What is it?

MYT1L syndrome

A rare genetic condition

- Caused by loss of function (haploinsufficiency) of the MYT1L gene
- Located on chromosome 2 (region 2p25.3)
- Described as a non-degenerative condition
- Mostly diagnosed in childhood, but possible at any age: consider it in adults too
- First described in 2011

Inheritance

- DE NOVO (most common): a variant that arose spontaneously in the child and is absent in both parents
- INHERITED: autosomal dominant transmission from a parent who carries the syndrome

Three genetic forms

- DELETION: part of the gene, or the whole gene, is missing on one copy
- POINT MUTATION: a “spelling mistake” in the DNA
- CERTAIN DUPLICATIONS, located within the gene, which disrupt it
- In all three cases: haploinsufficiency — only one working copy

Where is the diagnosis made?

- PCO (French early-assessment and referral platform) — entry point, ages 0–12
- CAMSP (French early medico-social care centre) — ages 0–6
- Genetics clinic — university hospital, reference centre
- Reference Centre for Developmental Anomalies — Rouen University Hospital (France)

How is it confirmed?

- Genetic testing: exome sequencing (WES), genome sequencing (WGS) or array CGH
- Rarely suspected on clinical presentation alone
- Molecular diagnosis is essential to confirm the syndrome

Epidemiology

- Rare disease — exact prevalence unknown
- About 100 families in France; families in more than 20 countries
- 3 national associations (France, United States, Spain); Poland and Germany in preparation

What is the impact?

Clinical presentation of MYT1L syndrome

Language & communication

- First words: median ~2.3 years
- First sentences: median ~5 years
- Wide variability between individuals
- AAC recommended if speech is absent or very limited
- Oro-myofunctional disorders and verbal dyspraxia

Intellectual development

- Intellectual disability is common, of variable severity
- Learning difficulties with or without intellectual disability
- ADHD: ~38% of cases
- Autism spectrum disorder (ASD): common

Behavioural difficulties

- Very common from early childhood
- Impulsivity, disinhibition, low frustration tolerance
- Anxiety, self-injurious or aggressive behaviour
- Risk of worsening in adolescence

Weight & eating

- 58% are overweight (23%) or have obesity (35%)
- 45% have eating disorders: hyperphagia, lack of satiety, rapid eating
- Food stealing or hiding is possible
- Can occur without excess weight

Motor & psychomotor skills

- Axial and/or peripheral hypotonia
- Delayed motor milestones
- Coordination difficulties
- Stereotypies in ~55% of cases
- Sensory processing difficulties

Other features

- Epilepsy: 23% of patients
- Eye and vision problems: 30% — neurovisual orthoptic assessment recommended
- Frequent sleep disorders, sleep apnoea (OSA)
- Mild, non-specific dysmorphic features

! Profiles vary widely — symptoms, their severity and their course differ greatly from one person to another.

Care and support

Management of MYT1L syndrome

There is currently no specific curative treatment for MYT1L syndrome.

RECOMMENDATION: multidisciplinary support, started as EARLY as possible

Medical care

- Paediatric neurologist / Neurologist
- Geneticist (coordination, lifelong follow-up)
- Child psychiatrist / Psychiatrist
- Endocrinologist / Nutrition specialist
- Obesity specialist
- Ophthalmologist + Orthoptist
- ENT specialist
- GP / Paediatrician

Therapies

- Speech and language therapist (language + AAC)
- Psychomotor therapist
- Occupational therapist
- Physiotherapist
- Orthoptist (neurovisual)
- Neuropsychologist / Psychologist
- Dietitian
- Adapted physical activity instructor

Key points

- AAC: start early if speech is absent or limited
- Adapted physical activity: recommended at all ages
- Structured eating routine: fixed mealtimes, limited access to food
- Melatonin may be considered for sleep problems
- Therapy at any age: late progress is possible

PARADOXICAL EFFECTS OF MEDICATION: the same treatment can have opposite effects in different patients. **Weight gain:** valproate, methylphenidate. **Worsening of behaviour:** aripiprazole, risperidone, methylphenidate. **Partial improvement in appetite:** methylphenidate, atomoxetine. Regular clinical monitoring is essential.

