AGO2 Association: Patient Family Conference 2022

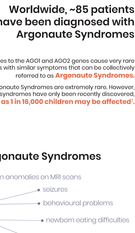
Only 10% of rare diseases have an approved therapy

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[1].**

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 10% of rare diseases have an FDA-approved therapy. There is an urgent need for more research, and earlier and more accurate diagnoses of and interventions for these disorders.
>
> *-Pariser, Director of NIH NCATS Office of Rare Diseases Research*

The AGO2 Association is dedicated to improving the lives of children and families impacted by Argonaute Syndromes



The AGO2 Association was founded in 2021 and aims to:

- Find and fund paths to treatment
- Connect families
- Raise awareness

Help us accelerate research, connect families and educate, to help our children maximise their potential.

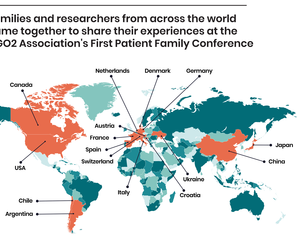
Please donate on our website!

Worldwide, ~85 patients have been diagnosed with Argonaute Syndromes

Changes to the AGO1 and AGO2 genes 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[2].**

Known symptoms of Argonaute Syndromes



~20% of patient families with Argonaute Syndrome came together with researchers in Regensburg and online

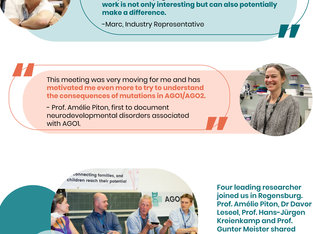
> I learnt something new about my child's condition
>
> 91% agreed or strongly agreed

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

This feedback is from families who attended in-person.

Families and researchers from across the world came together to share their experiences at the AGO2 Association's First Patient Family Conference

Educational. Informative. Emotional.



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 increased education and the development of wide networks, so they can share experiences and know they are not alone.

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

Leaders in rare diseases
Learn from other rare diseases and position AGO gene research as a role model for other rare diseases going forward.

> The relationships formed through the AGO association among families who truly 'understand' [how] to live with AGO provides support beyond measure. This conference has created **long-term connections & hope for our shared cause** which will positively impact children & entire families.
>
> ~Ashley

> Proud that my son could give a real example for what research is needed.
>
> ~Antja, Mum of Finn

Researchers were able to network with patient families

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

- The **variability** of the disease
- The **different approaches** taken by families in different countries
- **Novel aspects** of the natural history of the disease

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

> I could develop an understanding of the variability of the disease, I saw that many children have shown progress which suggests strongly that intervention can work.
>
> Lastly, it is always good to remind oneself that **our work is not only interesting but can also potentially make a difference.**
>
> ~Marc, Industry Representative

> This meeting was very moving for me and has **motivated me even more to try to understand the consequences of mutations in AGO1/AGO2.**
>
> ~ Prof. Amélie Piton, first to document neurodevelopmental disorders associated with AGO1.

Four leading researcher joined us in Regensburg. Prof. Amélie Piton, Dr Davor Lessel, Prof. Hans-Jürgen Kreienkamp and Prof. Gunter Meister shared their research and learnt from the experiences of patient families.

1. Wakap SN et al. European Journal of Human Genetics. 2020;28:165–173.
2. López-Rivera et al. J. Brain. 2020;143:099–1105.