



# Welcome

## DYRK1A SYNDROME

### Get Started

#### STEP 1

##### GET SUPPORT

- Community
- Meetups



#### STEP 2

##### GET INFORMED

- Common Questions



#### STEP 3

##### GET INVOLVED

- Research
- Volunteer



✉ [dsia@dyrk1a.org](mailto:dsia@dyrk1a.org)

🌐 [www.dyrk1a.org](http://www.dyrk1a.org)

📱 [@dyrk1a.syn](https://www.facebook.com/dyrk1a.syn)

📷 [@dyrk1asyndrome](https://www.instagram.com/dyrk1asyndrome)

🐦 [@dyrk1aorg](https://twitter.com/dyrk1aorg)

📌 [@dyrk1asyndrome](https://www.pinterest.com/dyrk1asyndrome)

### WE ARE YOUR COMMUNITY



Welcome to the DYRK1A Syndrome community. Receiving this diagnosis is met with a wide range of feelings from shock and fear of the unknown to relief of finally having answers. You're not alone. Many in this community can relate and want you to know it's OK to have those feelings. Together, we will build an inclusive community that redefines normal for our loved ones living with DYRK1A Syndrome.

### UNDERSTANDING THE DIAGNOSIS



You may have many questions about how DYRK1A Syndrome might affect your child and family. It is best to ask your doctor or specialist any medical questions you may have. However, because this is a rare diagnosis, literature is limited and symptoms present differently from person to person they may not have the answers you're looking for.

With the help of our medical & scientific advisors, we have put together answers to some of the most common questions which you will find in Step 2. We are hopeful that with more information gathered from our registry we will be able to address more questions and shed light on this rare condition. Connecting with other families, virtually or in person, is another way to help understand your loved one's diagnosis.



# Get Support

DYRK1A SYNDROME



## Adrienne Apodaca

*"I felt so alone even though I was surrounded by friends and family who adored Emily and loved our family. I couldn't seem to shake this overwhelming sense of sadness and loneliness caused by Emily's diagnosis.*

*I needed this community more than I realized. I needed to be able to cry and have people understand why I felt the way I did. I needed to hear other people say their children had the same victories and struggles that Emily does. I needed to see other kids with DYRK1A to know everything would turn out okay.*

*We are no longer alone, we found our second family."*

 [dyrk1a.org/fbgroup](https://dyrk1a.org/fbgroup)

 [dyrk1a.org/conferences](https://dyrk1a.org/conferences)



### ONLINE COMMUNITY

One easy way to connect with other families is to join our private online community on Facebook. We work hard to keep this a safe and supportive space. Apart from discussion threads, you will find information, resources, a map of self-identified families that have received the diagnosis, and more. Connecting internationally is not a barrier, with the translation feature families can communicate with each other regardless of language.



### LOCAL COMMUNITY

Since our DYRK1A community is so rare and spread out across the world, it is important to find support in your local community. Every local community may have different offerings and it may help to find a local organization to help point you in the right direction.



### FAMILY MEETUPS

In-person gatherings are another way to connect with other families. The US & UK organizations hold an annual event where families can meet one another, listen to speakers on a variety of topics and enjoy some fun activities. Families can also plan smaller gatherings with other families that are nearest to them.




# Get Informed

## DYRK1A SYNDROME



### Amy Clugston

*"From the moment Lorna was born I had so many questions and concerns that couldn't be answered. There are no published articles or list of common symptoms when they are undiagnosed. Even after we found her change on the DYRK1A gene in 2014 and 18 years later, there was very little information. I was left speechless when I saw an article with a picture of a girl that resembled my daughter even though their change was different. I'm excited to know families will now have more information when they get a diagnosis"*

-  [dyrk1a.org/questions](http://dyrk1a.org/questions)
-  [dyrk1a.org/research-updates](http://dyrk1a.org/research-updates)
-  [dyrk1a.org/genereviews](http://dyrk1a.org/genereviews)



### COMMON QUESTIONS

With the support of our Medical & Scientific Advisory Committee, we were able to answer some of the most common questions people tend to have about DYRK1A Syndrome. These questions and answers can help you become more familiar and better able to explain it to others.



