The Lived Experience: Infantile Spasms

At Positive about Down syndrome (PADS), we work hard to ensure parents have the knowledge and access to support that they need for their children. This publication has been created to provide information about getting a diagnosis for infantile spasms (IS) and tips on advocating for that.

For further information, the UK Infantile Spasms Trust (UKIST) has a brilliant booklet about the tests and treatment. For any medical advice we would always advise you to speak with qualified medical professionals.

This document does not set out to give any medical advice, but rather to raise awareness of IS and to direct you to organisations that can provide further information and support.

What is Infantile spasms?

Infantile spasms is a rare but serious form of epilepsy, which occurs mostly in babies under the age of one year, although occasionally in children with Down syndrome it can begin later. IS affects around 400 children a year, and around 3 per cent of children with Down syndrome. It is also sometimes referred to as West syndrome.

Infantile spasms is caused by chaotic electrical activity in the brain. This activity can result in a range of symptoms, the most obvious being spasms, which are a form of

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seizure. Spasms normally happen in clusters, with a pause of a few seconds between each one. They can look like a head-drop or an exaggerated startle reflex. Some children may also begin to slow down in their development or lose skills, this is known as a developmental regression and is less likely if the spasms are caught early. The chaotic brain activity caused by IS can be seen on a test called an EEG (electroencephalogram). IS is diagnosed by the combination of spasms, loss of skills and an EEG showing chaotic brain activity.

The single most important thing you can do to protect your child is to be aware of what IS looks like so that you can take prompt action if your child has spasms or begins to lose acquired skills. UKIST has an awareness video on their website which all parents of young children with Down syndrome should watch.

IS can look quite subtle but can have big consequences, including increased learning disability and an increased likelihood of seizures in the future. Quick diagnosis and treatment of IS gives your child the best chance of a good outcome.

Remember: there are several things that can cause worrying movements, most of which are not serious. However, if you have any concerns that your child is experiencing spasms you should seek expert advice as it is a medical emergency. It is not the time to worry about making a fuss!

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If your child does have IS, it can be a very scary time. You're not alone – several PADS parents have been on this journey with their children, and UKIST can also offer support.

What to do if you think your child has IS?

Infantile spasms are a medical emergency. If you think that your child is having spasms, you should:

- Try to film the spasms if possible. This will allow a paediatrician or neurologist to review the movements and identify seizures.
- Take your child to the nearest paediatric A&E.
- Ask for your child to be reviewed urgently by an experienced paediatrician and/or neurologist. As IS is relatively rare, it is important that your child and the video (if you have been able to get one) are assessed by someone with experience in IS.
- Insist on an EEG. An EEG is a test which will allow a neurophysiologist to assess the electrical patterns in the brain. IS comes with a distinct pattern of chaotic brain activity called hypsarrhythmia, which can be seen on an EEG. An EEG is the only way to accurately diagnose IS. Not all hospitals have the ability to do EEGs, so you may need to be referred elsewhere. However, it is important this happens quickly (within three days).

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I'm worried that my concerns are not being taken seriously

Infantile spasms is relatively rare and causes damage more quickly than some other common types of epilepsy. The standard NICE guidelines for childhood epilepsy are to have a referral to a specialist within two weeks and an EEG within four weeks, and this will be the protocol most familiar to a non-specialist doctor. **However, IS is different and requires more rapid action**, so there is a chance that you may need to advocate strongly for your child to get them the urgent attention that they need.

The doctor who first sees your child may not have come across IS before – on average a GP would have to practice for 300 years before seeing a case of IS. Therefore, being able to guide a non-specialist doctor to good research and guidelines on IS may help to get the appropriate tests and treatment quickly.

There are some things that it may be worth highlighting to the doctor reviewing your child, particularly if that doctor is not a specialist.

