Aicardi Syndrome

Corpal

DCC & Aicardi Syndrome UK

What is Aicardi Syndrome?

Aicardi syndrome is an incredibly rare genetic condition. The cause is not known. Because it occurs predominantly in girls, it is believed that it is linked to an X chromosome and is the result of a genetic change very early on in development. It is believed to be a first-time genetic change and is not hereditary.

Aicardi syndrome is diagnosed through MRI and an eye exam. Most cases are diagnosed due to the classic three features. These will be introduced further on in this booklet.

The early stages of diagnosis are a frightening and confusing time for families. In this booklet you will find messages from families who have children with Aicardi syndrome, which we hope will offer comfort and perhaps a little clarity.

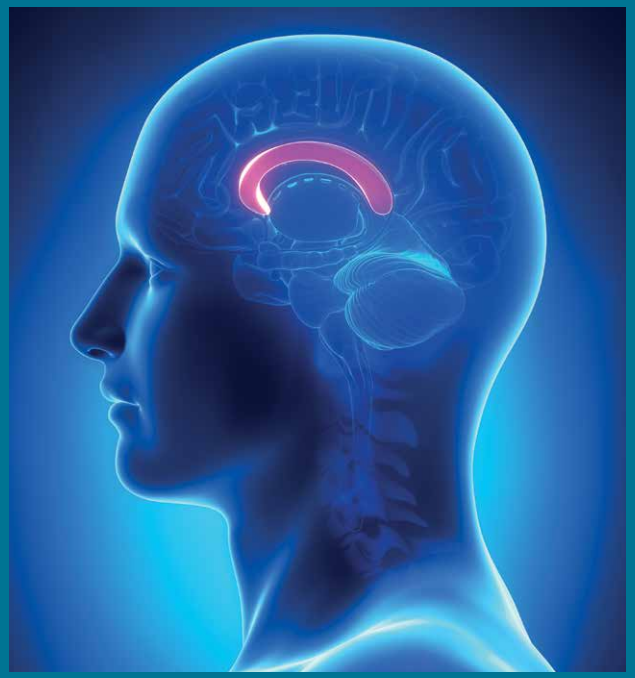
The Classic Diagnostic Triad

1. **Infantile spasms** are a form of epileptic seizure that cause a child's upper body and arms to rhythmically stiffen in clusters. Children with Aicardi syndrome will usually have this type of seizure along with other types. For most people with Aicardi syndrome, seizures become an aspect of day-to-day life. There are many different anticonvulsants and therapies to try which can give some control to symptoms.

2. **Chorioretinal Lacunae** are lesions on the retina of the eye. These may cause sight impairment and blindness. From a young age children can have access to ophthalmology to give their vision the best support. For those who have more significant visual impairment, there are many opportunities to support their other senses through play and therapy.

3. **Agenesis of the corpus collosum** means that the major pathway between the two sides of the brain is either fully missing, partially missing or malformed. For the majority of people with Aicardi syndrome this is one of several brain malformations that occur.

The Corpus Collosum



The corpus collosum is a structure which connects both hemispheres of the brain. There are other pathways by which the two hemispheres of the brain can "communicate" but the corpus collosum is the main one.

Every child born with Aicardi syndrome is different. There is no way, at this early stage, to fully anticipate how they will progress. Some children are significantly affected and require a lot of support, care and intervention through out their lives. Occasionally there are children diagnosed who are less affected and only experience minor developmental delays. Wherever your child lands on this spectrum, it is a challenging diagnosis and it is natural to have a lot of questions. Here are some answers to questions that may be on your mind:

- Most children diagnosed with Aicardi syndrome will require support from physiotherapy, occupational therapy, neurology, paediatrics and eventually special educational needs teams.

- Many children will have difficult to treat seizures and most will need to remain on anticonvulsants throughout their lives.

- Some children will learn how to walk and talk, but many will not. This does not mean to say they won't learn their own way of communicating. You'll be amazed at what can be said without using words.

- Many people with Aicardi syndrome require tube or peg feeding. If this is the case for your child, you will be supported by a team to become an expert in using this equipment.

- The majority of children with Aicardi syndrome have significant developmental delay. When hearing this, it can be easy to focus on the things your child won't learn to do. It is important to remember that they will develop into their own wonderful person, who will thrive on your love and care.

- Some children with Aicardi syndrome experience scoliosis and there is a chance that they will need to use a brace or have surgery when they are older. Your child's physiotherapist will keep an eye on this.

- There is nothing you could have done to have stopped this from happening. It is believed that Aicardi syndrome is caused by a first-time genetic change in early development. Neither the father nor the mother is to blame for this and it is no one's fault.

- Building a team you trust around your child, who can signpost you to appropriate support depending on when you need it, will help you to feel more in control on this journey. Speaking to your health visitor, GP, local carers centre and local disability groups can help you to find the information you need. You can also contact us at Corpal and we can signpost you to online groups and useful websites.