### PUBLISHED RESEARCH

There is published research about the DYRK1A gene though it is more limited when it comes to DYRK1A Syndrome specifically. We've gathered a list of existing data and included it on our website. If you are unable to access a particular study due to a paywall or registration, please reach out as we may be able to share a copy. The technical language in which these studies are written may be difficult to understand. We welcome your questions and encourage you to discuss them amongst the community.



### COMMON SYMPTOMS

According to GeneReviews, DYRK1A Syndrome has a recognizable pattern of symptoms that can be seen clinically. They include the following (listed most to least common): Developmental delay or intellectual disability, speech impairment, microcephaly, feeding problems, similar facial patterns, eye abnormalities, epilepsy, gait disturbances, underweight, Autism Spectrum Disorder, short stature, sleep disturbance, urogenital anomalies, hypertonia, gastrointestinal problems, hyperactivity, anxiety, cardiac defects, dental anomalies & musculoskeletal features.



# Get Involved

## DYRK1A SYNDROME



### Ana Downey

*"We are eager to learn more about DYRK1A syndrome and help others lead more fulfilled lives. Our way to contribute is providing data, so scientists can help put the pieces together. Luna started her participation in the "Tiger Study" this past April. The data collection is 100% virtual and they are great to accommodate the family and kids time. It is a big time commitment, but the Tiger team broke up some sessions to make sure Luna could participate in full. We can't wait to see where all the research can take us."*

-  [dyrk1a.org/research](http://dyrk1a.org/research)
-  [dyrk1a.org/volunteer](http://dyrk1a.org/volunteer)
-  [dyrk1a.org/donate](http://dyrk1a.org/donate)



### PARTICIPATE IN RESEARCH

There is currently a small grainy snapshot of what DYRK1A Syndrome is and how it could affect people. As more people take part in the research and data collection opportunities, researchers can help piece together a clearer picture of DYRK1A Syndrome. There are a few efforts underway to collect data that will inform future studies. We encourage you to visit our website to learn how to volunteer data anonymously and check back for new opportunities.



### VOLUNTEER TIME OR EXPERTISE

We rely on volunteers who provide their time and/or expertise for the programs and opportunities that are currently available. We can always use more volunteers to help us accomplish the goals we have set out in our strategic plan that will ultimately improve the lives of families that have individuals with DYRK1A Syndrome. If interested, you can find and complete the volunteer form on our website.



### DONATE TO OUR CAUSE

We fundraise throughout the year to help us achieve our mission and we are grateful for the donations our community has generated. If you'd like to support, donations can be made with cash, check, or credit card. Details can be found on our website. Other ways to give include employer donation matching programs, participating in our annual run, Facebook fundraisers, shopping with AmazonSmile, and buying t-shirts on Bonfire.

### WHAT IS DYRK1A?

DYRK1A is another name for the dual specificity tyrosine phosphorylation regulated kinase 1A gene. It is located on chromosome 21 in the q22.13 region and provides instructions for making a kinase enzyme. The DYRK1A enzyme is important in the development of the nervous system.

### WHAT IS DYRK1A SYNDROME?

DYRK1A Syndrome is described by a specific set of signs and symptoms. These signs and symptoms can include; intellectual disability, speech impairment, feeding difficulties at birth or during infancy, epilepsy or history of seizures, autistic traits, reduced head size (microcephaly), and certain facial characteristics.

### WHAT CAUSES DYRK1A SYNDROME?

DYRK1A Syndrome is caused by genetic changes that occur in the DYRK1A gene which affects the function of the DYRK1A protein. Genetic changes most often happen very early in a pregnancy, during or soon after conception. These genetic changes are often called variants or mutations. This gene has an important role in regulating the function of many other proteins in the cell. However, some variants in the DYRK1A gene may not affect the protein.

### HOW IS DYRK1A SYNDROME DIAGNOSED?

DYRK1A changes can be found with molecular genetic testing. Chromosomal microarray analysis (CMA) can detect large deletions or duplications that affect the DYRK1A gene. If a change is too small to be detected there are a few other methods. Multigene panels are used to look for changes in groups of genes related to a specific symptom. The DYRK1A gene is included in multigene panels for intellectual disability (ID), microcephaly, autism & epilepsy. Whole exome or genome sequencing can also detect changes in the DYRK1A gene. A geneticist can explain the results of these tests.





