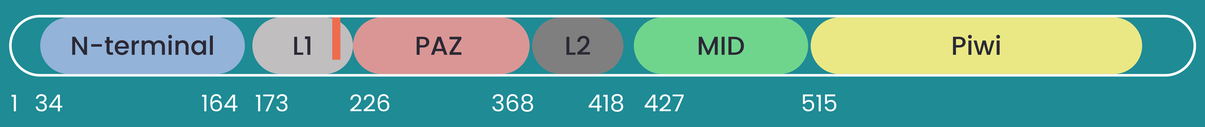
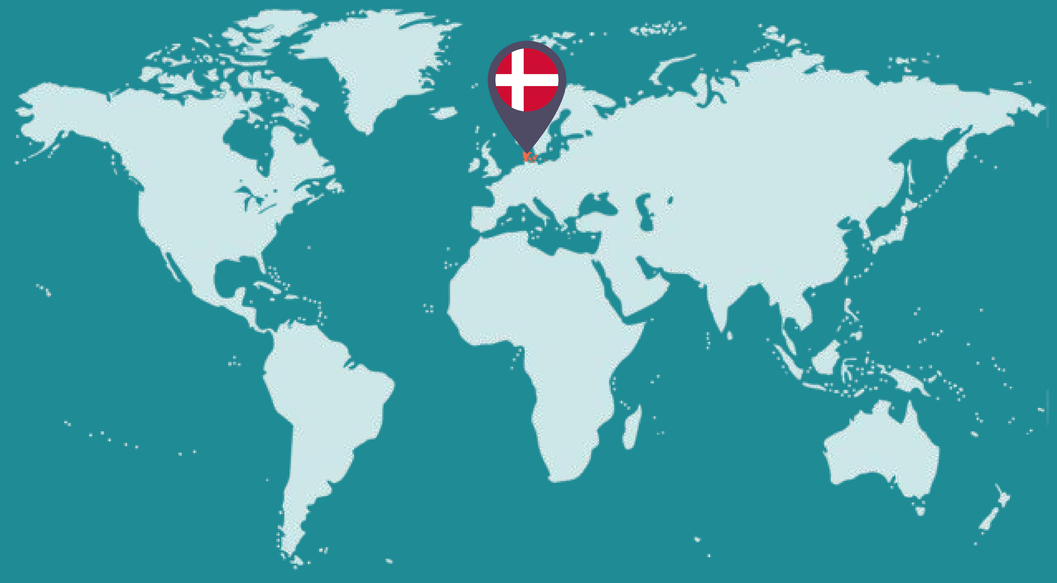
JACOB

Mutations:

de novo AGO1 c.569T>A, p.(Leu190Gln)





Age	12 years old
Family	Mother, Father, Older brother
Age of Diagnosis	Psychomotor retardation developmen at 6 months, vision impairement at 3 months, floppy child syndrome at 3 months, focal epilepsy at 1-2 year and ongoing, AGO1 8 years,

Being easy to excite
High pain threshold
Long response time
Trouble with sleep
Seizures/epilepsy
CVI visual impairment
Poor impulse control
Stiff or jerky movements

Psykomotor retardation
Challenges with koordination
Mental disability
Low muscle tone
Hygenic problems
Little speech
Easily excited
Smiling and laughing often

INTERESTS

- Music
- Watching traffic
- Biking
- Playing pretend kitchen
- Playing with cars

FAVOURITES

- Singing
- Water activities

AGO1

DEVELOPMENT

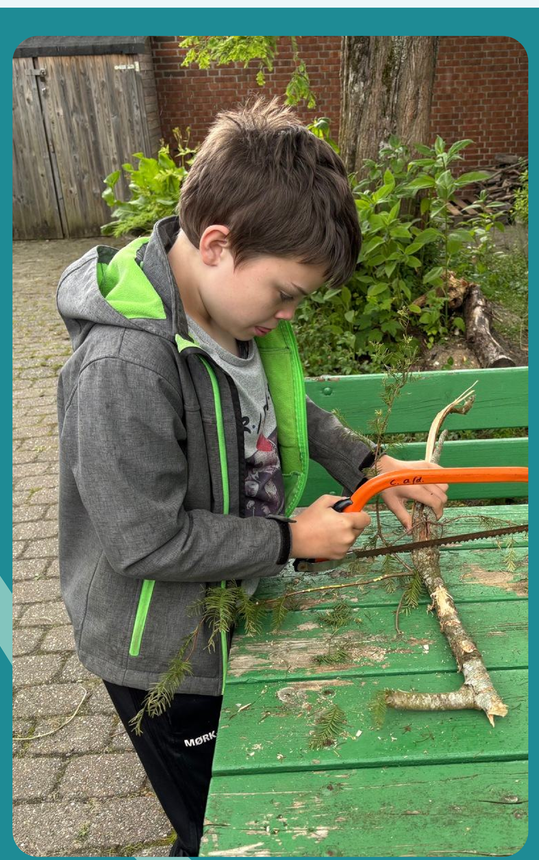
Slowly, he is happy and learning new things, he can ride a bike with put support but cannot understand traffic, he has a non homogenic development as he is further along in some areas than others

THERAPIES

Physical, speech, sense, visual

Jakob is a happy child, and he has the need of constant scaffoling by adults (as you would a small child) exept he is 12 and has 12 years of experience. He is not able to self-regulate, control his impulses or differantiate between safe/dangerous situations. It is all hands on deck, as he is quite the little rascal and loves to interact, tease, smile and make mischief.

In the morning we wake him up, and dress him (he cannot do it himself), get him ready for "school". In the afternoon we typically eat a snack and do some chores or activities Jakob likes, such as loading the waschingmaschine and turning it on or riding his bike. For dinner he needs support and verbal cues for eating with utensils. After dinner we make activities, play or watch ipad and right before bed we sing along to music videos on youtube and play guitar. He is tucked in and we stay with him til he sleeps. He usually wakes 1-3 times during the night.



AGO