AGO Alliance: Patient Family Conference 2025

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

In Europe, rare diseases are defined as diseases that affect 1 in 2,000 people or fewer.[2] **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 feelings of isolation**.

> Coming together is a beginning; keeping together is progress; working together is success.
>
> *- Henry Ford, American Industrialist and Businessman*

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

AGO Alliance was founded in 2021 and aims to:

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

The AGO Alliance Patient Family Conference 2025 was made possible through collaboration with our partners: Svoboda lab at IMG, AGO Alliance Poland and Sindromes AGO.

To hear more from the families and researchers who attended the conference, please click here.

> In this 3rd conference, the mood has shifted. In the past there was hesitancy and the need to cover more basics. Now, I see how the field is progressing and we are taking steps towards a treatment.
>
> *- Nora, President of the AGO Alliance*

Support us in helping our children maximise their potential.

Please click here to donate.

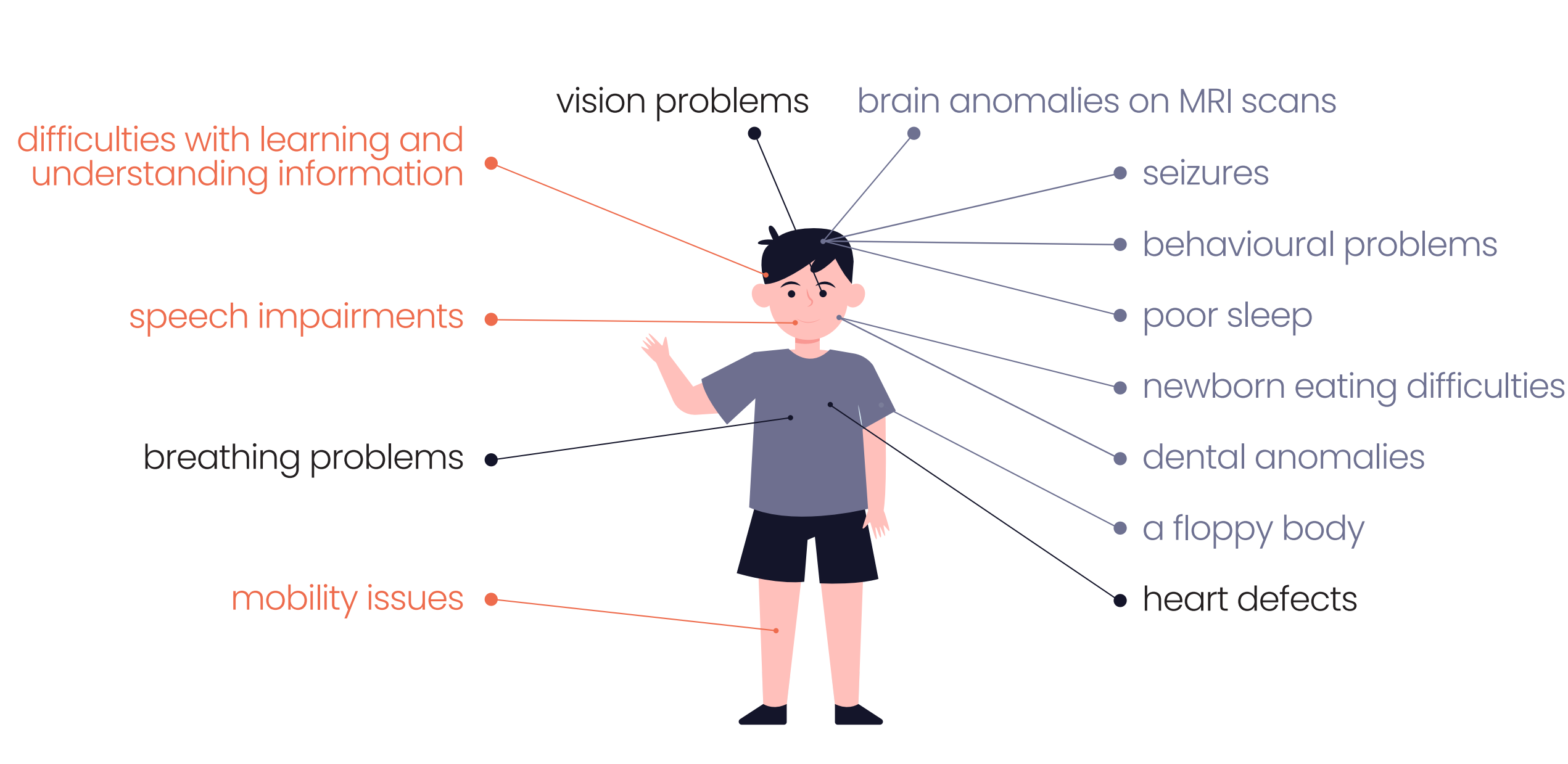
Worldwide, >180 patients have been diagnosed with AGO Syndromes.

