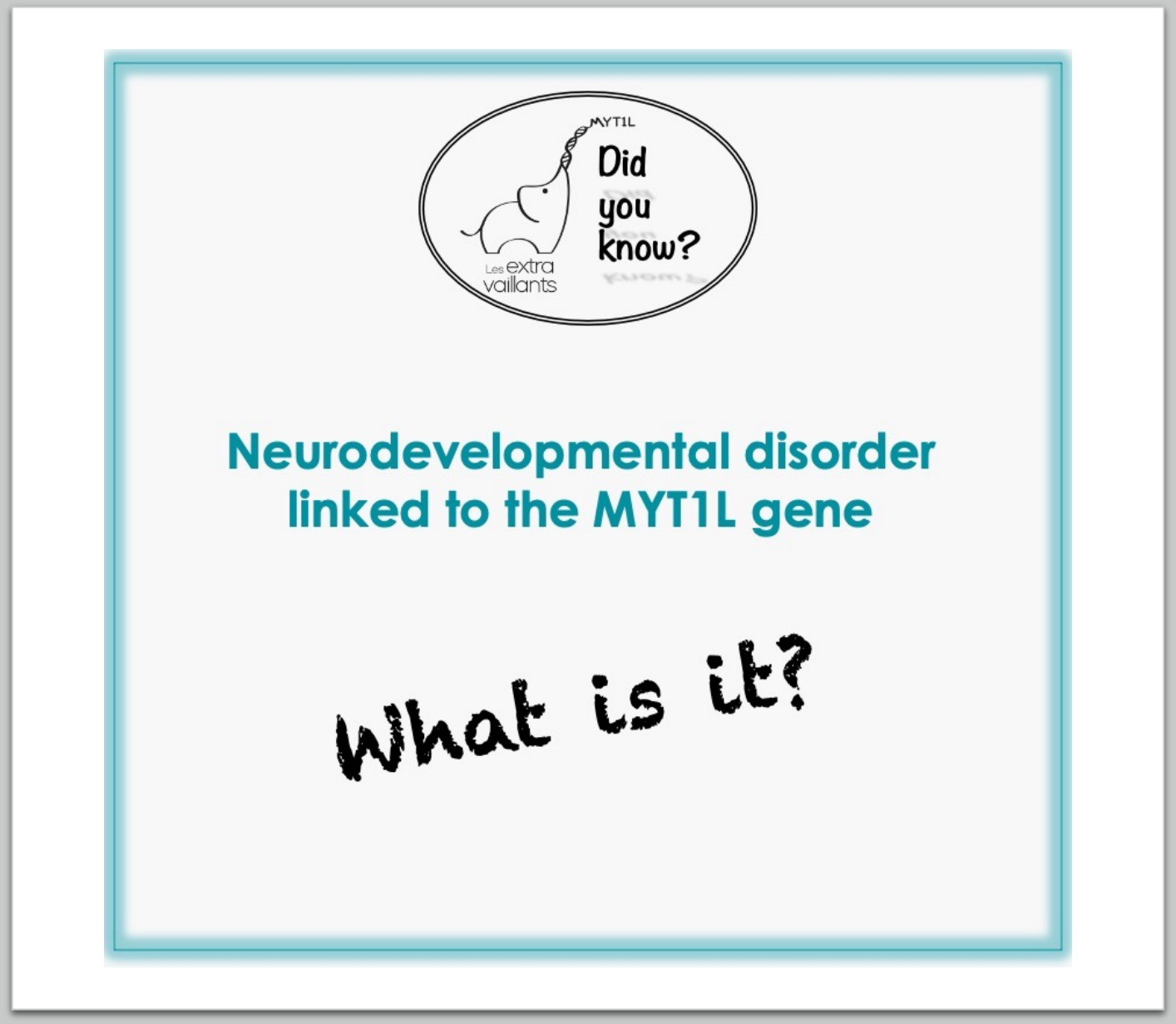


Created by :

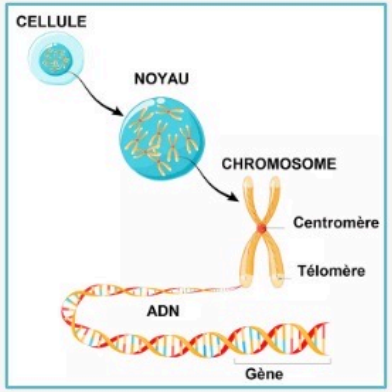
Association Les Extra-Vaillants MYT1L

in collaboration with the geneticists

Dr Coursimault, Dr Guerrot, Dr Lecoquierre, from the reference center for developmental abnormalities of the Rouen University Hospital (2022)

