AGO Alliance: Patient Family Conference 2024

Only 5% of rare diseases have an approved therapy[1]

In Europe, rare diseases are defined as diseases that affect fewer than 1 in 2,000 people. **But rare diseases are only rare in isolation; when grouped together, they affect 1 in ~25 people.[2]**

The rarity makes it challenging to research, diagnose and provide appropriate care, and **a lack of awareness can lead to a lack of understanding and feeling of isolation.**

> Only about 5% of rare diseases have an FDA-approved therapy.[1] There is an urgent need for more research, and earlier and more accurate diagnoses of and interventions for these disorders.
>
> - *Anne Pariser, Former Director of NIH NCATS Office of Rare Diseases Research*

AGO Alliance is dedicated to improving the lives of children and families impacted by the ultra-rare Argonaute Syndromes

AGO Alliance, formerly known as the AGO2 Association, was founded in 2021 and aims to:

1. Find and fund paths to treatment
2. Connect families
3. Raise awareness

Support us in helping our children maximise their potential.

Please click here to donate

> This is the second time I have participated in organising this meeting, and it is clearly the best one for Argonaute scientists. It truly inspires us to push boundaries and accelerate our research. We're here, dedicated to working for you, AGO families.
>
> *- Prof. Martin Simard, Centre de Recherche du CHU, Quebec*

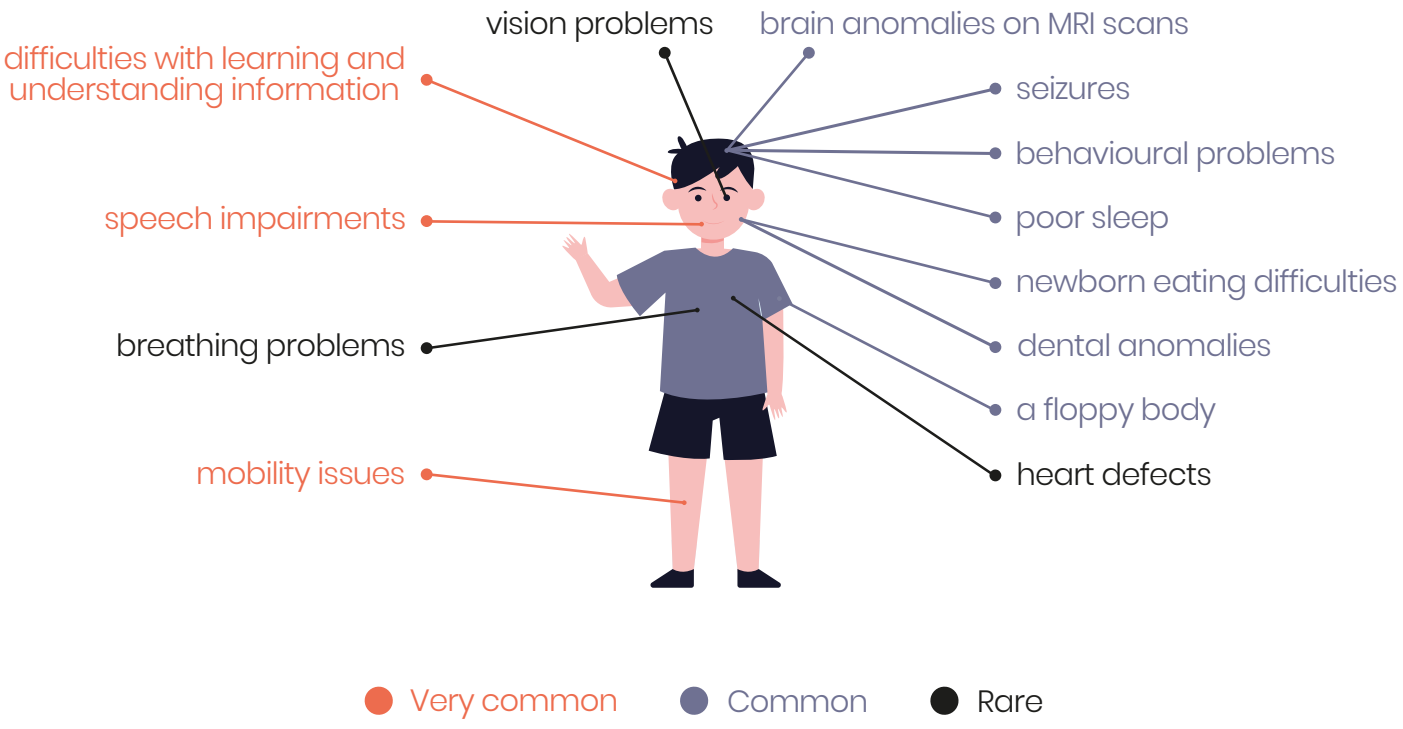
Worldwide, more than 110 patients have been diagnosed with Argonaute Syndromes

