ENDO ERN General Assembly
Crowne Plaza Hotel, Athens

MTG3 HCP meeting

Tuesday 21 April 2026

Pietro Maffei, Felix Reschke, Valeria Corradin, Bernd Rosenbichler, Nolwen le Floch

MTG3 chairs

Funded by
the European Union

Contact

The representatives for MTG3 Genetic Disorders of Glucose & Insulin Homeostasis are:

Pietro Maffei Adult and Paediatric chair

Azienda Ospedaliera di Padova (AOP) (Padova, Italy)

Pietro Maffei

Felix Reschke Paediatric chair

Hannoversche Kinderheilanstalt, Diabetes Centre for Children and Adolescents (Hannover, Germany)

Felix Reschke

Valeria Corradin ePAG representative

AILIP (Ass.ne Italiana Lipodistrofia) (Dossobuono, Italy)

Valeria Corradin

Nolwen LeFloch ePAG representative

Association du syndrome de Wolfram (ASW) (Grand-Champ, France)

Nolwen LeFloch

Bernd Rosenbichler ePAG representative

Alström Syndrom e.V. (Unterföhring, Germany)

Bernd Rosenbichler

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Thank you for attending this meeting !

WP1
Management & Coordination

SAVE THE DATE: European Congress of Endocrinology (ECE) 2026

This event will be from May 9th to May 12th, 2026

The European Congress of Endocrinology (ECE) will take place in **Prague, Czech Republic, from 9 – 12 May 2026**.

Save the Date! Further details will be announced in due course.

Event summary

Event	European Congress of Endocrinology (ECE) 2026
Date	May 9th - May 12th, 2026
Location	Prague, Czechia
Organisation	European Society of Endocrinology (ECE)
Website	Visit event website
WP	WP6 Education and Training
	Add this event to your calendar

SAVE THE DATE: European Society of Paediatric Endocrinology (ESPE) 2026

This event will be from September 8th to September 10th, 2026

Save the date for the 64th ESPE 8-10 September, 2026 to be held in Marseilles, France.

More information will be available closer to the event.

Event summary

Event	European Society of Paediatric Endocrinology (ESPE) 2026
Date	September 8th - September 10th, 2026
Location	Marseilles, France
Organisation	European Society for Paediatric Endocrinology
	Add this event to your calendar

WP1
Management & Coordination

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SNOW FOUNDATION FOR WOLFRAM SYNDROME RESEARCH
A GLOBAL VOICE FOR RARE DISEASE

11th International Wolfram Syndrome Symposium
Berkshire, England 3rd-4th June 2026

WOLFRAM SYNDROME UK
Inform, Support, CURE
together with
SNOW FOUNDATION FOR WOLFRAM SYNDROME RESEARCH
A GLOBAL VOICE FOR RARE DISEASE

WP1
Management & Coordination

ALSTRÖM SYNDROME INTERNATIONAL

About Support The Latest

← Back to All Events

11th Alström Syndrome International Family Conference & Scientific Symposium

Thursday, May 21, 2026, 6:00 PM –
Monday, May 25, 2026, 10:00 AM

Someone (Florida) gave
$111 1 month ago

WP2
Communication & Dissemination

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MTG 3 tasks

Next special issue in Endocrine Oncology
1 paper for each MTG
We will provide a temptative Title and possible Authors….
Work in progress and open collaboration

Emerging therapies in rare diabetes and insulin resistance: need for structured exchange

- Increasing availability of **new metabolic therapies:**

  GLP-1 receptor agonists / metreleptin / setmelanotide / other off-label / mechanism-based approaches/ etc. [1,2,3]

- **Small but growing evidence base** for selected rare forms of:

  insulin resistance / lipodystrophy/ rare / monogenic diabetes

- In clinical practice, these therapies are likely already used **heterogeneously:**

  different indications/ different timing/ dosing/ different access and reimbursement pathways

Current practice, access and experience may differ substantially between centres and countries.

