SUMMARY · STUDY PUBLISHED IN 2026

Eating behaviour phenotype in the neurodevelopmental disorder related to the *MYT1L* gene

This multicentre study describes in detail the eating behaviour of 21 people (14 children and 7 adults, aged 6 to 37) with a neurodevelopmental disorder (NDD) related to the MYT1L gene and overweight, obesity or eating behaviour disturbances. It highlights features whose intensity and consequences vary from one person to another.

Read this first

The original publication is in English. This document is not a translation of it but a summary written by the association Les Extra-Vaillants MYT1L (originally in French), as the best balance between faithfulness to the data and readability. For full details, please refer to the original publication.

The results describe a group of participants, not any particular person: for your relative's situation, the team that supports them remains the best contact. **Only the reference publication is authoritative.**

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Study reference

Coursimault et al., *Orphanet Journal of Rare Diseases*, 2026. **Eating behaviour phenotype in the MYT1L-related neurodevelopmental disorder: a deep phenotyping study using standardized questionnaires.**

Work by Dr Coursimault (Department of Genetics, Rouen University Hospital), Prof. Poitou and Prof. Dubern (PRADORT reference centre, Pitié-Salpêtrière and Trousseau hospitals respectively, AP-HP, Paris). Collaboration with hospital departments and research teams in France, Belgium and Luxembourg.

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1. Background and objectives

The neurodevelopmental disorder related to the MYT1L gene (also called MYT1L syndrome) is a rare genetic condition, described recently. It is caused by loss of function of one of the two copies of the MYT1L gene, located on chromosome 2 (region 2p25.3). This loss of function can result from:

- a variant (point change) in the gene, truncating or missense;
- a deletion of the 2p25.3 region removing all or part of the gene;
- an intragenic duplication, i.e. one whose breakpoints lie inside the gene. According to the PNDS (French national care protocol), other duplications, which keep a complete copy of the gene, do not lead to this mechanism.

Features vary from one person to another, in nature as well as intensity. The article and the PNDS mention in particular the following features, which are not all present in every person:

- delayed motor and language development, and hypotonia (low muscle tone);
- intellectual disability and/or learning disorders, of variable intensity;
- behavioural difficulties: intolerance of frustration, anxiety, aggression, autism spectrum disorders;
- epilepsy;
- physical features (facial dysmorphism);
- early-onset obesity, which is common.

The key 2022 study (Coursimault et al., *Hum Genet.* 2022), describing a new cohort together with a literature review, reported obesity in about 60% of people and eating behaviour disturbances in about 45%, with a median age at onset of weight gain of about 3 years. However, these disturbances had never been specifically characterised.

Objectives of the study

- describe the eating behaviour phenotype;
- compare it with other rare genetic obesities;
- draw leads for diagnosis and support.

2. Participants

Number	21 people with a confirmed genetic diagnosis; 10 new, 11 already described in 2022
Sex	12 female, 9 male
Age	14 under 18, 7 adults; aged 6 to 37 (median age: 10)
Weight	Of 20 people: 14 with obesity, 3 overweight, 3 of normal weight. Rapid weight gain (or parents' concern) began at about 3 years on average
Genetics	16 loss-of-function changes (14 truncating, 2 deletions removing the gene) and 5 missense changes, all located in the zinc-finger domains; *de novo* in 20 out of 21 people, unknown origin for 1
Inclusion criteria	French-speaking people with a confirmed molecular diagnosis, with overweight or obesity, or clinically reported eating behaviour disturbances
Reported eating behaviour disturbances	16 out of 21 people according to their parents, including 12 with overweight or obesity

The article does not detail the split between girls and boys among children and adults.

3. Method

Standardised questionnaires, used in research and recommended in the French national care protocol (PNDS) for obesities of rare causes, were completed by families or by the referring doctor, as part of routine follow-up. Each tool explores a different dimension of eating behaviour.

Tool	What it measures	Completed for
Dykens hyperphagia questionnaire	food-focused thoughts, food seeking, severity	21 people
Visual analogue scale (VAS) for hunger	hunger over the last 24 hours	19 people
CEBQ	attraction to food, satiety, speed of meals, selectivity, emotions	21 people
Trousseau impulsivity questionnaire	food impulsivity	18 people
FCQ-T-r (adults)	food cravings and food addiction	8 people (7 adults, 1 teenager)
PAR-DD-QoL	parents' quality of life linked to eating behaviour	20 parents

Questionnaires are not the only source. The study also relies on clinical and genetic data from medical follow-up: molecular diagnosis, weight status (body mass index according to WHO standards), age at onset of weight gain (reported by parents or collected by the referring doctor), specialist follow-up, current treatments, endocrine assessment. Three additional questionnaires explored social situation (EPICES), anxiety and depression in adults (HADS) and physical activity (RPAQ).