# Common Questions

## DYRK1A SYNDROME

### WHAT VARIANTS ARE FOUND IN DYRK1A SYNDROME?

The variants reported in current research articles cause a loss of functional DYRK1A protein from one of the two copies of the gene. There are many different variants that can lead to a loss of function to the protein. These could be variants that lead to an absence of DYRK1A expression, or to a protein missing one part of its sequence, and sometimes to a protein with a very subtle change in its sequence, but this small change is sufficient to impair its activity.

### DO DIFFERENT VARIANTS CAUSE DIFFERENT SYMPTOMS & SEVERITY?

There is no evidence to date that different variants also present different symptoms or severity. It is possible though not certain that variants that only partially affect the DYRK1A activity if they exist, may lead to less severe clinical manifestations. In contrast to other syndromes that result in forms of intellectual disability, there are similarities among individuals carrying a pathogenic variant in DYRK1A. We hypothesize that other factors, from genetic or environmental origins, may influence the clinical manifestations and the severity of the syndrome.

### IS 21Q22.13 DELETION THE SAME?

Yes, DYRK1A is the gene responsible for the symptoms observed in the individual carrying a deletion of this 21q22.13 region. A deletion may also be described as a partial deletion or micro deletion. Individuals with 21q22.13 deletion and anomalies/variants in DYRK1A present the same clinical manifestations.

### HOW IS DYRK1A SYNDROME CONNECTED TO DOWN SYNDROME?

People are typically born with 23 pairs of chromosomes, 46 chromosomes in total. In Down Syndrome, there are three copies of chromosome 21, and therefore three copies of the DYRK1A gene. With three copies of the DYRK1A gene and then makes too much of the DYRK1A protein. You may see research that aims to inhibit or decrease the impact of this third copy. The reason this approach does not work for people with DYRK1A Syndrome is that while there is a pair of chromosome 21, one of these chromosomes has a missing or faulty DYRK1A gene.

### WHAT SHOULD BE EXPECTED OF A CHILD'S DEVELOPMENT?

Each person with this diagnosis is unique and may not be susceptible to developing all the features that have been reported. It is common to present with a delay in psychomotor development, significant difficulties with communication and language, and epileptic seizures in early childhood, which may or may not continue into adolescence and adulthood. Some behavioral issues such as anxiety or stereotypic behavior, often referred to as stimming and are a common trait among autistic individuals may occur. They may have feeding challenges beginning at birth with difficulty coordinating a suck, swallow, and breath pattern that can progress with strong preferences to different textures.

### IS THERE A CURE?

There is currently no cure for DYRK1A Syndrome.

### WHAT ARE THE TREATMENTS?

Early intervention and regular therapies such as speech, occupational, and physical therapies can help manage symptoms and also help reach developmental milestones. Medical conditions related to DYRK1A Syndrome are treated by multiple different specialists.

### WHAT IS THE PREVALENCE OF DYRK1A?

It is difficult to know the exact number of cases because a formal registry does not exist. However, according to large-scale studies, around 0.3% to 0.5% of individuals with intellectual disability are likely to carry a genetic anomaly (variant) in DYRK1A based on genetic testing. The number of individuals identified will increase in the coming years as genetic testing becomes more utilized. As an example, there are more than thirty affected individuals identified in France and we expect ten more identified per year.