- **Guidelines are that children with suspected IS should have an EEG within three days.** Many forms of epilepsy do not require the same rapid diagnosis and treatment as IS. An example of clinical guidelines, from Sheffield Children's Hospital, and you may wish to share these. Please also see page 13 of this pdf for more information.
- Be clear about what you are asking for: *'I would like my child to be reviewed by an experienced paediatrician today and referred for an urgent EEG in line with specialist clinical guidelines.'*

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- **Spasms can be less obvious in children with Down syndrome.** Low muscle tone can make the spasms less obvious, and the developmental delay associated with Down syndrome means that it can be harder to spot developmental changes or loss of skills. If you feel that your worries are being dismissed because of Down syndrome, this is called 'diagnostic overshadowing'. You could ask, *'how would you deal with this if my child didn't have Down syndrome?' Or point out that 'Given IS is more common in children with Down syndrome than in the general population, I feel that these concerns should be taken more seriously because my child has Down syndrome, as the risk is significantly higher.'*
- **Any delay in diagnosis and treatment can have an impact on outcomes.** A large-scale, study by O'Callaghan et al found that 'increasing lead time to treatment is significantly associated with decreasing developmental score at 4 years in all infants with infantile spasms', and that this was true for any delay in treatment. If you are struggling to get an urgent referral, please ask the doctor to look up this study before making a decision. (O'Callaghan et al, The effect of lead time to treatment and of age of onset on developmental outcome at 4 years in infantile spasms: *Evidence from the United Kingdom Infantile Spasms Study, Epilepsia, 52(7):1359–1364, 2011, doi: 10.1111/j.1528-1167.2011.03127.x)*
- **If you are still worried that your concerns are not being addressed**, it may be worth being explicit about what you feel the risk is: *'Are you 100 per cent confident that delay in referral/diagnosis/treatment will not risk permanent damage, and if not, are you saying that you are comfortable taking that risk?'*

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Jacob's story

One Thursday dinner time in May, Jacob was sitting in his highchair eating his tea, when I thought I noticed his head drop slightly. I didn't think too much of it as he wasn't very used to sitting in a highchair because we had only started weaning him a few weeks before. But I did think it was a bit strange as he had always had good head control. That evening, when he was in bed, I did a bit of Googling about head drops and some information came up about IS. That was when I remembered that I'd read information about this before on the Positive about Down syndrome (PADS) New Parents Facebook group, so I knew that it was more common in babies with Down syndrome.

The next day, my husband noticed Jacob doing some strange movements that he had never made before. He was repeatedly pulling his arms upwards and backwards and scrunching his legs inwards towards his tummy. He did this a few times with a short break in between. We knew something wasn't right. We videoed his suspect movements and phoned the GP, who said that he would refer us to the hospital. But having read more about IS, we knew that this was a medical emergency, so we took Jacob to A&E.

I was prepared for a fight in order to be taken seriously but the hospital staff were fantastic. Jacob's paediatrician was on duty and asked me what I thought the movements were. I shared my concerns that it was IS and he also thought it was possible. Jacob had another cluster of spasms while we were waiting in the hospital which meant that the staff witnessed them first-hand and referred us for an emergency EEG. The EEG showed that Jacob had hypsarrhythmia, the chaotic brain patterns associated with IS, so we were admitted overnight to begin his medication.

Jacob was treated with a high dose of the steroid Prednisolone and the anti-convulsive drug vigabatrin. It was a harsh regime for his little body, but hypsarrhythmia is incredibly damaging

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to the brain, so it was vital to stop this as soon as possible.

Fortunately, within 48 hours of starting the medication, Jacob's spasms had stopped. The next few weeks are a bit of a blur: it was incredibly difficult to administer the medicines to a grumpy and disagreeable baby four times a day; the steroids made him ravenous and sleepy in the day, and ravenous and sleepless at night. We were back and forth to the hospital for checks to ensure that his body was coping with the medicine. Our strong, smiley boy lost his cheeky smile, some muscle tone and any interest in the fun things in life. Instead, he simply sat in his chair or on our laps groaning and rubbing his head from side to side in discomfort. Despite all this, to our delight, two weeks later, a follow-up EEG showed that the hypsarrhythmia was gone.

I won't lie – it was an incredibly difficult time. I had forgotten really, until I sat down to write this!

However, although this time was hard for us all, we were so blessed to receive lots of support. The hospital staff were great and so kind. The parents on PADS New Parents group were supportive and encouraging and we also received fantastic advice from the UKIST support group on Facebook.

Having support from people who have experienced the exact same thing has been invaluable, as they are the ones who can tell you things even the doctors can't. They especially put our minds at rest about the real-life side effects of the medication and prepared us for what might happen during weaning off the drugs. I am so grateful for those who have become Jacob's 'cheerleaders' and have listened to my tired ramblings when the drugs affected his sleep yet again!

Eventually, the time came for Jacob to be weaned off the Prednisolone and then finally the vigabatrin. This was a nerve-racking time (as there was the possibility that the spasms would return), but also one that brought us a lot of joy and relief. Our funny, brave little man emerged from the drug-induced fog, we saw his smiles return and he got stronger every day.