Ref: (1) Kühnen P, et. Al. Proopiomelanocortin Deficiency Treated with a Melanocortin-4 Receptor Agonist. *N Engl J Med*. 2016;375(3):240-246. doi:10.1056/NEJMoa1512693, (2)Brown RJ, et al. The Diagnosis and Management of Lipodystrophy Syndromes: A Multi-Society Practice Guideline. J Clin Endocrinol Metab. 2016;101(12):4500-4511. doi:10.1210/jc.2016-2466
(3) Jagomäe T, et al. Early Intervention and Lifelong Treatment with GLP1 Receptor Agonist Liraglutide in a Wolfram Syndrome ….*Cells*. 2021;10(11):3193.

MTG3 Approach: Mapping Practice & Next Steps

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Key questions across MTG3:

- Who is using which therapies — and in which indications?
- At what stage of disease / treatment pathway?
- What are the perceived benefits and limitations?
- What are the main barriers?
- access & reimbursement
- national differences
- off-label use
- Where are the key evidence gaps?
- Is there potential for shared approaches or expert alignment?

Proposed approach

1. **MTG3 survey**
   → capture current use, indications, experience, access & unmet needs
2. **2. Data overview**
   → identify commonalities, differences, and priority areas
3. **3. Multi-stakeholder discussion**
   → clinicians, researchers, patients (+ optional partners)
   → focused on evidence, access, risks & benefits

Perspective: → Towards shared understanding, potential expert alignment, and future collaborative activities

Endocrine Oncology Special Issue

Call for proposals:

- Endo-ERN plans a special issue on Endocrine Oncology
- Aim:
- highlight rare endocrine cancers
- explore long-term effects of treatments in rare conditions
- Limited number of contributions (~10 papers across MTGs)

Proposed MTG3 topic:

- **Monogenic insulin resistance as a model for insulin/IGF signalling and endocrine tumor biology**
- Rare disorders with **chronic hyperinsulinaemia,** Activation of key pathways (insulin / IGF axis/ etc.)
- Potential **translational link** between rare metabolic disease & endocrine oncology
- 👉 Opportunity to position MTG3 within this field

Discussion:

Do we agree to submit this as an MTG3 proposal? Who would be interested in contributing / co-authoring? Are there alternative topic suggestions from the group?

Early Stage Type 1 Diabetes & ENDO-ERN

Background:

- Growing focus on **Early Stage T1D (stage 1–2)**: screening, prediction, disease-modifying approaches
- Increasing need for **European collaboration**:
- Cross-border trials & cohorts, Registries / data infrastructure, Structured **transition (pediatric → adult care)**
- Existing initiatives: **INNODIA, SWEET, PRE-T1D, Breakthrough T1D**

Why discuss within ENDO-ERN?:

- Potential interface with: **Rare / monogenic diabetes /** Complex, early-stage disease trajectories
- Possible ENDO-ERN role: **Bridging / coordination function** (not duplication) / Focus on **transition & care pathways**
- Existing contacts with key initiators (e.g. Chantal Mathieu)

Key questions for MTG3:

- Does **Early Stage T1D** fit within the **ENDO-ERN mandate**?
- Can it be considered under a **“rare / complex condition” perspective**?
- Where is the **added value vs. existing networks**?
- Should ENDO-ERN take a **pilot / exploratory role**?

WP3 Evaluation

- Ensure that HCP members are entering data into e-REC for the continuous monitoring process
- Please feedback any challenges you are having

e-REC

e-REC is an electronic reporting (e-reporting) programme that captures new clinical encounters. This allows for a better understanding of the occurrence of the rare endocrine and bone conditions covered by European Reference Networks (ERN) such as Endo-ERN and ERN BOND.

Compared to the Core Registry, where more detailed and longitudinal data is collected, e-REC only captures the number of new cases, suspected or confirmed, seen monthly at the participating expert centres. Because e-REC does not collect any personal data, there is no informed consent needed from the patient. It is a clinician-only platform designed to support the ERN's in their continuous monitoring program.

e-REC is open to all centres that look after people with rare endocrine and bone conditions. It allows continuous reporting of core indicators of activity and enables clinical networks to objectively map the conditions and related activities. e-REC assists Endo-ERN and ERN BOND and its members in the continuous monitoring programme.