# Organization

DYRK1A SYNDROME US

TIN: 83-3342162

## Board of Directors

### Amy Clugston - President

- DYRK1A Parent
- Internal Affairs Chair

### Priscilla Guzman - Treasurer

- DYRK1A Parent
- Finance Lead

### Adrienne Apodaca - Secretary

- DYRK1A Parent
- Family Support Lead
- Meetup Committee Lead

### Ana Downey - Director

- DYRK1A Parent
- External Affairs Chair
- Fundraising & Awareness Lead

### Johnna Graves - Director

- DYRK1A Parent
- Volunteer Lead

### Sarah Glass - Director

- DYRK1A Parent
- Medical / Scientific Chair



✉ [dsia@dyrk1a.org](mailto:dsia@dyrk1a.org)

🌐 [www.dyrk1a.org](http://www.dyrk1a.org)

📍 1745 Lorna Ln  
Otsego, MI 49078

## DYRK1A SYNDROME US



DYRK1A Syndrome US is a nonprofit 501c3, a tax-exempt organization with the main purpose of improving the quality of life for those affected by DYRK1A Syndrome. Through support, education, research, and collaboration we are working to improve family life, daily living, and clinical care for those with DYRK1A Syndrome. The objectives identified in our strategic plan for the years 2021 to 2024 are to; build organizational capacity, provide support and education to the DYRK1A patient and caregiver community, provide education and outreach to the DYRK1A clinical and scientific communities, push the envelope in research, patient care and therapies in ways that significantly improve the quality of life for DYRK1A patients.

The organization's accomplishments are all made by volunteers, people who give their time and experience to the mission. The Board of Directors meets quarterly, three times virtually and once in person. We have two main committees that meet monthly, internal affairs & external affairs. The Internal Affairs Committee handles all the internal and operational areas of the organization which includes; governance, finances, and volunteers. The External Affairs Committee handles areas of the organization that are external, such as; family support programs, meetup conferences, fundraising, communications, public relations, and marketing.

We are also in charge of maintaining a unified international community of families and professionals focused on improving the lives of individuals affected by DYRK1A Syndrome through the DYRK1A Syndrome International Association.





# Organisation

THE DYRK1A UK COMMUNITY  
Registered Charity Number 1179083

## Charity Trustees

### Janet Banks - Chair

- DYRK1A Parent
- Meetup Committee

### Gemma Brady - Treasurer

- DYRK1A Parent
- Meetup Committee

### Tably Wright

- DYRK1A Parent

### Lynne Hearn

- DYRK1A Parent

### Aaron Johnson

- DYRK1A Parent



✉ uk@dyrk1a.org

🌐 www.dyrk1a.org

📍 32 Belgrave Road  
Tamworth, Staffs  
B77 2LW

## THE DYRK1A UK COMMUNITY



The DYRK1A UK Community is a registered charity in the UK with the main purpose of raising funds to support individuals with DYRK1A syndrome and their families. The funds are primarily used to hold annual meet-ups that allow families to learn more about the condition from professional speakers. The meet-ups also allow families the opportunity to socialize, enjoy family fun activities, and share valuable experiences and advice.

In addition to the charity trustee meetings, we have a Meet-up committee that handles all the details for the Meet-up. The organisations accomplishments are made by all of the volunteers.

### Meetup Committee Members

- Bhavana Mullanpudi, DYRK1A Parent
- Gemma Brady, DYRK1A Parent
- Janet Banks, DYRK1A Parent
- Frances Bush, DYRK1A Parent
- Kelly Mcleod-Andrews, DYRK1A Parent
- Keren Franks, DYRK1A Parent
- Magdalena Pachucy Jalocha, DYRK1A Parent
- Sarah Stevenson, DYRK1A Supporter



# Organization

## SPANISH ASSOCIATION OF DYRK1A SYNDROME

### Board of Directors

**María Ángeles Alcalá Del Águila**  
President

- DYRK1A Parent

**María Jesús Pérez Llerena**  
Vice President

**David Conesa Garcia**  
Secretary

- DYRK1A Parent

**María Teresa Pérez Mariné**  
Treasurer

- DYRK1A Parent

**María Del Mar Pagán Solana**  
Member

- DYRK1A Parent

**Palma Milagrosa Rivera Castro**  
Member

- DYRK1A Parent



+34 617 76 79 62



info@aesdyrk1a.org



www.aesdyrk1a.org



Avenida Juan Carlos I 41  
30700 Torre-Pacheco  
Region of Murcia

### SPANISH ASSOCIATION



The Spanish Association of DYRKIA Syndrome was established in January 2022 and has as its goals: The investigation and dissemination of DYRKIA syndrome, through scientific/medical personnel in hospital centers and national laboratories. As well as the search for new cases and the acceptance of affected families at the national level, for the exchange of knowledge and experiences that can help the daily lives of families.