Neurodevelopmental disorder linked to the MYT1L gene

- **A rare genetic disorder**
- **Located in the chromosomal region 2p25.3**
- **Involving the MYT1L gene**
- **Non-degenerative**



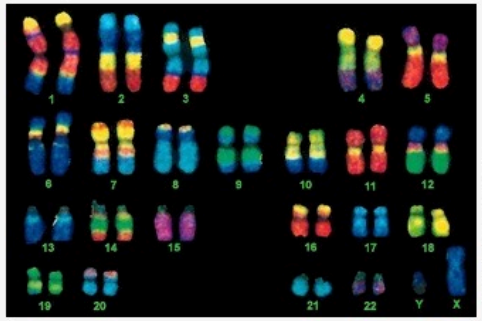


Image : medicinus.net

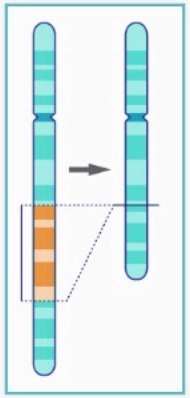
Most of us have 23 pairs of chromosomes

Neurodevelopmental disorder linked to the MYT1L gene

Manifesting in two ways

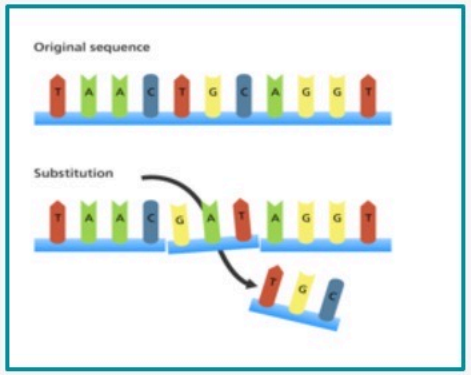
DELETION

The loss of a part or the totality of a gene



MUTATION

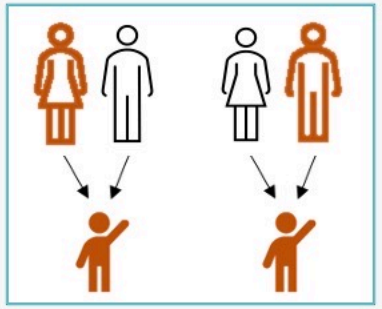
Anomaly in the gene sequence



Neurodevelopmental disorder linked to the MYT1L gene

Hereditary

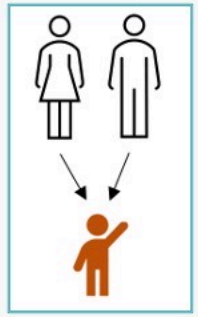
from a symptomatic parent



A person with this genetic disorder has a 50% chance of transmitting it to their offspring

or

« DE NOVO »



De novo gene birth is when a new gene evolves from ancestrally non-genic DNA sequences

MYT1L

Did you know?

Les extra vaillants

Neurodevelopmental disorder linked to the MYT1L gene

Practically Speaking?

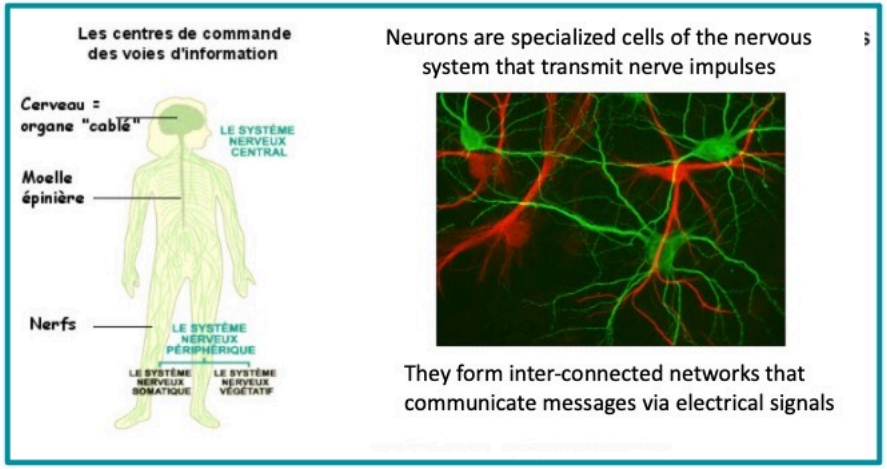
MYT1L

Did you know?