Changes in the Argonaute genes, AGO1 and AGO2, cause very rare conditions with similar symptoms that can be collectively referred to as **Argonaute Syndromes.**

Argonaute Syndromes are extremely rare. However, as these syndromes have only been recently discovered, **as many as 1 in 16,000 children may be affected.**[3]

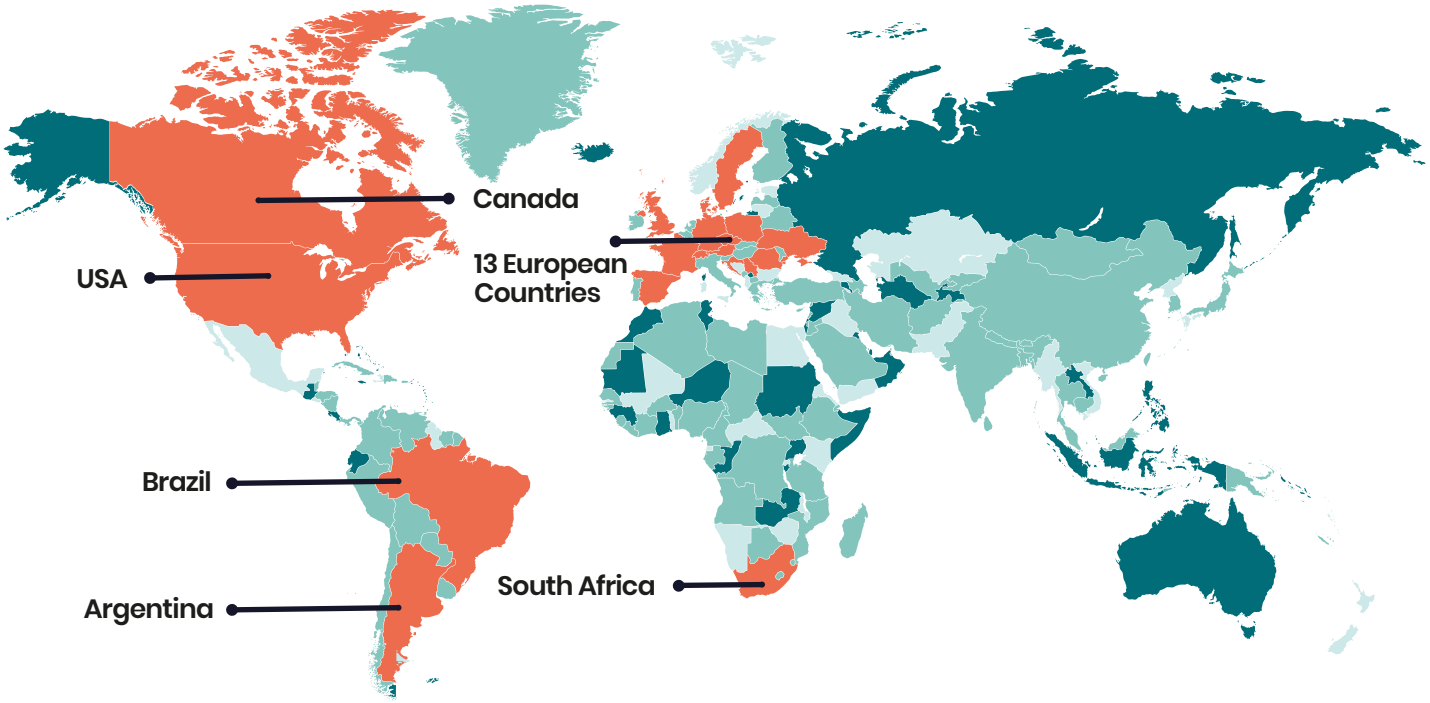
AGO1: AGO1-related syndrome. AGO2: Lessel-Kreienkamp or Leskres syndrome.