Fundraising will be carried out, with sporting and cultural events to obtain economic resources that will be used exclusively for scientific research on the syndrome.

Seminars, assemblies, and webinars will be prepared with the participation of medical and scientific personnel. We will give visibility to the syndrome through social networks and other means that we have at our disposal.



# International

## DYRK1A SYNDROME

### Member Organizations

#### DYRK1A Syndrome US

- Amy Clugston
- Board Members

#### The DYRK1A UK Community

- Janet Banks
- Trustees & volunteers

#### Spanish Association of DYRK1A Syndrome

- Mari Angeles Alcalá
- Board Members

### Representatives

Australia - Tracey McDonnel

Canada - Janet Thompson

Denmark - Linda Klindrup

France - Julie Duval Derrien  
• Valérie Gonzalez

Hungary - Diana Lantos

Italy - Silvia Landini  
• Miriam Grigoletto  
• Elena Caterina Di Noto

Mexico - Monica Ornelas

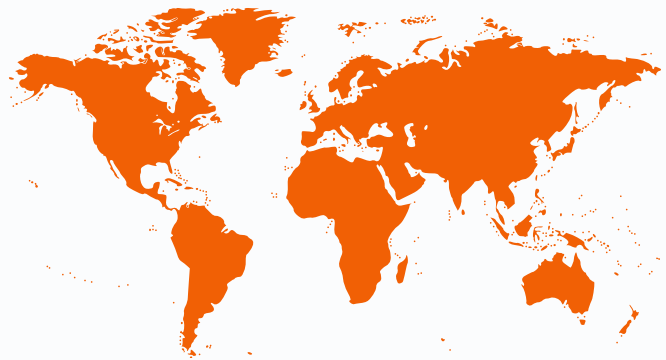
Poland - Szymon Dziomba



### INTERNATIONAL ASSOCIATION



The DYRK1A Syndrome International Association includes organizations & groups throughout the world who are working together to improve the quality of life for those affected by DYRK1A Syndrome. These groups include DYRK1A Syndrome US, the DYRK1A Community UK, the Spanish Syndrome Association DYRK1A, and smaller groups in Italy, Australia, Cyprus, Denmark, France, Poland, and Canada. An International Advisory Committee is planned to create more opportunities for our international community to work together on common goals that align with the strategic plan. We are working with international family representatives on the goal of our strategic plan to formalize relationships with international partners.



## Medical and Scientific Advisory Board

### **Sarah Glass** - Chair

- DYRK1A Parent
- Geneticist, Chief Development Officer, n-Loxem

### **Amy Clugston**

- DYRK1A Parent
- DYRK1A Syndrome US & International Association

### **Nadia Deba Giwani**

- DYRK1A Parent
- Leader in Healthcare Communications and Marketing

### **Dr. Bregje van Bon**, MD, PhD

- Clinical geneticist
- Radboud University Medical Center, Nijmegen, The Netherlands

### **Dr. Lucas Bronicki**, PhD, FACMG, DABMGG

- Assistant Professor, Clinical Genetics
- University of Ottawa, Canada

### **Dr. Amelie Piton**, PhD

- Neurogenetics and Translational medicine, Institute of genetics and Mol/Cell Biology
- University of Strasbourg, France

### **Dr. Craig McIntosh**, PhD

- Postdoctoral Fellow, Centre for Molecular Medicine and Innovative Therapeutics
- University of Murdoch, Australia



## **MEDICAL AND SCIENTIFIC ADVISORS**



The Medical and Scientific Advisory Board meets quarterly to discuss matters involving medical and scientific information related to DYRK1A Syndrome. They offer their expertise on scientific projects, publications, and the clinical needs of the patient population. Our advisors help assure that the organization's policies, research, marketing, communications, and publications meet the highest scientific standards. They are focused on our strategic plan goals to; establish a research funding mechanism, deepen the understanding of individuals affected by DYRK1A syndrome, establish a drug development toolkit and path to therapeutic, define centers of excellence, and educate and communicate to the community.