Les extra vaillants

Neurodevelopmental disorder linked to the MYT1L gene

The MYT1L gene is involved in the process of neurogenesis (neuron maturation specifically)

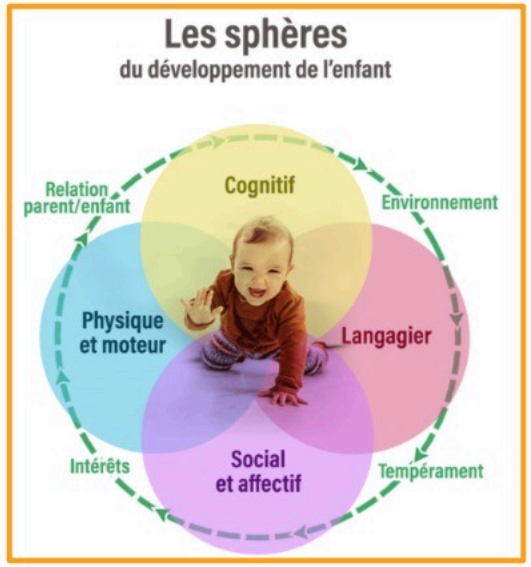


Source : Plateforme ACCES

Neurodevelopmental disorder linked to the MYT1L gene

This syndrome manifests itself through a

global developmental delay (GDD)



Neurodevelopmental disorder linked to the MYT1L gene

- **The typology is heterogeneous: from multiple disabilities to assisted autonomy**
- **Not all patients are impacted in the same way from this syndrome, and its severity varies from one patient to the next**

Neurodevelopmental disorder linked to the MYT1L gene

THE EFFECTS?

Neurodevelopmental disorder linked to the MYT1L gene

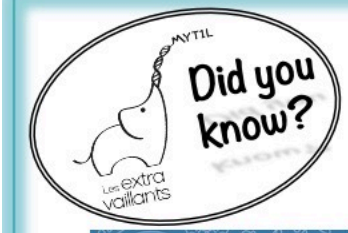
Almost 100% have singular or associated behavioral disorders

1 in 2 patients

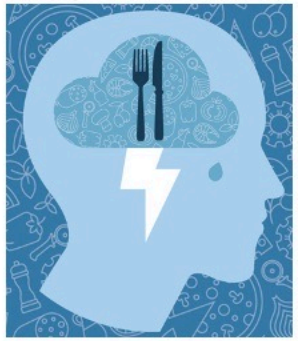
- **Has stereotypies (repetitive or ritualistic behaviors)**
- **Have an impulse control disorder, or frustration intolerance**

4 out of 10 patients

- **Have aggressive tendencies towards themselves or towards others**
- **Present with autism spectrum disorder**
- **Present with an attention deficit disorder**



Neurodevelopmental disorder linked to the MYT1L gene



Weight and/or Eating Disorders

1 in 2 patients
has an eating disorders
(hyperphagia, impulsivity, orality disorder)

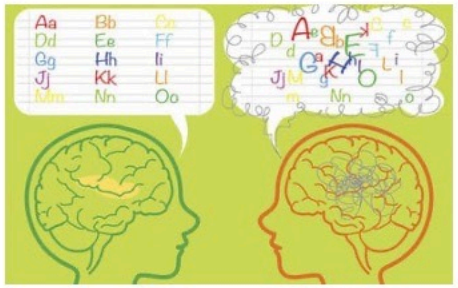
6 out of 10 patients
have a weight problem
(overweight or obese)

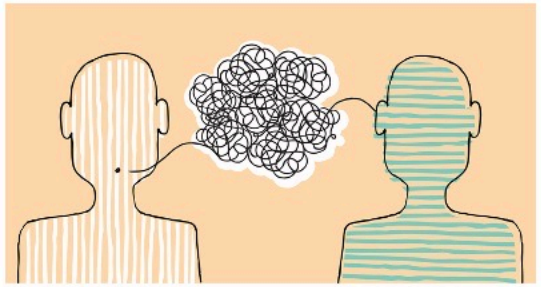
4 out of 10 patients
are of a normal weight

Did you know?

Neurodevelopmental disorder linked to the MYT1L gene

Predominant Language Delay





Speech Disorders

The median age for forming their first sentences is around 5 years old

Did you know?

Neurodevelopmental disorder linked to the MYT1L gene

Intellectual Disabilities or Learning Disabilities

7 out of 10 patients
have an intellectual disability of varying degree

3 out of 10 patients
do not have intellectual disabilities but have a combination of multiple learning disabilities

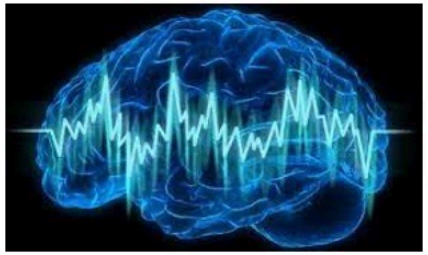
Neurodevelopmental disorder linked to the MYT1L gene

Source : Hoptoys

Psychomotor Development

- **Median age of walking is at 22 months, some require mobility aids.**
- **Coordination issues, unstable motor skills, impact on fine motor skills**
- **Hypotonia**