- Although this is a scary time, you are your child's advocate and champion. You will be amazed at what you are capable of handling as time goes on. You will discover so much happiness, joy and strength together. Please read the following messages from families who have a child with an Aicardi syndrome diagnosis.

Messages from families

“On 23 February 2011 we welcomed our daughter into the world.

We were lucky we got an early diagnosis. After a difficult labour our daughter was sent to ICU and within 24 hours, they confirmed fluid on her brain.

We were transported to the children’s hospital where she had many tests and after 4 days, we had our diagnosis. It crushed us completely. Our neurologist was pretty informative and he did tell us what to expect. However, we were then just sent home alone to wait for the seizures to get worse and oh boy they did. I wish we had been given full support from the start. I think any child with such a serious diagnosis should have access to outreach nurse etc from the start; someone who can be their link to all the medical help they need.

My help eventually came from the Aicardi support group on Facebook. It took me a while to join the group but it really helped me in the early days. My daughter is 10 next month and we have fought to make sure we have help and support around us. I really think connecting with people in the same world helps so much, you can ask anything and won’t be judged.”

– A Mother

“There is no denying that receiving a diagnosis of Aicardi syndrome is life changing for everyone involved. As a mum, I was devastated when my only daughter was diagnosed at 8 weeks. I very quickly decided to try not to focus on long term worries but to focus on enjoying life with her. She has taught me so much and brings joy to everyone she meets.

She has developed her own amazing personality and is very feisty. In a short space of time, I discovered some really great groups and have met some fantastic families. It’s great to be able to chat to other people who are going through what we are so I would say that it’s worth reaching out to other families.

It’s also definitely important to know that everyone’s journey is very different. At the start, I wanted to do so much research but I read a lot of terrifying information so instead I try to focus on positive things. By far the most difficult thing is the seizures which are hard to manage and it’s horrible watching your daughter suffer. It’s a constant battle to get medications correct but we decided early on that we would do what we felt was best so instead of giving Evie a lot of medication, we try to manage the seizures but allow her to develop. Allowing Evie to develop in her own time and having a lot of patience is what I find helps me most. Every little thing she learns is amazing to us so we just try to give her the best possible quality of life.”

– A Mother

"The day my granddaughter was diagnosed with Aicardi Syndrome was truly life changing. It is a whirlwind of confusion and a bit of heartbreak for my grandchild and her parents.

Having such a special, precious person in your life puts everything into perspective and you quickly learn that you only need a smile or very small development to help you stay positive.

Reach out! Never be afraid to ask for help. Learn about the seizure medications, research new skills like baby yoga and baby massage. Reach out to other parents to learn from them and become knowledgeable on medication and coping mechanisms.

The most important thing is that your Aicardi princess will surprise you every day and make you feel so blessed and privileged to be part of their life.
At the age of two, our Princess loves people, loves nature, horse riding, school, swimming and partying!

– A Grandmother *"*

"The days leading up to and after our daughter's diagnosis were some of the darkest days we have experienced as a family. We were given a list of all the things wrong with our daughter with very little in the way of what we could expect or initial support. Everyone will deal with this differently and it was so important for my wife and I that we were there for each other, while our extended family offered words of support, we were very aware that as parents we would have to change our life significantly to ensure our daughter has the best life possible.

My wife and I both have had different approaches to processing our daughters' condition, she delved right in to researching everything she could so that she knows everything there is to know about Aicardi syndrome. I have found it better personally to distance myself from the condition, not speculate too much and deal with change as it comes. Neither is right or wrong but it was important that we both understood that we have different approaches.

Over the years we have built up a fantastic support team of therapists and medical professionals with whom we have a great relationship with. They have helped so much with equipment, advice, therapy, funding and emotional support. Make sure you build good relationships with the professionals that are there to help.

Finally, the thing that gets me through every day is our fantastic daughter. I don't see a disability or condition I see our funny, cheeky, affectionate daughter. There will be many ups and downs but regardless of how down I am feeling one smile from her changes everything. Having a happy child will go a long way in helping you and your family get through this."

– A Father

Support

- Parent led Phone helpline to assist with information and emotional support at point of diagnosis and beyond.

Educate

- Collaborating with Healthcare Professionals, Foetal Medicine Units & Obstetricians to promote understanding of DCC.
- Engaging with Educators & Parents and building a platform for knowledge based learning.

Connect

- Our DCC network brings families and adults together through regular events and active social media platforms, while engaging with the IRC5 international research community.

Get Involved

- Join Corpal as a Supporter, spare some time & share your skills or donate via Justgiving or Paypal to help support our ongoing work. Visit **www.corpal.org.uk** to find out more.

Get Inspired

- From Bake Sales, to Marathons our Supporters actively fundraise for Corpal.
- Share your stories and challenges to inspire others on the journey.

Keep in touch

Phone: +44(0)7427 429388

Facebook: Corpal

Email: info@corpal.org.uk

Twitter: @officialcorpal

Web: www.corpal.org.uk

Instagram: @corpalcharity

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DCC & Aicardi Syndrome UK

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