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

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

AGO1: AGO1-related syndrome (also known as "NEDLBAS").
AGO2: Lessel-Kreienkamp or Leskres syndrome.

Known symptoms of AGO Syndromes:[3,4]

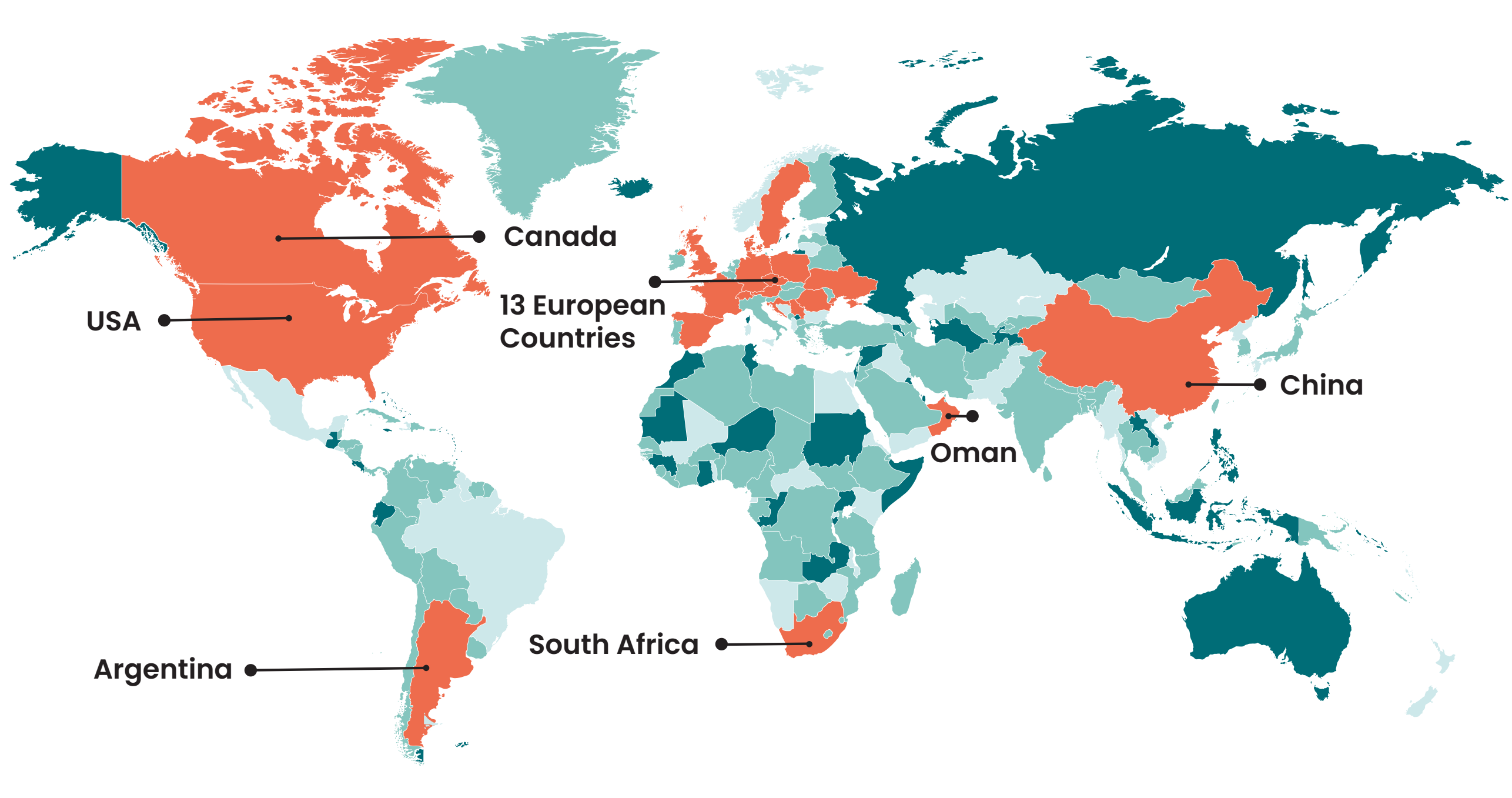


> Our basic research can have real impact for the understanding of the different mutations.
>
> *- Researcher and attendee of the AGO Alliance Patient Family Conference 2025*

Who attended the Patient Family Conference 2025?

~90% **of known patient families with AGO Syndromes worldwide are members of the AGO Alliance.**

~25% **of these families came together with researchers from across the world to share their experiences at AGO Alliance's Third Patient Family Conference.**



Attendees from Europe came from: China, Czechia, Denmark, Estonia, France, Germany, Netherlands, Poland, Romania, Slovakia, Spain, Sweden, Switzerland, United Kingdom.

> [The best part about meeting other families at the conference was] the sense of understanding and brotherhood.
>
> *- Lukasz, father of Alex*

> **I learnt something new about my child's condition.**
>
> 80% agreed or strongly agreed

94% of families and researchers would attend a similar meeting in the future.

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

What 3 words would you use to describe the family workshop?

heartfelt
friendly
life changing
real
supportive
connected
brave
valuable
moving
uplifting
honest
touching
hopeful
inspiring
comforting
interesting
personal
emotional
empowering
touching
challenges
tailored
compassion

Our patient families' hopes and dreams:

Awareness and understanding
To discover more patients living with AGO Syndromes and build a larger pool of data to better understand these conditions.

Clear guidance and support
Improved guidance for patient families to help children with AGO Syndromes to fulfil their potential.

Improved treatments
To develop treatments that can help to alleviate the more serious symptoms associated with AGO Syndromes.

> I am grateful for the opportunity to meet such giants as Professor Ambros, Professor Lessel and Professor Kreienkamp. I am in awe of their dedication and their great mission to help children live happy lives.
