The Lived Experience: Infantile Spasms

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At Positive about Down syndrome, 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 around getting a diagnosis for Infantile Spasms and tips on advocating for that.

For further information, UKIST has a brilliant booklet on the tests and treatment, and of course, for any medical advice we would always advocate you speak with relevant medical professionals.

This document does not intend to give any medical advice, but rather raise awareness of Infantile Spasms and signpost to organisations that can provide further information and support.

Infantile spasms: knowing the signs and advocating for your child with Down syndrome.

What are infantile spasms?

Infantile spasms (IS) are a rare but serious form of epilepsy, which occur mostly in babies under the age of one (although occasionally in children with Down syndrome, it can begin later). IS affects around 400 children a year, and around 3% of children with Down syndrome.

IS 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 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 less likely if the spasms are caught early. IS also causes chaotic brain activity, which can be seen on a test called an EEG. You may hear IS being referred to as West syndrome – this is the combination of spasms, loss of skills and 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. UKIST has an awareness video on their website that 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!

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 the UK Infantile Spasms Trust can also offer support.

What should I do if I'm worried my child is having infantile spasms?

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.
- Take your child to the nearest paediatric A&E. You can find your nearest A&E here, and whether they accept children.
- Ask for your child to be reviewed urgently by an experienced paediatrician and/or neurologist. As IS is relatively rare, your child (and the video, if you have been able to get one) must be assessed by someone with experience in IS.
- Insist on an EEG. An EEG is a test which allows 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 that this happens quickly (within 3 days).

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

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

The doctor who first sees your child may not have come across IS before – a GP, on average, would have to practice for 300 years before seeing a case of IS. Therefore, being able to signpost 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 3 days.** Many forms of epilepsy do not require the same rapid diagnosis and treatment of IS.

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 clinical guidelines."* The doctor may refer to NICE guidelines, which state that children with epilepsy should be seen within 2 weeks and have an EEG within 4 weeks. This is not appropriate for IS – please refer the doctor to the Sheffield clinical guidelines and the study by O'Callaghan et al below.

- **Spasms can be less obvious in children with Down syndrome.** Low 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 then the general population, I feel that these concerns should be taken more seriously because she has Down syndrome, as the risk is significantly higher."*

- **Any delay in diagnosis and treatment can have an impact on outcomes.** A large-scale, international 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 concerned that your concerns are not being addressed**, it may be worth being explicit about what you feel the risk is. *"Are you 100% confident that a delay in referral/diagnosis/treatment will not risk permanent damage, and if not, are you saying that you are comfortable taking that risk?"*

Jacob's Story

One Thursday dinner time in May, Jacob was sitting in his highchair eating his tea, when I thought I might have 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'd only started weaning him a few weeks before, but I did think it was a bit strange as he has 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 Infantile Spasms. That was when I remembered that I'd read information about this before on the PADS New Parents Facebook group, so I knew that it was much more common in babies with Down syndrome than in the rest of the population.

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

I was prepared for a fight 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 and I shared my concerns that it was Infantile Spasms 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 on the Monday. The EEG showed that Jacob had hypsarrhythmia, the chaotic brain patterns associated with Infantile Spasms, 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 the hypsarrhythmia is incredibly damaging to the brain and so it is 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 and our strong, smiley boy lost his cheeky smile, 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'd 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 Parent 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, especially putting your mind at rest about the real-life side effects of the medication and preparing you 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 wean off the Prednisolone and then finally the Vigabatrin. This was a nerve-wracking time (as there is the possibility that the spasms will return), but also one that brought us a lot of joy and relief as our funny, brave little man emerged from the drug-induced fog and we saw his smiles return and he got stronger every day.

It has been a slow process and Jacob only finally weaned off the last medicine about 8 weeks ago. He has some catching up to do after all those months of not making progress; however, he is doing fantastically. It has 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, and we cannot thank the people who posted about Infantile Spasms 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

Ollie's Story

My little boy Ollie was born at 36 weeks in January 2020. A surprise postnatal diagnosis of Down syndrome, but we came to terms with it, he was generally healthy - a small hole in the heart but nothing to worry about, a bit slow to feed and quite jaundiced, but beautiful.

Within 6 weeks he was feeding perfectly, by 3 months big smiles and starting to get better head control, by 5 months he was rolling. We were living in a lockdown, I was grateful for PADs support online and things were generally good.
I had read about Infantile Spasms on the PADs new parents' group with videos and warnings. I thought I noticed a few involuntary head drops but tried to ignore it, I didn't want to believe the worst - he was so happy and doing so well.