It has been a slow process and Jacob finally completed his weaning off the last medicine. He has some catching up to do after all those months of not making progress; however, he is doing fantastically well. It felt like a long and difficult bump in the road, but we are so proud of our gorgeous boy and all the things he is achieving. We cannot thank Jude and Helen, who posted about IS on the PADS page, enough. Without them, our actions would not have been so prompt, and our story could have been very different.

Amy Price

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Ollie's story

Ollie was born at 36 weeks in January 2020. We had a surprise postnatal diagnosis of Down syndrome, and we came to terms with it quite quickly. He was generally healthy with a small hole in his heart but nothing to worry about. He was a bit slow to feed and a little jaundiced, but he was beautiful!

Within six weeks he was feeding perfectly. By three months he had big smiles and was starting to get better head control. By five months old, he was rolling over. We were living in lockdown therefore I was so grateful for Positive about Down syndrome (PADS) support online.

I had read about IS on the PADS New Parents Facebook group and seen videos and read about warning signs. I thought I noticed Ollie having a few involuntary head drops occasionally, but I tried to ignore it. I didn't want to believe the worst as he was so happy and doing so well.

A month or so later he started making strange movements when he woke up. A bit like the startle reflex. His arms would outstretch, his legs would pull up to his tummy and his eyes got very staring and vacant, slightly popping out or rolling back. He would do this a few times in a row about once a day. It then became more frequent, a few times a day with a few more spasms in succession. I videoed all these episodes as it was suggested to do so in the groups.

I just knew what it was! So I spoke to my health visitor and showed her the videos. She agreed that Ollie needed immediate medical attention. I called the GP who rang ahead to the paediatric ward, and I took him straight there, where he was seen immediately.

After some observations and monitoring, they were able to arrange an EEG quite

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quicky. He had lots of probes and wires on his head to measure electrical activity in the brain. My videos were also shown to a neurologist who diagnosed IS.

It was the worst moment of my life. Way worse than discovering he had Down syndrome. There are so many scary things on Google about it!

However, we had caught it quickly. Ollie had yet to show any real signs of developmental regression.

The medication regime was awful: Vigabatrin and steroids that made him grumpy, sleepy, very puffy and constantly hungry. He was awake all through the night grunting and hungry. So I was constantly breast feeding him. He completely lost his personality and he lost a lot of his muscle tone. But it worked! The spasms were gone in 24 hours!!!

For many babies that is it and then they gradually wean off the medication and are fine.

Unfortunately, Ollie's journey was a long one. Like a very long dark tunnel filled with fear and anxiety. We are well and truly out the other side now and he is doing amazingly.

But, to get here, we had to endure two relapses, two courses of horrible steroids, weeks of upping the dose of vigabatrin to no gain, then finally adding another medication. Then suddenly it

worked! We got there three months after his initial diagnosis, and he has had no more spasms!

However, Ollie was quite developmentally delayed by this point. But, slowly and surely, he caught up, was able to sit for longer and longer, and then he started to stand.

He then began to show signs of another type of epilepsy, focal and absence seizures, but these are not as damaging as IS seizures.

Ollie remained on vigabatrin for one year. When he was finally weaned off, we did have to add in a different antiepileptic medication for the focal and absence seizures.

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Since being weaned of Vigabatrin, Ollie has come on in leaps and bounds. He is also the happiest little boy again now.

He can sit by himself, has started to move around in his own special way, he is standing for longer, he's feeding himself and he is just our wonderful little boy.

The medication he remains on doesn't have anything like the side effects of the initial ones and are keeping the focal and absence seizures at bay too.

The main difference between IS and normal movements or reflexes, is that they are involuntary, so you can't stop or interrupt them by cuddling or distracting the child. They usually happen before sleep or after waking up.

Remember, you know your child best. If something doesn't seem normal, get it checked. My advice to anybody who suspects IS, is video as many episodes as possible. If it is IS, the frequency and number of movements will increase and become more obvious.

It is so important to get seen and treated quickly! Don't panic but, if need be, go to A&E and show them the videos and get an EEG as soon as possible. Don't let them fob you off with a long wait for an EEG.

UKIST is a UK charity raising awareness of IS and it has a Facebook group which you can join and share your videos and ask for advice. There is also a PADS IS support WhatsApp group for families of children who have been diagnosed.