>
> *- Attendee of the AGO Alliance Patient Family Conference 2025*

> Everyone was so supportive and understanding. I felt comfortable with everyone immediately, as if we've been friends our whole lives.
>
> *- Attendee of the AGO Alliance Patient Family Conference 2025*

> Each conference, each research paper, each connection between families and scientists brings us one step closer to answers.
>
> *- Kerri, mother of Wyatt*

Researchers were able to network with patient families:

~170 researchers joined us in Prague to attend the science conference, view our posters and meet patient families.

~50 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 about:

- How **widespread** AGO Syndromes are globally
- The short- and long-term **needs** of patients, to improve their **quality of life**
- Emerging **treatments**
- The **varied** nature of AGO Syndromes
- The significant **challenges** that arise as **patients grow up**
- The **resilience** and **postive mindset of patient families** managing the impact of these conditions

75% of researchers established new, or strengthened existing, collaborations during the conference.

> [Interacting with patient families has motivated me] to introduce the disease in my teaching activities.
>
> *- Prof. Ronald van Rij, Researcher*

> The conference enabled me to meet in-person with scientists with whom we are collaborating on translational efforts, and that resulted in us **sharpening our focus and organisation priorities**. Such in-person brainstorming meetings are almost always more effective than virtual meetings or email.
>
> *- Prof Victor Ambros, Researcher and winner of the 2024 Nobel Prize in Physiology and Medicine*

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

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1. Braga LAM et al. Therapeutic Advances in Rare Disease. 2022;3:26330040221100840.
2. Wakap SN et al. European Journal of Human Genetics. 2020;28:165–173.
3. Lessel D et al. Nature Communications. 2020;11:5797.
4. Schalk A et al. Journal of Medical Genetics. 2022;59:965–975.

AGO: Argonaute; **AGO1:** AGO1-related syndrome;
AGO2: Lessel-Kreienkamp or Leskres syndrome;
NHS: National Health Service.