MYT1L

Les extra vaillants

MYT1L SYNDROME

Rare genetic neurodevelopmental disorder
Complex profiles

ORPHAcode: 647799

French PNDS available

PNDS: French national diagnosis and care protocol

- Rare genetic anomaly in chromosome region 2p25.3, which includes the MYT1L gene
- Haploinsufficiency: loss of function of one copy of the gene
- Caused by a deletion, a mutation or certain duplications
- Most often de novo; more rarely inherited from a carrier parent

It involves

- Global developmental delay
- Varied profiles: from multiple disabilities to supported independence
- Severity and number of effects vary from one person to another
- Lifelong care and support

Les Extra-Vaillants MYT1L

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- Medical lead: Dr Juliette Coursimault, clinical geneticist - Reference Centre for Developmental Anomalies and Malformation Syndromes, Rouen University Hospital (France), AnDDI-Rares network
- Member of Alliance Maladies Rares (France)
- Recognised public-interest charity in France
- Many tools: PNDS, guides, easy-read sheets... and resources on our channels

Site internet

Facebook

YouTube

About 100 families in France

- Families in more than 20 countries. Les Extra-Vaillants MYT1L, the world's first (France, 2021), The MYT1L Project (USA, late 2024), MYT1L Extravalientes (Spain, 2025); Poland and Germany in preparation
- International MYT1L Families-Clinicians-Researchers Days (2022, 2025 — replays on YouTube)
- Private Facebook groups, on request

Updated November 2026 — Ref.: PNDS MYT1L, CHU Rouen (HAS website, February 2026)

IMPACT

- Delay mainly affecting language
- Oro-myofunctional disorders and verbal dyspraxia
- Intellectual disability and/or learning difficulties (possible without ID)
- Neurovisual disorders
- Behavioural difficulties very common (alone or combined), often with ASD and/or ADHD
- Sensory processing difficulties
- Anxiety disorders from childhood to adulthood
- Psychomotor development disorder, hypotonia
- Weight problems (58% overweight/obesity) and/or eating disorders (45%, with or without excess weight)
- Epilepsy (~23%)
- Fatigue
- Frequent sleep disorders, sleep apnoea (OSA)
- Brain MRI anomalies
- Non-specific dysmorphic features

SUPPORT - TREATMENT

NO SPECIFIC CURATIVE TREATMENT EARLY, COORDINATED MULTIDISCIPLINARY SUPPORT IS ESSENTIAL

- Where to turn: French reference centre (CRMR) for developmental anomalies – CHU Rouen, and local reference and expert centres (CRMR, CCMR)

Multidisciplinary team involved (not exhaustive)

- **Medical**: paediatric neurologist, geneticist, child psychiatrist, endocrinologist / nutrition specialist, ophthalmologist, ENT, GP...
- **Therapies**: speech therapist, occupational therapist, physiotherapist, psychomotor therapist, neurovisual orthoptist, adapted physical activity instructor...
- **Psycho-educational** : neuropsychologist, psychologist, dietitian, social worker, school support assistant...

Symptomatic treatments are possible, with attention to paradoxical effects (agitation, weight gain, worsening); close clinical monitoring is essential

PATHWAYS

- **MYT1L MDPH guide** (French disability-rights guide), **full "J'ai 18 ans" (turning 18) file** (rights, protection, assets), **Family welcome guide.** In French, in the "Réservé aux familles" area (pink button) of the association's website, or on request by e-mail: extravaillants@gmail.com

- **Schooling: a pathway adapted to each pupil**
  In mainstream school, in a specialised setting or home-schooled, with adapted equipment, adjustments and one-to-one support set out in the schooling plan (PPS)

- **Adult life** : pathways still little known
  Plan from adolescence: work, rights, housing, social and medico-social support

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