Neurodevelopmental disorder linked to the MYT1L gene



Epilepsy

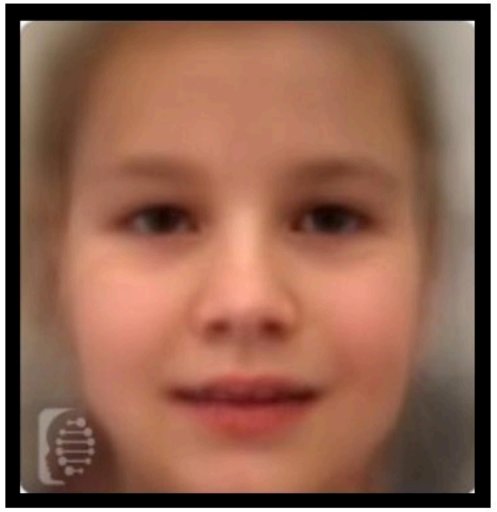
2 out of 10 patients have epilepsy

Brain MRI Abnormalities

3 out of 10 patients have brain imaging abnormalities

Neurodevelopmental disorder linked to the MYT1L gene

Non-specific dysmorphic features

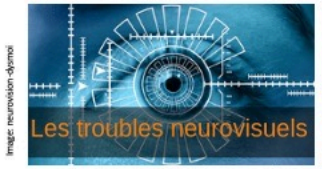


Patients do not necessarily have distinctive morphological features associated with this syndrome.

The portrait was generated using the research software Face2gene.

Neurodevelopmental disorder linked to the MYT1L gene

Other observed disorders



- **Neuro-Visual disorders**

- **Sleeping disorders**

- **Fatigability**

Neurodevelopmental disorder linked to the MYT1L *gene*

A TREATMENT?

Neurodevelopmental disorder linked to the MYT1L *gene*

NO SPECIFIC TREATMENT for this syndrome to date

- **Treatment may be suggested for some of the impacts of the syndrome**
- **Strict frameworks can be imposed for diet, sports**

Did you know?

Neurodevelopmental disorder linked to the MYT1L *gene*

WHAT TO DO THEN?

Did you know?

Neurodevelopmental disorder linked to the MYT1L *gene*

RECOMMENDATION

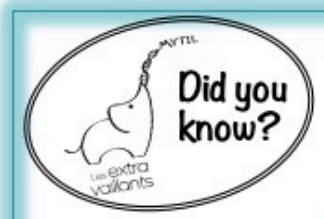
Multidisciplinary support as EARLY as possible

Did you know?

Neurodevelopmental disorder linked to the MYT1L *gene*

RECOMMENDATION

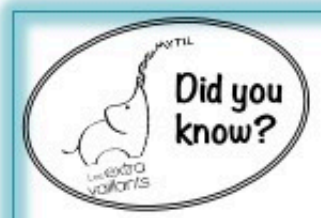
Multidisciplinary support as EARLY as possible



Neurodevelopmental disorder linked to the MYT1L *gene*

Care and rehabilitation to be considered

- Neuro-paediatrician / paediatrician / neurologist
- Endocrinologist
- Geneticist
- ENT
- Psychiatrist/child psychiatrist
- Doctor specialising in sleep disorders
- Doctor specialising in eating disorders
- Ophthalmologist and orthoptist



Neurodevelopmental disorder linked to the MYT1L *gene*

Care and rehabilitation to be considered

- Physiotherapist
- Speech therapist
- Neuropsychologist
- Psychomotrician
- Occupational Therapist
- Psychologist
- Specialised educator
- Nutritionist
- Osteopath...

Neurodevelopmental disorder linked to the MYT1L *gene*

WHAT COURSE?

Did you know?

Neurodevelopmental disorder linked to the MYT1L gene

THE PATHS ARE HETEROGENEOUS

NO UNIQUE SOLUTION

THERE ARE MANY DEVICES

it depends on :

- The patient's profile and needs
- Family resources (time, finances, mobility...)
- Access to care and social assistance
- Medical prescriptions
- Institutional decisions

Did you know?

Neurodevelopmental disorder linked to the MYT1L gene

AREAS OF FOCUS - examples

- Encouraging a 1:1 approach to learning
  - Reinforcing self-confidence
  - Reinforcing commitment to the task
  - Reinforcing attention
  - Helping to manage emotions positively
  - Report on progress

  ! Individual help is not an obstacle to independence
- Use planning tools and benchmarks.
- Support with visual aids and manipulatives
- Compensate for the gesture with a tablet or computer or mobile letters if necessary.
- Be prepared to give more time, and accept that the person may be tired.
- Have ambition and a project built in the zone of proximal development.