FOCUS

No. 05 - November 2026

Les extra vaillants

À LA UNE

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19 Nov. photo for the mural

26 Nov. JIMR testimony

5 Dec. March, Paris

28 Feb. mural unveiled

Click the underlined words or scan the QR codes to open the links

Editorial

Since February 2026, MYT1L syndrome has its own national diagnosis and care protocol, the PNDS. Doctors, carers, therapists and families finally share a common reference, and it is the focus of this issue (page 3).

This document does not cure: there is still no curative treatment. But it sets one shared course of action for diagnosis and follow-up that anyone can read, and its summary helps your GP draw up the care plan. My advice: read it, then take the summary to your next appointment.

We owe this progress to the doctors and researchers who move forward alongside us, and to you: every questionnaire completed, every day spent in Rouen, every test result shared counts. This same partnership made it possible this year to publish the first detailed description of eating behaviour in the syndrome (page 7).

It carries on into your daily life: the families' area now brings together the MDPH disability guide, the turning-18 guide and many other tools (pages 4 and 5), and the events calendar picks out useful dates for you (page 12). They answer the questions you ask us every week. They are free, and they are yours.

We have reached a turning point. For five years, a handful of volunteers has carried all of this. Our 2026-2030 project is ambitious, and it can only live if it is shared. I remain fully committed, and I hope that over time responsibilities will be better shared: we need new energy, in line with what each person can give. An hour counts, a skill counts, a membership counts.

Our next general meetings will be the time to talk about it together: you will receive your notice by e-mail.

Valérie Salomone, President

4 things to do before the end of the year

1. I take part in the JIMR

My photo for the mural before **19 November**, and if I wish, my testimony for the Alliance before **26 November**. I take my photo (p.8)

2. I create my account

The families' area opens with your e-mail and a password. Two minutes, just once. Go (p.4)

3. I walk

On **Saturday 5 December** in Paris, behind the « MYT1L syndrome » banner, for the Téléthon's 40th anniversary. I sign up (p.8)

4. I get involved

I join, I offer a little time. After the May meeting, a new notice will invite me to vote on our statutes and elect the board. Act with us (p.13)

Focus on the PNDS

A national diagnosis and care protocol, or PNDS, describes the diagnostic and therapeutic management of a rare disease and the care pathway. The one for MYT1L syndrome has been online since 5 February 2026. It is for everyone around a person with the syndrome: doctors, carers, therapists and families.

PNDS

What it brings

A shared reference

The same benchmark everywhere in France, for diagnosis, follow-up and referral.

Support for teams

Help for professionals meeting the syndrome for the first time.

A clearer pathway

For families, a document to read and share, to know what to ask for and whom to ask.

Three documents, freely available

The summary

Written for the GP. It helps them draw up the care plan.

The full PNDS

The reference document for care teams, therapists and families.

The rationale

The scientific data on which the protocol is based.

What you can do with it

- **Read it** at your own pace, then note your questions for the next appointment.
- **Give the summary to your GP.** It helps them fill in the care plan and the application for long-term condition status (ALD). If the French health insurance accepts it, care related to the syndrome is covered at 100% of the Social Security rate.
- **Share it** with the professionals who support your relative: therapists, care-home staff, school.
- **Quote the Orphanet code 647799** in your exchanges and applications.

Good to know

Drawn up by the Rouen reference centre following the method proposed by the HAS (French National Authority for Health), it has not been validated by the HAS. It will evolve as knowledge grows. There is currently no specific curative treatment: medical and paramedical care targets each person's needs. The PNDS does not replace the advice of your medical team.

Thank you

To Dr Juliette Coursimault (Rouen University Hospital, Inserm U1245), lead author, and to the working group: Béatrice Dubern, Christine Poitou-Bernert, Anne-Marie Guerrot, François Lecoquierre, Lucie Kropfeld, Stéphane Rondeau, Anthony Soter, Soizic Maingant, Flore Schnitzler, Aurélie Chupin, Marie Gentin and Laura Lechevalier.

Your « Families Only » area

A protected space on the association's website, open since July 2026, for French and French-speaking families. You will find all the up-to-date documents there, whenever you need them, without having to ask us.