Care pathways

How care is organised in France

PATHWAYS VARY — no single solution — French services adapt to each person's needs · MDPH: local disability office

Entry point (ages 0–12)

- PCO: functional assessment + funded early-intervention package (24 months max.)
- CAMSP (ages 0–6): early medico-social support
- GP / Paediatrician / PMI (child health service)

Schooling & medico-social support

- Mainstream school with AESH (support assistant) and/or PPS (school plan)
- PAS (school support hubs) — no MDPH application needed
- ULIS, SEGPA, UEEA (special units in schools)
- DAME / DIME: flexible medico-educational services after 2026

Care & therapies

- Private practice: speech, psychomotor and occupational therapists, physiotherapist
- CMP / CMPP (mental-health centres)
- CAMSP
- Hospital / Reference Centre — CHU Rouen

Rights & benefits (MDPH)

- AEEH (child allowance) → AAH (adult allowance, from age 18)
- PCH (compensation benefit), now covers neurodevelopmental disorders
- AESH + adapted learning materials
- CMI (inclusion card) — parking card recommended
- Respite for carers
- MDPH guide and "J'ai 18 ans" (turning 18) file, in French — families-only area of the website, or on request

Transition to adulthood

- Pathways are still little known and are built around each person's own plans
- Plan ahead before age 16–18 (Creton amendment, legal protection)
- Work: mainstream (RQTH status), supported employment, ESAT (sheltered work)…
- Living: family home, supported or inclusive housing, care homes (FAM, MAS)…
- AAH via the MDPH from age 18

Resources & patient education

- Patient education (ETP): AnDDI-Rares / Défiscience rare-disease networks
- Families–Clinicians–Researchers Days 2022 & 2025 (replays on YouTube)
- ERHR (rare-disability relay teams): support in complex situations
- Communautés 360: 0 800 360 360 (French helpline), local solutions

And the families?

The association · “Vaillants”: people living with MYT1L syndrome

Les Extra-Vaillants MYT1L association — The world’s first association dedicated to the MYT1L gene, founded in 2021

Who are we?

- The only French-speaking association dedicated to MYT1L syndrome
- Based in France (Normandy)
- Public-interest charity (France)
- Member of Alliance Maladies Rares (France)
- Partners: AnDDI-Rares, Défiscience, CHU Rouen

For families

- Welcoming new families, welcome booklet
- La Bulle des Extra-Vaillants, international group and WhatsApp groups (access on request)
- Online events, video calls and meet-ups
- MDPH guide and “J’ai 18 ans” file (in French)

For the Vaillants

- Tools for talking about the syndrome with your child
- Easy-read sheets and hands-on cards
- “J’ai 18 ans”, “Ton dossier MDPH” and “Ton argent”, in easy-read French

Care and research

- Families–Clinicians–Researchers Days 2022 and 2025
- Research projects: follow-up and family participation
- Guides: “Understanding research” and “Understanding an ASO”
- Study summaries and publications
- “Impacts and care” tools in several languages

Raising awareness

- FOCUS magazine
- Videos and replays on YouTube
- Website and social media
- Taking part in rare-disease conferences

Around the world

- **FR:** extra-vaillants-myt1l.com (pioneer, 2021)
- USA: The MYT1L Project (late 2024)
- ES: Extravalientes MYT1L (2025) · Poland, Germany: in preparation
- Families in more than 20 countries, on every continent
- About 100 families in France

Where to find resources?

Resources and contacts

Join us

- **Website:** extra-vaillants-myt1l.com
- **Facebook:** /extravaillants
- **YouTube:** Les Extra-Vaillants MYT1L channel
- Donations, memberships and volunteers welcome

Available resources

- Introductory videos and replays of the Days — YouTube Extra-Vaillants
- Tools for the Vaillants, easy-read sheets — on our website
- Hands-on easy-read cards — on request via the website
- MDPH guide, “J’ai 18 ans” file — families-only area of the website, or on request
- Family welcome booklet, FOCUS magazine — on the website
- PNDS MYT1L care protocol (HAS, Feb. 2026) — has-sante.fr

Quick access

Scan with your phone camera

Our videos

The website

Tools for the Vaillants

The PNDS

Write to us

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