A month or so later he started doing strange movements on waking a bit like the startle reflex, arms outstretched, legs pulled up to his tummy and his eyes got very staring and vacant slightly popping out or rolling. He would do this a few times in a row once a day. It then became more frequent a few times a day a few more spasms in succession. I videoed all these episodes.
I just knew what it was. I spoke to my health visitor showed her the videos, she agreed this 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 quickly, lots of probes and wires on his head to measure electrical activity in the brain. That, alongside my videos, were shown to a neurologist who diagnosed Infantile spasms.

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 caught it quickly he had yet to show any real signs of developmental regression.

The medication regime was awful, vigabatrin + steroids it made him grumpy, sleepy, very puffy, and constantly hungry. He was awake through the night grunting and hungry. So, I was constantly breast feeding through the day. He completely lost his personality, and he lost a lot of 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 2 relapses, 2 courses of horrible steroids weeks of upping the dose of vigabatrin to no gain then finally adding another medication to his vigabatrin. Then suddenly it worked we got there 3 months after his initial diagnosis no more spasms!
He was however quite developmentally

delayed by this point, but slowly and surely, he caught up able to sit for longer and longer starting to stand.

He did then show signs of another type of epilepsy focal and absence seizures, but they are not damaging in the same way as IS.
He remained on Vigabatrin for 1 year.
When he finally weaned off, we did have to add in a different antiepileptic medication for the focal absent seizures.
Following the wean from Vigabatrin he's come on leaps and bounds he's really starting to catch up, he's the happiest little boy.
He can sit himself up, starting to move around in his own special way, standing

for longer, feeding himself and he is a wonderful little boy.

The medication he remains on don't have anywhere near the side effects and are keeping the focal and absence seizures at bay.
My advice to anybody who suspects IS is to get as many videos as possible, if it is IS the frequency and number of movements will increase and become more obvious.

The main difference between IS and normal movements or reflex is it's involuntary so you can't stop them by cuddling them or distracting them. It usually happens before sleep or on waking.

It is important to get seen and treated as soon as possible, don't panic but, if needs 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 for IS and they have a Facebook group to join and show videos and ask advice.

We also have as PADS IS support WhatsApp group for those diagnosed.

You know your child best so if something doesn't seem normal get it checked.
The other advice I would give is there is light at the end of the tunnel, there are a huge number of different medications that can be used to treat it and sometimes it takes time and trial and error.

The sooner it is caught and diagnosed the better the outcome and they will catch up slowly and surely developmentally. The drugs they start with are harsh and it does feel like they lose their personality but its short term and its what's needed to treat IS. They will get their personality back I promise.

Quinn's Story

We're sharing our story to raise awareness for Infantile Spasms week, as our beautiful little Quinn was diagnosed with IS two years ago almost to the day, in November 2019. Awareness is so important - infantile spasms are a rare form of paediatric epilepsy, but if it hadn't been for another Mama's post and video in the Positive About Down Syndrome group back in August 2019, 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, called hypsarrhythmia. Quinn's extra chromosome meant that he was at a slightly 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 was informed when discussing with professionals. One of the paediatricians we saw at PAU was not convinced and I feel had I not known about infantile spasms, and knew what to ask for, he wouldn't have booked the EEG. I know from reading others' stories that some have not be as fortunate in identifying or getting a diagnosis. The longer that the spasms go uncontrolled, the greater the risk is that development will be affected. The spasms themselves can be so slight, a movement like the startle reflex, often being misdiagnosed as such or as reflux. When we first noticed Quinn's, they were such a small jolt but progressed quite quickly to more regular 'clusters' of spasms whilst waiting over a weekend for our EEG appointment.

Quinn was admitted to hospital immediately following the results of the EEG to commence 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, the steroids were so hard on him. I reached out to support 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 (moon face). It was an extremely difficult time.

Thankfully, the steroids worked first time, and once we started to ween Quinn off, 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 however, but that's another story!

Quinn is thriving now, he attends nursery and loves spending time with his friends, loves singing and dancing, and is an absolute whizz at his phonics cards, he makes us proud every single day. He still takes medication, Sodium Valporate, and has regular checks ins work his epilepsy consultant. We had one recently in fact, and the plan is to start reducing the medication in the New Year. 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, don't Google (easier said than done I know), reach out to our amazing community and to the UK Infantile Spasms Trust Support Group (West Syndrome) on Facebook, their support was invaluable.

Louise Burry

Resources

Website

https://ukinfantilespasmstrust.org/

Video - An introduction to Infantile Spasms for children with Down syndrome

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.

Research and guidelines relevant to clinicians are:

- 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