How do I get in? Two minutes, just once

1. On the website's home page, click the pink **« Réservé Familles »** (Families Only) button.
2. Click **« S'inscrire »** (Sign up): your e-mail and a password of your choice, or your Google account.
3. **We approve your request**: this keeps the area strictly for families. Your access is then permanent, on computer, tablet or phone.

Families' area

Your account replaces the old access code: one login opens every page, including future ones. A problem? extravaillants@gmail.com

What you will find there

MYT1L families welcome booklet

July 2026. The first landmark after diagnosis.

MDPH guide for MYT1L syndrome

The French disability file explained section by section, with template letters.

Turning 18

Legal protection, assets and inheritance, easy-read versions.

Understanding research

June 2026. Studies on MYT1L syndrome, in simple words.

Family survey results

May 2026. What 30 participants told us about their needs.

Contacts to bring family life together

Discussion groups, testimonies, ideas: the area will live thanks to you.

A 5-minute mission: « Let's get to know each other »

Carine and Éric, volunteers with the association, are building family life with you. Their questionnaire is waiting for you in your area. Your answers will tell them what would help you most, close to home.

Living outside France? We are also expanding our resources in several languages, open to all on the website. The Facebook group **« La Bulle des Extra-Vaillants MYT1L »**, run by families, remains your place for parents to talk to each other.

Four tools for your daily life

MDPH guide: fill in and understand your file

More than 40 families have asked us about it since April 2026. (The MDPH is the French disability office.) It comes in four tabs:

- the **full 160-page guide**, with template letters to copy;
- a **section-by-section explanation** of the two additional forms, rare diseases and neurodevelopmental disorders;
- two **easy-read versions**, for parents and for the person concerned.

Open the MDPH guide

Turning 18: legal protection, assets, inheritance

At 18, parental authority ends. For a young person who cannot express their wishes, it is best to prepare early. Two detailed parts (legal protection, assets and inheritance) and three easy-read versions: for the young person, for parents, for the adult concerned.

From 17. The application can be filed during the last year as a minor.

6 months before 20. The AAH disability allowance is not backdated: apply early.

Already 18? Nothing is lost: a whole chapter is there for you.

Open the turning-18 guide

Impacts page: 27 features explained

An interactive page open to all: 27 features ranked by frequency, with a description and guidance on what to do. Filters by area and language. To show at school, or to a new professional.

Discover the page

Easy-read cards: understanding my difference

« I understand my difference »: cards to handle, to better understand daily life, spot strengths and find ways to get help. Available on request, commercial use prohibited.

Ask for them

And the PNDS? It is not just for doctors: carers, therapists and families can read and use it. It has its own page in this issue (page 3).

These support documents explain the procedures. They replace neither your support nor the advice of a legal professional, and practice varies from one MDPH and court to another. A bill on the legal protection of adults is under review: check before any binding step.

Where does research stand?

Call to families: brain MRI and EEG

Rouen University Hospital and Heidelberg hospital are collecting brain MRIs and EEGs from as many people with MYT1L syndrome as possible, for a centralised, double-blind review. The aim: to better understand the syndrome and offer appropriate neurological follow-up.

No new test needed. Talk to your geneticist or paediatric neurologist, and ask them to send existing results to Dr Juliette Coursimault: j.coursimault@chu-rouen.fr. Every file counts.

Language, cognition, behaviour

Ongoing, Rouen University Hospital. 22 Vaillants took part from March to May 2025. The study aims at reliable benchmarks and therapy protocols specific to the syndrome, for children, teenagers and adults.

Eating behaviour

Published in 2026. First detailed description, in 21 people, with the PRADORT centre. The results are on page 7.

Humanities and social sciences

Priority of the 2026-2030 project. Natural history: how does the syndrome evolve in adolescence and adulthood?

Episignatures

Rouen University Hospital. The association supports research on episignatures of the MYT1L gene.

GenIDA

Open to families. You enter your relative's data yourself, in French, English, Italian or Spanish, alongside the Rouen study. genida.unistra.fr

International cooperation

Heidelberg and St. Louis. The teams share their work, from neuron biology to the clinic. Meeting in St. Louis on 13 and 14 November.

Stay informed about research

To receive our research news, FOCUS magazine and key dates by e-mail, we need your consent (GDPR). Simply reply **« YES »** to our last e-mail, or write **« YES »** to **extravaillants@gmail.com** with your first and last name. Your details are never shared and you can unsubscribe at any time. Our website's **families' area** also gives an update on research.