https://eurreb.eu/registries/e-rec

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WP3 Evaluation

MTG Stats slides 2025

Funded by the European Union

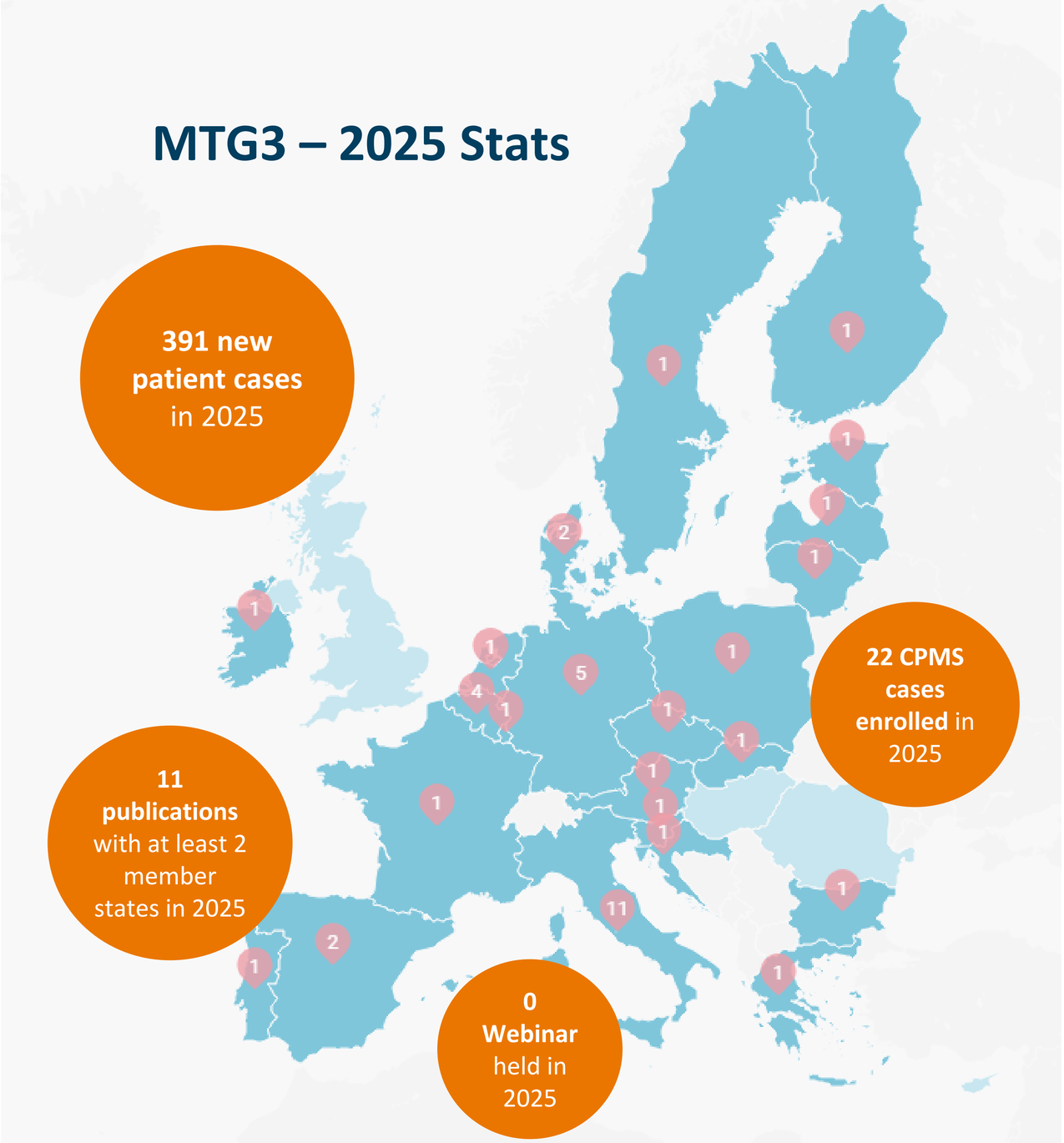
MTG3 – 2025 Stats

391 new patient cases in 2025

22 CPMS cases enrolled in 2025

11 publications with at least 2 member states in 2025

0 Webinar held in 2025



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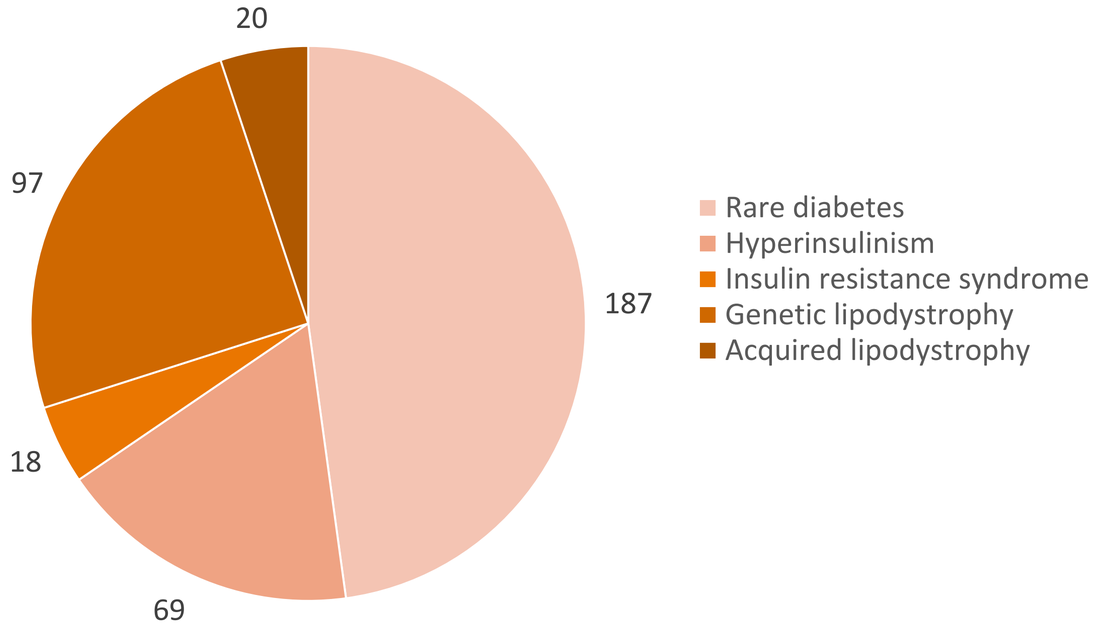
No. of new patients per MTG3 sub thematic area of new patients



MTG3 HCPS - 2025 Stats

42 HCPS in MTG2 formally

39 HCPs reporting in **e-REC** registry

28 HCPs reported they attended at least 1 **MTG meeting**

21 HCPs participated in at least 1 **CPMS panel**

- 30 HCPs have **different** EC & e-REC totals
- 8 HCPs **do not have** Endo-ERN logo/url at their website
- 12 HCPs who did not measure patient satisfaction – this is based on your HCP **NOT your department** usually

WP6 Education & Training

MTG3 improvement is needed in 2026 !

Overview of Webinars for MTG3

Showing 12 entries

Theme / Title	Date	Time CET	Speaker(s)	Register	View	CPMS