The other piece of advice I would give is that there is light at the end of the tunnel. There is a huge number of different medications that can be used to treat IS and sometimes it takes time with trial and error to find the correct ones for your child. The sooner it is caught and diagnosed, the better the outcome.

The drugs they start with are harsh and it does feel like they lose their personality. But this is short term, and the priority is to treat IS. They will get their personality back I promise.

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Quinn's story

We are sharing our story to raise awareness about IS, as our beautiful little Quinn was diagnosed with IS two years ago. Awareness is so important. IS is a rare form of paediatric epilepsy, but if it hadn't been for another mama's post and a video in PADS group, I may not have been so quick to seek treatment or so pushy for an EEG, which is required to diagnose IS by identifying irregular electrical activity in the brain. Quinn's extra chromosome meant that he was at a higher risk of infantile spasms.

We will be eternally grateful for that mama's post. It meant that I knew what to look for, what to record, and I was well informed when discussing Quinn's treatment with professionals. One of the paediatricians we saw at the hospital was not convinced and I feel had I not known about IS, and what to ask for, he wouldn't have booked the EEG. I know from reading other stories that some people have not been as fortunate in identifying symptoms or getting a diagnosis. The longer that the spasms go uncontrolled, the greater the risk that the development of the child will be affected.

The spasms themselves can be so slight. They are a movement like the startle reflex, often being misdiagnosed as such or as reflux. When we first noticed Quinn's spasms, they were such a small jolt but as we waited over a weekend for our EEG appointments, they progressed quite quickly to more regular 'clusters' of spasms.

Quinn was admitted to hospital immediately following the results of the EEG to start treatment. He also had bronchiolitis, which may well have brought the spasms on in the first place. Treatment for Quinn's IS was a very high dose of prednisolone three times a day for what felt like an eternity. We lost our smiley, happy boy and the steroids were so hard on him. I reached out to support

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groups for their experiences as I worried Quinn would never smile again. The steroids made Quinn distant, angry and hungry all the time. He didn't even look like our baby anymore; his face became so puffy (which is called moon face). It was an extremely difficult time.

Thankfully, the steroids worked first time, and once we started to wean Quinn off them, slowly our boy came back to us.

The first smile was just beyond amazing! Good job too, as the lack of sleep throughout those dark months reminded me that I'm too old to have another child! Quinn still needs to get the memo that 4am is not an appropriate time to start the day, but that's another story!

Quinn is thriving now. He attends nursery and loves spending time with his friends. He loves singing and dancing, and he is an absolute whizz at his phonics cards. He makes us so proud every single day. He still takes medication, sodium valproate, and has regular checks-ups with his epilepsy consultant. We had an appointment recently and the plan is to start reducing the medication soon.

Our boy is an absolute superstar, a warrior, a force of nature, and he makes his Mama and Dada better people! If we can ever help another parent with their journey, we would be honoured. One piece of advice would be... don't Google (easier said than done, I know)! Reach out to our amazing community and the online support groups, as their support was invaluable.

Louise Burry

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Where can I get more information?

The UKIST website and Epilepsy Action both have clear, useful information about IS. UKIST has information on treatment, and a Facebook support group where other parents can provide support.

Some research and guidelines relevant to clinicians:

- Eastern Paediatric Epilepsy Network, Management of Infantile Spasms in Infants Under One Year of Age.
- Lux AL, Edwards SW, Hancock E, et al. The United Kingdom Infantile Spasms Study (UKISS) comparing hormone treatment with vigabatrin on developmental and epilepsy outcomes to age 14 months: a multicentre randomised trial. Lancet Neurol 2005; 4: 712–17.
- O'Callaghan et al, The effect of lead time to treatment and of age of onset on developmental outcome at 4 years in infantile spasms: Evidence from the United Kingdom Infantile Spasms Study, Epilepsia, 52(7):1359–1364, 2011
- O'Callaghan et al, Safety and effectiveness of hormonal treatment versus hormonal treatment with vigabatrin for infantile spasms (ICISS): a randomised, multicentre, open-label trial, Lancet Neurol 2017; 16: 33–42
- Sheffield Children's NHS Foundation Trust, Infantile Spasms (West Syndrome)

With thanks to Jude Simpson for writing this document and her unerring passion to raise awareness of IS. Thanks too to Helen Kel for also supporting PADS parents experiencing IS.

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