he can't go more than two days without pooing or it will become very much impacted and too hard to shift, resulting in medicine and stress and then maybe followed by a few loose days which is awful for him. He also learnt to hold his poo, even loose poo, at the age of seven... if we were in the car and I could smell the bottom burps, I would tell him. "No pooing Ollie. We are nearly home. No poo in the car". This has worked really well.

So... we coped with it all. I coped with it all. Still coping with it really. It's been a long time, ten years. I wouldn't wish it back. For all of you reading this; You can do it! You can do anything! Take care and thank you for reading my story so far.

PADS has a private Facebook group for parents and carers of children and young people with Down syndrome who have ARMs about-rectal malformations, Hirschprung's Disease, and who are wanting information and support specifically around toileting.

https://www.facebook.com/groups/dsukhirschprungsdownsyndrome

PADS also has potty training pages where there is more information about the benefits of early potty sitting on bowel function and control.

DSUK Going Potty 0-5

This hugely successful group is for both the parents of and professionals working with children with Down syndrome aged under 5, to encourage and support the child to be out of nappies and become toilet trained.

https://www.facebook.com/groups/219984462212935

Toileting 5+

For parents and professionals working with children aged 5+ around toileting issues.

https://www.facebook.com/groups/2696424470414491

Further information:

https://childrenswi.org/medical-care/gastroenterology-liver-and-nutrition-program/conditions/hirschsprungs-disease

https://www.niddk.nih.gov/health-information/digestive-diseases/hirschsprung-disease

https://maxtrust.org/

Acknowledgement:

We are fortunate enough to work with June Rogers MBE who has extensive knowledge and experience of working with families with children with Down syndrome who also have Hirschsprung's disease.