Therapies: cautious information

Antisense oligonucleotides (ASOs) are being studied in the United States: the idea would be to help the working copy of the gene produce more MYT1L protein. **This ASO does not exist yet**, and the work will take several years. There is currently no specific curative treatment for MYT1L syndrome. Our « Understanding research » guide, in the families' area, gives an overview.

Update as of November 2026. This information does not replace the advice of your medical team.

Eating behaviour: the study is published

Announced in FOCUS no. 4, the study was published in 2026 in Orphanet Journal of Rare Diseases. For the first time, it describes in detail the eating behaviour of 21 French-speaking people, 14 children and 7 adults.

Work by Dr Juliette Coursimault (Department of Genetics, Rouen University Hospital), Prof. Christine Poitou and Prof. Béatrice Dubern (PRADORT reference centre, AP-HP). Thank you to all the families who completed the questionnaires.

16 out of 21
reach or exceed the threshold for marked hyperphagia (score ≥ 19)

5.1 / 10
average hunger, 5.3 on waking: hunger present throughout the day

5 out of 8
people assessed (7 adults, 1 teenager) have a score suggestive of food addiction, with no diagnostic value

37%
of parents describe a quality of life severely or extremely affected by eating behaviour

What the study shows

- Moderate but persistent hyperphagia, weaker satiety response and very fast meals, with chewing that is sometimes difficult.
- First signs of interest in food around age 3½; preoccupation that is most often stable over time.
- Food impulsivity less frequent than in other rare obesities (5 children out of 14).
- Half of the people actively seek food; no night-time food seeking was reported.
- Hormone tests without any notable abnormality.

What it does not say

Only people with overweight, obesity or eating behaviour disturbances took part. **So these figures do not describe all people with the syndrome.**

The group is small, includes few adults, and the answers mainly come from relatives. Results for adults remain to be confirmed.

Suggestions from the research team

- **Inform families in advance** that hunger and food preoccupation may persist.
- **Structure meals**: meal routines, adapting the environment.
- **Watch for oral-motor difficulties**: speed of meals, chewing, large pieces swallowed.
- **Adapt follow-up to age**, within early, multidisciplinary, family-centred care, with preventive measures from the genetic diagnosis: nutritional advice, routines, adapted physical activity.

These suggestions are general. For your relative, talk to their doctor or reference centre. A summary in English, written by the association, is available on our website: Research page. **Only the reference publication is authoritative.**

Read the original article (open access): Coursimault et al., Orphanet Journal of Rare Diseases, 2026.

Article

JIMR 2027: your photo, your testimony

The AnDDI-Rares network wants to gather hundreds of portraits to create a giant photo mural. Let's be many to represent the Vaillants!

Before 19 November 2026

1. Click the participation link, from a phone or a computer.
2. Strike a pose. Happy with the photo? Click « J'aime » (I like it). If not, « Recommencer » (Start again). Three photos maximum.
3. Your portrait joins the big national mural.

Take my photo

Who can take part? Patients, families, associations, professionals, researchers. **What next?** Unveiling on Sunday 28 February 2027, in several network centres and online. The original mural will be exhibited at the Imagine Institute in Paris.

Your testimony for the French Rare Diseases Alliance, before 26 November

For JIMR 2027, dedicated to equity, the Alliance is collecting testimonies: « Diagnose early, treat everywhere ». Your choice: the diagnostic odyssey, the care pathway or daily life (disability file, school, work, family), and what made a difference for you. Send a few lines, a good-quality photo and the completed publication consent form to jimr@maladiesrares.org. They may be published during the JIMR month.

Rare Diseases March



Saturday 5 December 2026, Paris

A festive 6 km walk for the Téléthon's 40th anniversary. Let's carry the « MYT1L syndrome » banner together!

- **1.30 pm**: meet near the bandstand, Luxembourg Gardens.
- **Dress code**: blue, green and pink, hats and ribbons welcome.
- **Travel**: the Alliance reimburses the train ticket of one person living outside Paris, per association. Let us know before booking.
- **Téléthon TV set**: in the evening in Saint-Denis, limited places for walkers.

Sign up with us, extravaillants@gmail.com, or on the Alliance form.