L'ipoglicemia in età pediatrica: dalle raccomandazioni internazionali alla pratica clinica (Childhood Hypoglycemia)	2026-03-05	16:00 – 17:00 hr				
SWEET-the role of a worldwide registry to investigate rare diabetes	2024-10-09	16:00 – 17:00 hr				
The mysterious links among cilia, centrosomes and insulin resistance: What have we learned from human genetics?	2024-04-02	15:00 – 16:00 hr				
Endo-ERN webinar HNF1B diabetes	2023-12-13	15:30 – 16:30 hr	Jarno Kettunen, Helka Parviainen, Tiinamaija Tuomi and Päivi Miettinen			

WP6 Education & Training

2026 MTG3 temptative plans of webinars:

- **Alstrom syndrome ePAGs**: AS-Germany, AS-UK, AS-Italy, AS-International
- **Wolfram syndrome ePAGS:** WS Italy, WS UK, WS France, WSC USA
- **Lipodystrophy e PAGS**
- **Congenital Hyperinsulinism in Europe**: Advances and care disparities (leaded by Henrik Christesen, Odense, DK)
- **Wolfram syndrome** webinars: T. Barrett (Birmingham); F. Urano (Washington)
- **Discovery of new medication by rare disease pathways** (V Marion, Strasbourg)
- **Paediatric rare diabetes**