Scores were compared, where possible, with published data for control groups (children, students) and for other rare genetic obesities.

4. Results, tool by tool

Each card presents one tool: what it explores, the key takeaway, then the key figures.

Hyperphagia · Dykens questionnaire 21 people

What the tool explores: hyperphagia: food-focused thoughts, food seeking, severity (score from 11 to 55). In France, a score ≥ 19 is used as the threshold for marked hyperphagia (expert consensus from the Prader-Willi PNDS), without formal validation as a universal threshold.

In plain words: in most people, moderate but persistent hyperphagia, less intense than in MC4R or LEPR deficiency, and also apparently less intense than in Prader-Willi syndrome or Bardet-Biedl syndrome.

- 16 out of 21 (76%) reach or exceed 19; mean score 25.9 (median 27; range 12 to 41).
- First signs of interest in food appeared at 3½ years on average (range 0 to 7); food preoccupation described as stable over time in 63% of cases.

Mean Dykens score (published data)	
LEPR deficiency (biallelic) · MC4R deficiency (monoallelic)	32 · 31.4
Prader-Willi syndrome (2 studies)	30.5 and 29.3
Bardet-Biedl syndrome	27.6
MYT1L-related NDD	**25.9**
16p11.2 deletion	21.4

Hunger · visual analogue scale (VAS) 19 people

What the tool explores: hunger felt over the last 24 hours, from 0 (not hungry) to 10 (extremely hungry), reported with the help of relatives if needed, on 4 criteria (average hunger, hunger on waking, hunger at its highest and at its lowest).

In plain words: hunger is present throughout the day, from waking, including at its lowest level.

- Means out of 10 over the last 24 h: 5.1 (average hunger), 5.3 (on waking), 5.7 (at its highest), 2.5 (at its lowest).

Relationship with food · CEBQ 21 people

What the tool explores: eight dimensions of eating behaviour (score from 1 to 5), compared with published data from children without rare diseases in 4 countries (United Kingdom, Spain, Switzerland, Portugal). A tool validated in children, also used here in adults.

In plain words: greater attraction to food, weaker satiety response, faster meals and low food selectivity.

Higher than in controls	Lower than in controls
• Enjoyment of food (4.5 out of 5) **• Food responsiveness: wanting to eat at the sight or smell of food (3.7)** **• Emotional overeating: eating more under the influence of emotions (2.7)**	• Satiety responsiveness (2.1) • Slowness in eating (1.7) • Food fussiness (2.1) • Emotional undereating: eating less under the influence of emotions (1.8)

Significant differences after correction for multiple comparisons; desire to drink does not differ.

Impulsivity · Trousseau questionnaire 18 people

What the tool explores: food impulsivity (in-house tool of Trousseau hospital; exploratory threshold ≥ 7, not validated in this disorder). 18 patients assessed, including 14 children.

In plain words: food impulsivity is uncommon, and no correlation was found with the intensity of hyperphagia.

- 5 out of 18 (28%) above the impulsivity threshold, all children (5 out of 14, i.e. 36%).
- Additional questions completed by families: difficulty adapting to the unexpected (found in 78%), food preoccupation outside meals (found in 44%), no night-time food seeking in any of the 18 patients.

Food cravings · FCQ-T-r 8 people

What the tool explores: irrepressible food cravings, score from 15 to 90; a score ≥ 50 suggests « food addiction ». Completed by 7 adults and 1 teenager aged 17, with the help of relatives.

In plain words: in most people assessed (7 adults, 1 teenager), food cravings are strong.

- 5 out of 8 (62.5%) have a score ≥ 50: a suggestive score, not a diagnosis.
- Mean score 52.9, significantly higher than in unaffected German (38.5) and Iranian (37.6 to 38.4) students.

Parents' quality of life · PAR-DD-QoL 20 parents

What the tool explores: the impact of eating behaviour on parents' quality of life (questionnaire focused, for the study, on eating behaviour).

In plain words: eating behaviour weighs heavily on families' quality of life.

- Severe or extreme impairment: 37%; mild to moderate: 63%.
- Median score of 42, close to those reported in parents of children with an autism spectrum disorder (46.7) or a severe chronic illness (44.7).

Clinical and additional data Varying numbers

What the tool explores: information from medical follow-up and additional questionnaires.

In plain words: oral-motor difficulties (very fast meals, chewing) are common.