Our 2026-2030 project

Together, going further for the Vaillants. Our goal: that every family finds the right answers faster, close to home, and that research keeps moving forward with them.

Know better to understand better. Understand better to support better. Support better to open up what is possible.

1 Advance knowledge and research

- Give priority to brain imaging and EEG with our German partners.
- Support research on episignatures at Rouen University Hospital.
- Build a French registry with a European scope.
- Open research to the humanities and social sciences: natural history in teenagers and adults, daily life, pathways, needs.
- Carry families' voices into the French National Rare Diseases Plan, and give clear and cautious information on innovative therapies.

2 Meet the challenge of useful local support

- Keep our tools up to date: condition, disability file guide, turning 18, calendar, syndrome news.
- Strengthen support for the transition to adulthood.
- Identify the right contacts in each region, and keep listening to you.
- Explore residential care for adults, older age, employment.
- Build recommendations with professionals on school, tools and daily life.
- Help buy equipment not otherwise covered, when dedicated funding allows.
- Bring the community to life: discussion groups, days in Rouen, ideally every two years.

3 Build a strong, shared association

- Fund our fixed costs on a regular basis: insurance, digital tools, website, travel.
- Create a half-time coordinator post, if multi-year funding allows.
- Share tasks among volunteers who are welcomed and supported.
- Free up time for joint projects with other associations, such as inclusive housing.

And beyond borders

The syndrome is rare everywhere: no country moves forward alone. We want to develop European cooperation around the France-Germany partnership, coordinate our key events with our sister associations, sign agreements with our partners, and guarantee reliable information, together with medical teams.

Five years: the review

It all started with two families and a genetics team committed to the subject. Here is what we have built together, with a 100% volunteer team.

1st
association in the world dedicated to MYT1L syndrome

80 to 100
families in touch with us in France

200+
participants from 10 countries in Rouen in 2025, including more than 50 children

40+
families helped with their disability file since April 2026

2,300+
website visitors, in more than 20 countries

€20,000
budget per year, approximately: every euro counts

Key milestones

2021	Association founded.
2022	Description of 40 new patients by the Rouen team. 1st international day for families, clinicians and researchers in Rouen.
2023	Recognition of the syndrome: Orphanet code 647799.
2025	2nd international day in Rouen, replays in French and English. Mini-symposium in Heidelberg.
2026	PNDS published in February. Publication of the eating behaviour study. 2026-2030 association project presented at the general meeting on 21 May. Families' area opened in July.

Our momentum has inspired others

The MYT1L Project Foundation in the United States, ExtraValientes MYT1L in Spain, active WhatsApp groups and new associations being set up in Europe. All of them share our elephant.

Tools for everyone

Disability file guide, turning 18, easy-read sheets and cards, videos, flyers, FOCUS magazine, translations: for families and professionals alike.

They made all this possible. Thank you!

The AnDDI-Rares network and Rouen University Hospital; Gan Assurances and the Groupama Foundation (€10,000, « Step by Gan » challenge); the schools and secondary school of Domfront (€8,124, charity run with more than 300 pupils); Ferrero France; the Bornéo agency, for our logo and brand guidelines; the Rouen medical school; Flaubert high school; and the companies, schools, volunteers and families of our regions.

Looking ahead: a small team will no longer be enough. Our resources must become regular to last, and tasks must be shared. That is what area 3 of our project, and our next general meetings (p.13), are all about.

In brief: news you can use

We have sorted the news received in recent weeks. Each item says where it comes from and why it matters to you.

Our international partners

Spain

First family meeting of ExtraValientes MYT1L in Burgos, from 30 October to 1 November.

For you: a sister association, to recommend to Spanish-speaking families.

Source: ExtraValientes MYT1L.

United States

Family-researcher meeting in St. Louis on 13 and 14 November.

For you: follow remotely on Zoom, in English, registration before 9 November.

Source: The MYT1L Project Foundation.

Germany

The MRI and EEG study extends the cooperation started in Heidelberg in 2025.

For you: your relative can contribute without any new test (page 6).

Source: Rouen University Hospital.

Our health network, AnDDI-Rares

Sharing the diagnosis

Four new video testimonies: carers, a couple, a sister, associations, from 9 November.

For you: words to share with your loved ones after a diagnosis.

Source: network newsletter, November 2026; videos on its LinkedIn page.