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Alström Syndrom e.V. -

Positive Impressions from Germany

Funded by the European Union

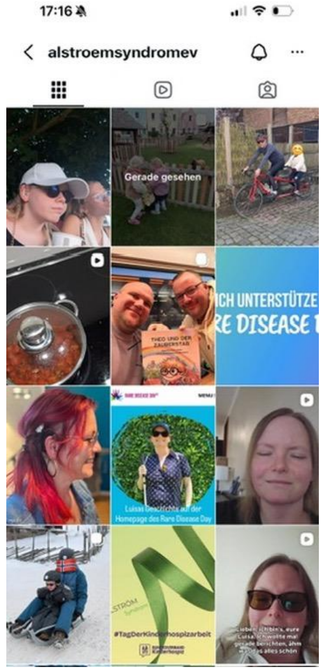
Alström Syndrom Germany e.V.

- Founded in 2022
- Currently 180 members
- 25 members with Alström syndrome
- Covering the German-speaking region of Europe
- Funded through donations
- Board member of Alström Europe (ASEU)

Projects

- **Alström vs. BBS:** We cooperate with University of Essen, which is currently establishing a center of expertise for Bardet-Biedl syndrome. A first joint project involved a detailed assessment of our Alström patients, generating a database that was compared with the BBS cohort, delivering a unique, new perspective.
- **N-of-1 Study:** Together with Charité Berlin, the University of Göttingen, and other partners, we have developed a new study format for obesity based on an N-of-1 approach, comparing the effects of different drugs such as Wegovy, Imcivree, and others.
- **Patient Registry Project:** Supported by the Berlin Institute of Health at Charité (BIH), the University of Frankfurt, and other partners, we are developing an Alström patient registry. This is one of the first rare disease projects to serve as a pilot for the German registry standard defined within NARSE, the National Registry for Rare Disease
- **Application** for EP PerMed JTC2026 “CARMEN2026”

Encouragement!



- We place strong emphasis on positive activities and constructive communication.
- Despite facing severe conditions, including visual impairment or blindness, our members lead active lives — they ski, dance, cycle, play the piano, practice judo, and much more.
- This perspective is also a central focus of our social media, which we continuously refine and improve.
- Healthy nutrition and everything that supports a balanced life(style) is another major area of focus.

Enjoying live with Alström Syndrome

With the first warm days of spring, the tandem bike comes back to life. For **Helena**, every ride brings joy: fresh air, movement, speed, and rich sensory experiences. Recently, we spent **3 hours and 20 minutes** on the road together – a milestone that means so much to us.

It shows that despite **Alström syndrome**, life can still be full of happiness, activity, and meaningful achievements.

Our focus is on **healthy nutrition, movement, and celebrating every success along the way**

For Amalia, the world is defined by sound rather than sight.

Even though Alström Syndrome has caused her to lose most of her vision, she refused to let that stop her from learning the piano.

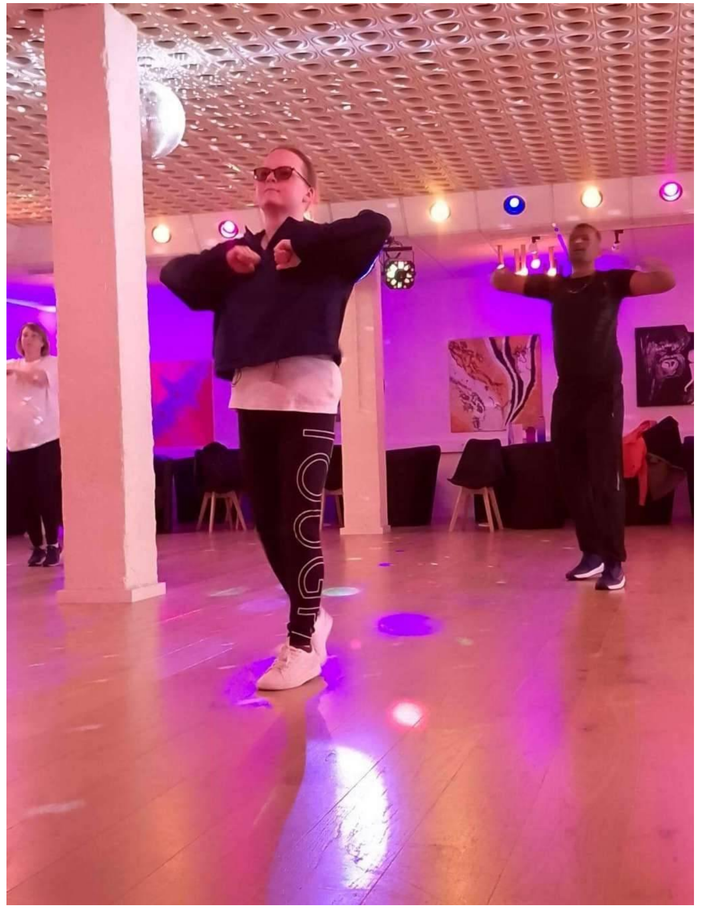
Learning to play required a lot of patience and practice. Since she cannot read traditional sheet music easily, she relies on her hearing and her memory to find the right notes. For Amalia, the piano is more than just an instrument; **it is a way to express herself and find peace.**

