UXUE

Mutations:
de novo AGO1 c.539_541delTCT, p.(Phe180del)

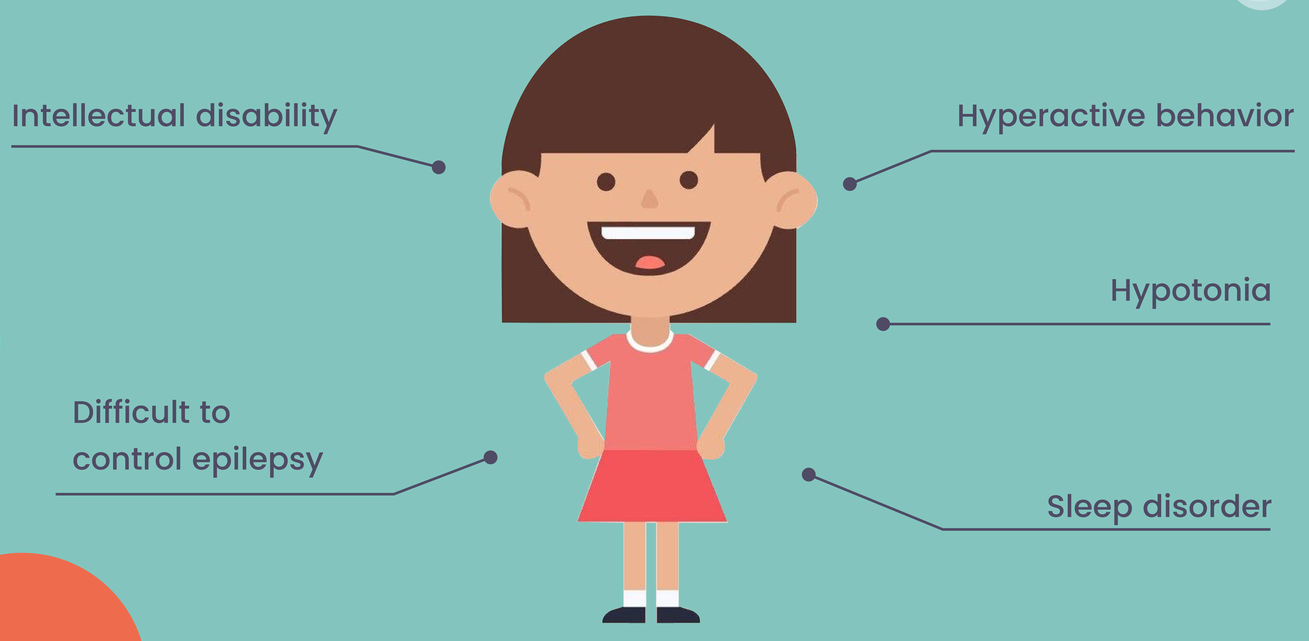






Looking in the mirror, tickling, Watching the kids go down the slide, watching his brother chase him (play tag) across the couches at home, watching the automatic doors go up and down (the garage door for example) make me smile

Age	5 years old
Family	1 sibling (2 years old) and another brother or sister on the way.
Age of Diagnosis	1 year of age: Global Developmental Delay 2 years old: Autism Spectrum Disorder grade 3 3 years and 2 months: Mutation in AGO1 gene



AGO1

DEVELOPMENT

She is non-verbal, however, she "sings" songs (hums them) making verbal approximations, intoning the songs very well. Uxue also makes verbal approximations to colors and numbers (she counts up to 25). Her comprehension is very good (she understands most things). No sphincter control. Disability 69%. Severe dependency

Mental age: 2 years old

THERAPIES

- Global stimulation: 2 - 2,5 years
- Speech therapist: 4.5 years

INTERESTS

- Swinging
- Swimming
- Singing
- Watching TV with songs
- Looking in the mirror

FAVOURITES

- Game: Musical tree
- Watching the singing games on TV







Nocturnal epileptic seizures of 30 minutes duration is the main everyday story/struggle.

AGO