Known symptoms of Argonaute Syndromes[4,5]



of known patient families with Argonaute Syndromes worldwide are members of the AGO Alliance

of these families came together with researchers from across the world to share their experiences at AGO Alliance's Second Patient Family Conference



Attendees from Europe came from: Austria, Croatia, Denmark, France, Germany, Netherlands, Poland, Romania, Serbia, Spain, Sweden, Switzerland, United Kingdom.

"I learnt something new about my child's condition."

83% agreed or strongly agreed

100% of families and researchers would attend a similar meeting in the future.

This feedback is from families who attended virtually or in-person.

Inspiring. Informative. Hopeful.

giving context
encouraging
meaningful
impactful
emotional
moving
touching
listening
impressive
hopeful
inspiring
empathy
interesting
connection
important
motivating

Our patient families' hopes and dreams

Awareness

Improved public awareness of the disease so patients can be diagnosed sooner.

Support networks

Improved support for patient families through education and global networks, so they can share experiences that help them take better care of their children and know they are not alone.

Improved treatments

The development of treatments to improve the day-to-day lives of patients and their families.

Stronger understanding

Improved understanding of the natural history and disease progression of Argonaute Syndromes, empowering parents to know what to expect for the future of their child.

We felt very alone after the diagnosis, but **the association and science committee gave us hope**. This is what the conference represents for us – hope to understand the disease, to come up with a treatment, and have the **support of like-minded people** long-term. We are really grateful.

- Aldona, mum of Albert & AGO Alliance Science Officer

The best part of attending virtually was meeting other families and not feeling so alone. My daughter, Lillie, is not alone anymore. I want to do whatever I can until my last breath.

- Jessica, mum of Lillie

Researchers were able to network with patient families

100 researchers joined us in Copenhagen to attend the science conference, view our posters and meet patient families.

~40 researchers attended our parent panel to learn from the experiences of patient families.

From interacting with patients and their families at the conference, researchers learnt:

- The **variability** of the disease
- What **life is like** for patient families, in particular their worries and hopes for the future
- The **value of diagnosis**
- Novel aspects of the **natural history** of the disease
- The **impact** of scientific information and research in **empowering families**

100% agreed that meeting patient families gave beneficial and meaningful context to the science section of this conference.

It was my first time hearing directly from affected families. Everything they described was new.

- Assistant Prof. Gainetdinov, New York University

I have been super impressed, two years later after that first meeting with the families, at how far the field is already pushing to understand the mutations that have been identified and what they could be doing. In our own work where we look at infection models and basic biology, I genuinely think about the patients now and hope that maybe something we're learning can help them. And so, that interaction, I wish we had more of it.

- Prof. Amy H Buck, University of Edinburgh

Meeting with the families motivates us to work harder and faster. The field has moved forward tremendously in the past two years, but there is still so much to do. Every piece of information matters.

- Researcher who attended the Patient Family Conference 2024

Prof. Amélie Piton, Prof. Davor Lessel, Prof. Hans-Jürgen Kreienkamp and Dr. Olena Ielesicheva presented the latest knowledge about Argonaute Syndromes to the patient families, including first results from the NHS and what this means to families.

1. United States Government Accountability Office. 2024. Rare Disease Drugs. Available at: https://www.gao.gov/products/gao-25-106774. Accessed: February 2025.

2. Wakap SN et al. European Journal of Human Genetics. 2020;28:165–173.

3. López-Rivera et al. J. Brain. 2020;143:1099–1105.

4. Lessel D et al. Nature Communications. 2020; 11:5797.

5. Schalk A et al. Journal of Medical Genetics. 2022; 59:965-975.

AGO: Argonaute; **AGO1:** AGO1-related syndrome; **AGO2:** Lessel-Kreienkamp or Leskres syndrome; **CHU:** Center Hospitalier Universitaire de Québec; **FDA:** Food and Drug Administration; **NCATS:** The National Center for Advancing Translational Sciences; **NHS:** National Health Service; **NIH:** National Institute of Health.