- More than 2 out of 3: tachyphagia, chewing difficulties or swallowing large pieces.
- Half: active food seeking (taking food without relatives knowing, persisting to obtain it).
- No significant difference according to the type of change (loss of function or missense), nor between treated and untreated people (11 participants treated, 10 of them with a medicine that may change appetite or weight): numbers too small to conclude.
- No notable endocrine abnormality.
- Physical activity (14 children, 6 adults): more light activity in children (71 min/day versus 20 in adults); comparable moderate activity, mainly walking; vigorous activity and supervised training rare.
- Context: 28.5% of families in a precarious situation (EPICES score > 30); in adults, no pathological anxiety or depression score (4 borderline cases).

5. Putting the results together

Taken on its own, each tool sheds light on only one dimension. Putting them together gives an overall description:

Dimension	Tools	What stands out
Hunger	VAS	Present throughout the day, from waking
Satiety	CEBQ	Weaker satiety response than in controls
Food preoccupation	Dykens, CEBQ	Marked in most, usually stable over time (63%); high enjoyment of food; low selectivity
Way of eating	CEBQ, clinical data	Fast meals; chewing difficulties common
Impulsivity	Trousseau	Uncommon (5 out of 18, all children); no correlation with intensity of hyperphagia
Food cravings	FCQ-T-r	Strong in 5 of the 8 people assessed (7 adults, 1 teenager)
Families	PAR-DD-QoL	Impaired quality of life, severely or extremely for 37% of parents

Conclusions of the study

The observed phenotype. In this group, the MYT1L-related NDD is characterised by early weight gain, associated with moderate but persistent hyperphagia, food preoccupation, hunger persisting throughout the day and impaired satiety. Food impulsivity is low in children, craving is marked in adults, and oral-motor difficulties are common.

The impact on families. These eating behaviour disturbances clearly affect families and justify early and proactive care, as well as targeted interventions.

Prevention from the genetic diagnosis. Measures put in place from the genetic diagnosis (nutritional advice, structured meal routines, promotion of adapted physical activity) could limit weight progression and the risk of obesity-related complications. They could thus help avoid combining a neurodevelopmental disorder with metabolic comorbidities.

A multidisciplinary approach. Age-appropriate, multidisciplinary, family-centred care remains essential, ideally within centres specialised in rare diseases and complex obesities.

What these results describe. They characterise a group selected on the presence of symptoms and do not give the frequency of these features among all people with the MYT1L-related NDD.

Compared with the PNDS

Several observations are consistent with the PNDS for MYT1L syndrome, which reports eating behaviour disorders in about 45% of cases from childhood, frequent and early weight gain, and oral feeding difficulties with chewing problems in some children. It recommends care as early as possible. On the other hand, it mentions marked food selectivity in some children with oral feeding difficulties, whereas the study describes broad acceptance of foods.

6. Strengths and limitations of the study

Strengths of the study

- First detailed and standardised characterisation of eating behaviour in the MYT1L-related NDD.
- Standardised tools, used in other rare obesities and in clinical practice, which allows comparisons.
- French-speaking multicentre study (France, Belgium, Luxembourg); 21 participants, a limited number reflecting the rarity of the condition.
- Multidimensional approach: hunger, satiety, impulsivity, cravings, quality of life, context.

Methodological limitations of the study

- Participants selected on the presence of overweight or obesity, or eating behaviour disturbances; people without weight problems or obvious eating difficulties were not included, which may have increased the frequency and intensity observed.
- Small number, including only 7 adults; assessment at a single point in time.
- Data reported by relatives, with a risk of recall bias; some tools not validated in this disorder or in adults (CEBQ), exploratory impulsivity threshold.
- Comparisons based on published means; possible influence of treatments.

7. Leads for clinical practice

The research team suggests the following leads, mainly for care teams:

- Consider an NDD related to the MYT1L gene in case of early weight gain associated with developmental delay, even without endocrine abnormality.
- Inform families in advance that hunger and food preoccupation persist.
- Structure meals: meal routines, adapting the environment.
- Watch for oral-motor difficulties, such as tachyphagia (very fast meals).
- Adapt follow-up to age, in particular by monitoring food cravings and loss of control over eating in adults.

It stresses the importance of early, multidisciplinary, family-centred care, ideally within centres specialised in rare diseases and complex obesities, and suggests preventive measures from the genetic diagnosis: nutritional advice, structured meal routines, promotion of adapted physical activity. Applying them to an individual situation is a matter for the medical team that follows the person.

8. What next after this study?

For research

- Carry out follow-up (longitudinal) studies, in larger groups, to describe changes with age and after care.
- Confirm the results in independent cohorts, especially in adults and for food impulsivity.
- Guide future drug trials targeting appetite regulation. In the study, only one participant had received semaglutide (1 mg per week), with a temporary benefit: an isolated observation, with no value as evidence.