Music has truly brightened her life. When she sits down to play, her disability doesn't matter anymore. The melodies give her strength and bring joy to her every day, proving that talent and passion can overcome any obstacle.

Dance is Luisa’s passion. Her biggest dream: to become a choreographer.

But for now, dancing remains a hobby where she can express her emotions.

Especially with Zumba, she can forget everything around her and, alongside feelings of happiness, release negative emotions as well.

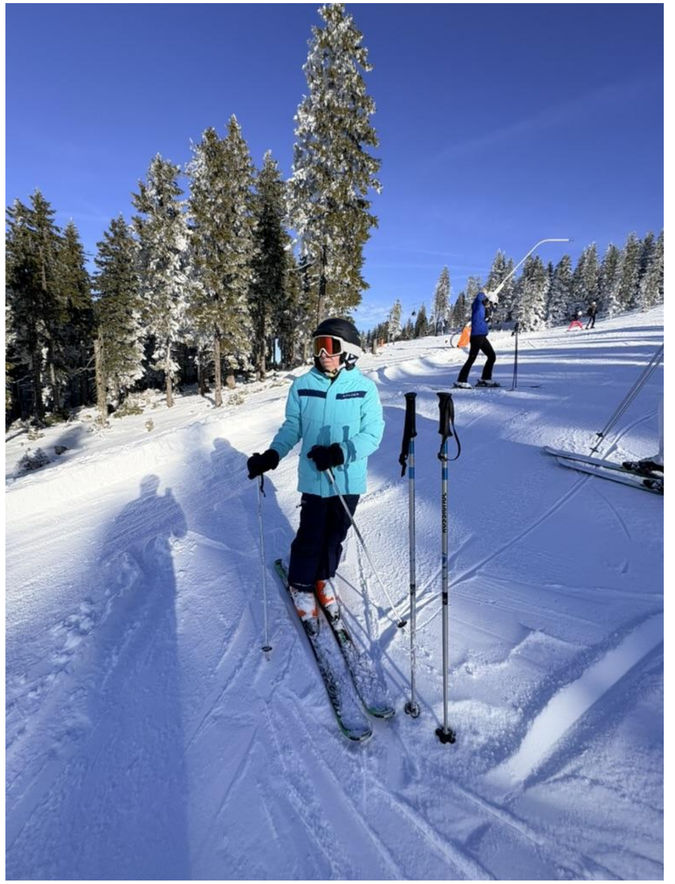


Ben loves to ski. Guided only by voice, he takes on even the steepest slopes with remarkable confidence.

Despite being almost completely blind, he maintains control, reads the terrain through instruction and instinct, and moves with a natural sense of rhythm.

He can spend entire days on the mountain, pushing his limits, until his body finally tells him it’s time to stop.

For him, skiing is more than an activity. It’s freedom, independence, and a way of experiencing the world on his own terms.



Louis and Ben love Judo. Louis fights on his knees – and wins.

NOLWEN LE FLOCH

Video from Nolwen Le Floch

Video from Valeria Corradin

AILIP
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