GENEKIT, in Lille

Patient education workshops: genetics, emotions, procedures, future plans.

For you: open to individuals and relatives, from diagnosis to the teen-adult transition.

Source: Lille University Hospital reference centre. The flyer

19 November event

11th multidisciplinary day in Paris: research with the people concerned, GenIDA, alternative communication.

For you: a photobooth for the mural (page 8).

Source: network invitation.

Your rights, your health

Mobility Inclusion Card

The French ministry for disability answers questions on the card's endorsements, conditions and related rights.

For you: worth reading before your disability file.

Source: ministry, shared by the network.

Genomic medicine

A report by France's IGAS and IGÉSR inspectorates recommends a second national plan for 2027-2036.

For you: what is at stake is access to genome analyses, which help with diagnosis.

Source: public report, shared by the network.

FraMaRa

The French national rare diseases app will have a library of reliable resources.

For you: verified content in one place. Our public resources could be offered there.

Source: French Rare Diseases Alliance, October 2026.

Genetics has no expiry date

To pass on to a family without a diagnosis: going back to a genetics clinic for new tests (exome, genome) is possible at any age. Find a centre (AnDDI-Rares, France).

Events calendar

Conferences, workshops, webinars, carer training: more than 100 events on rare diseases and neurodevelopmental disorders, in France, online and abroad. Filter by region, topic, audience and format. The calendar is updated **twice a month**. Open the calendar

Agenda

Our selection for you

Date	Event	Where
13 Nov.	Webinar « Self-determination, autonomy and safety », iMIND centre, 12.30 pm	Online
13 Nov.	Webinar « Sleep disorders », Dijon NDD Federation, 1 pm	Online
13-14 Nov.	MYT1L Family-Scientist Meeting, in English, Zoom registration until 9 Nov.	Online, St. Louis
14 Nov.	Organising and passing on family assets (Autisme Paimpol)	Paimpol
17 Nov.	« My first disability application for my child », designed for Finistère	Online
18 Nov.	Rare diseases teen-adult transition day, ages 15 to 25	Nice
19-20 Nov.	Eating, sleep and behaviour in young children (ANECAMSP)	Paris or online
24 Nov.	« When the body speaks »: expressing pain with alternative communication	Online
26-27 Nov.	National disability conference, open to carers	Niort or remote
26 Nov.	**JIMR testimony** to send to the Alliance before this date (p.8)	By e-mail
27 Nov.	4th GénoPsy congress: genetics, behaviour and mental health	Rouen
4 Dec.	« Rare diseases: from school to work », Paris Nord platform	Paris or online
5 Dec.	**Rare Diseases March**	Paris
10 Dec.	2nd day of the Normandy rare diseases platform	Normandy
11 Dec.	Review of the 3rd national rare disabilities plan, registration before 1 Dec.	Online
25 Feb.	**MYT1L Day**	Everywhere
28 Feb.	**Rare Disease Day** and unveiling of the mural	Network centres, Paris, online

Check times, prices and registration in the calendar: some events are paid or have limited places. An event to suggest? Write to us.

Our next general meetings

Online

The date will be sent to you by e-mail

Extraordinary general meeting: adoption of the new statutes.

Ordinary general meeting: review of 2026, 2026-2030 project, 2027 budget, membership fees, election of the board.

After the general meeting on 21 May, we are meeting again to decide on these matters together. Your notice will give the date and agenda. Can't attend? You can give a proxy.

We are looking for new energy

Treasury first, but also families and local support, transitions and rights, communication, fundraising, association life. From home, at your own pace: each task is described step by step, and nobody is alone with their task. **Applications are open**: extravaillants@gmail.com

There are a thousand ways to act with us

Join

To give more weight to families' voices.

Give time

An hour or two a week, from home.

Offer your skills

Design, translation, law, IT, accounting.

Organise an event

Race, concert, sale, company challenge.

Share your story

Nothing is published without your written consent.

Spread the word

Share our posts around you.

Tax-deductible donations and membership (France)

66% for individuals: a €100 donation costs you €34. **60%** for companies, under French tax rules. The association is recognised as being of general interest.

Act with us

Donate, join

Follow us

Facebook for real-time news, Instagram, YouTube for videos and Rouen replays, and the website for in-depth information.

en.extra-vaillants-myt1l.com

Website

MYT1L Les extra vaillants