For support

- Put preventive measures in place from the genetic diagnosis: nutritional advice, structured meal routines, adapted physical activity, which could help limit weight gain and obesity-related complications.
- Develop adapted physical activity (APA), taking into account the obstacles specific to the disorder: motor difficulties, hypotonia, fatigue, behavioural difficulties, limited access to adapted programmes.
- Support families individually and identify early those in a precarious situation.
- The association's proposal, based on the PNDS: APA on medical prescription, and an oral-myofunctional assessment (speed of meals, chewing, swallowing) with a view to possible therapy by a speech therapist or physiotherapist.

9. For families: questions and resources

If you think it may help care: a few questions to raise with the team supporting the person

These suggestions do not come from the study. They are given for guidance, to be adapted to each situation.

- **Treatments:** if you have doubts about the effect of a treatment on appetite or weight, you can talk to the doctor who prescribes it. Noting the changes observed and when they appeared can help make the link. The PNDS for MYT1L syndrome notes that some psychotropic drugs can interfere with appetite regulation.
- **Meals and chewing:** if meals are very fast or chewing is difficult, you can ask for an assessment, with a view to possible therapy by a speech therapist or physiotherapist. The PNDS provides for assessment of oral-myofunctional and feeding skills.
- **Adapted physical activity:** if adapted physical activity (APA) seems useful, you can raise it with the team supporting the person, including the possibility of a prescription. The PNDS lists the APA teacher among the professionals involved in follow-up and provides for assessing physical activity.
- **Future studies:** if you would like to take part in future studies, you can ask whether the team will contact you or whether you should come forward, and ask to be informed and advised so you can decide whether to take part.

The association's resources

- The association's website: en.extra-vaillants-myt1l.com (French site: www.extra-vaillants-myt1l.com, with an area for families).
- Videos of the MYT1L International Days (talks by clinicians and researchers): YouTube channel

Publications and reference documents

- The original article (in English, open access): read the article
- The PNDS for MYT1L syndrome (French National Authority for Health website, in French): see the rationale
- The 2022 study on 40 new people: read the article
- The 2017 study linking the MYT1L gene to intellectual disability and obesity (open access): read the article
- All publications, regularly updated: association website

This document can be passed on to the professionals who support the person: doctor, dietitian, educational or social care team.

10. Source and credits

Original article: Coursimault et al., *Orphanet Journal of Rare Diseases*, 2026. Eating behaviour phenotype in the MYT1L-related neurodevelopmental disorder: a deep phenotyping study using standardized questionnaires. doi.org/10.1186/s13023-026-04497-x

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Writing: summary written by the association Les Extra-Vaillants MYT1L. Only the reference publication is authoritative. Any comment or request for correction can be sent to the association: extravaillants@gmail.com.

Thank you to the families who took part in this study and to all the research teams.

11. Glossary

NDD	Neurodevelopmental disorder.
Eating behaviour	The way of eating in the broad sense: hunger, fullness, cravings, speed, food choices, emotions. Its disturbances, studied here, are distinct from eating disorders (anorexia, bulimia, binge-eating disorder), which are not the subject of the study.
Phenotype / phenotyping	Phenotype: all the observable characteristics of a person, here those of eating behaviour. Phenotyping: detailed and standardised description of these characteristics.
Eating behaviour disturbances	Term used in the study: particular or atypical features in the way of eating (hunger, satiety, interest in food, speed of meals, cravings, impulsivity).
Hyperphagia	Excessive food intake combining strong hunger, reduced satiety, persistent food seeking and psychological impact.
Satiation	The feeling that leads to stopping eating during a meal.
Satiety	Absence of hunger after a meal, until the next meal. « Satiety responsiveness » (CEBQ) measures the ability to reduce intake after eating.
Craving	An intense food desire that is hard to resist.
Tachyphagia	Eating very fast.
Oral-motor	Relating to mouth movements: chewing, swallowing.
Food impulsivity	Difficulty delaying the immediate urge to eat.
Food addiction	Very strong food cravings with loss of control; here assessed by a questionnaire score, not diagnosed.
De novo	A genetic change that appeared in the person, not inherited from the parents.
Truncating / missense change	Truncating: leads to a shortened protein or none at all. Missense: a single amino acid in the protein is replaced by another.
Loss of function	A change that prevents one copy of the gene from producing a functional protein.
Mean / median	Mean: sum of the results divided by the number of participants. Median: the value that splits the group into two equal halves.
Statistically significant	A difference clear enough that it is unlikely to be due to chance.
PNDS	Protocole national de diagnostic et de soins: French national diagnosis and care protocol, a reference document published by the French National Authority for Health (HAS).