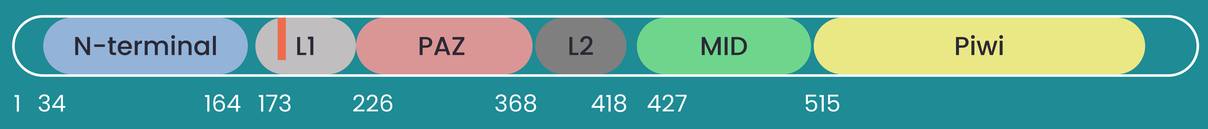
ALBERT

Mutations:
de novo AGO1, p.(Phe180del)



de novo PP2R5E p.(Glu191Lys) - uncharacterised

Age	6 years
Family	Lives with parents and younger brother (4.5) and younger sister (9 months)
Age of Diagnosis	Autism at age 13 months

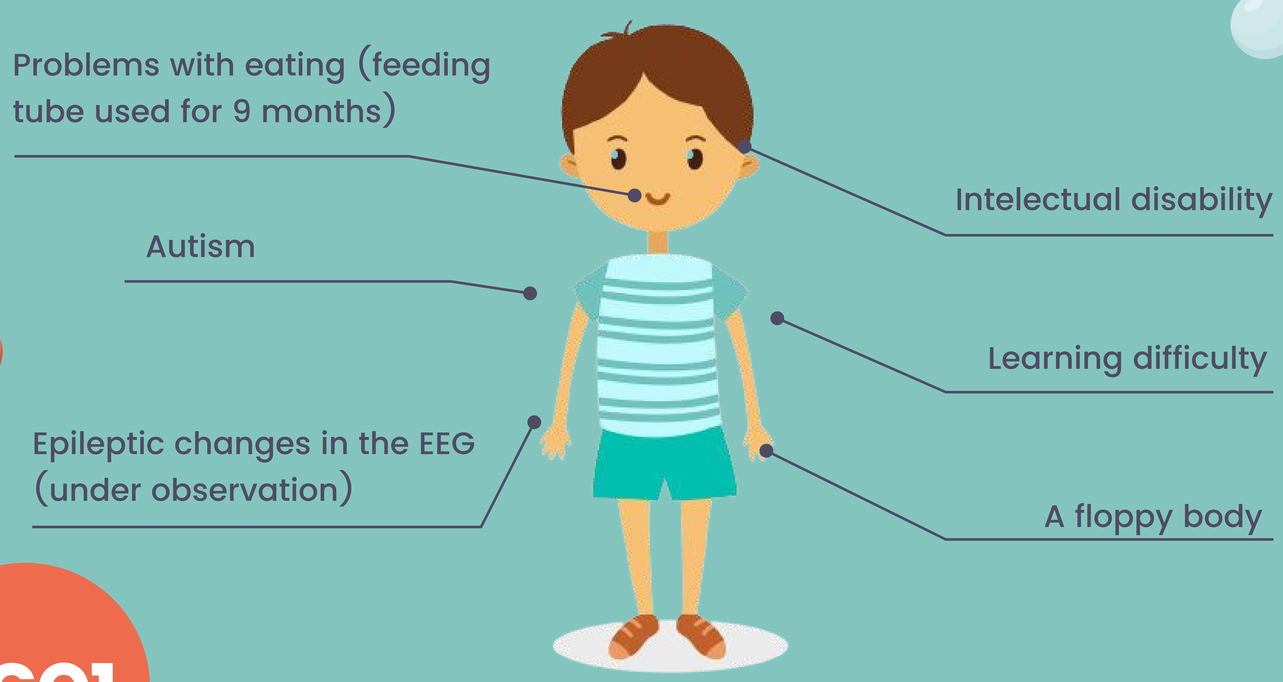
INTERESTS

- Songs
- Running
- Rotating objects
- Swing
- Splashing in water
- Train

FAVOURITES

Toy: train

Song: Wheels of the bus



AGO1

DEVELOPMENT

Nonverbal, attending the special needs pre-school
Mental age: 1.5 year old
Physical age: 3.5 years old

THERAPIES

- Physiotherapy from month 11 due to difficulties in sitting and crawling
- Behavioural therapies from age 13 months

Albert has feeding difficulties which led him to receive feeding tube for 9 months. Currently we are in the process of reintroducing food for last 8 months. We started off with jars for 6-months old kids. He has 5 meals a day which can take up to 6.5h a day.

He continuously develops new behaviours to protest food - covering his mouth, standing up at the chair, running away, turning around, laughing for extended periods. Everyday is a new challenge, but we have managed to provide him with enough calories in meals to remove the feeding tube. Currently, he eats jars for 8-months old kids and one solid meal himself.

AGO