ELIZA

Mutations:
de novo AGO1 c.1073A>G, p.(Gln358Arg).

A new book, a silly face, playing hide and seek, and seeing the people I love make me smile

Age	3 years old
Family	1 younger brother
Age of Diagnosis	AGO1 at 2 years old

INTERESTS

- Books
- Outdors
- Water play
- Singing
- Active play

FAVOURITES

- Reading
- Singing

AGO1

DEVELOPMENT

Speech - near age appropriate, 30 month age equivalent;
Fine Motor Skills - poor, 10-13 months age equivalent;
Gross Motor Skills - poor, 15-18 months age equivalent

THERAPIES

- Physical Therapy - Ongoing, started at 10 months old
- Occupational Therapy - Ongoing, started at 10 months old
- Speech Therapy - Ongoing, started at 19 months old

Eliza can get complex febrile seizures from a low-grade fever that can last over 20 minutes, with focal convulsions. We are constantly fearful of minor/common illnesses. One of her seizures happened late at night, with no vocalization and we caught it by chance. We worry that she will have another silent seizure, and because there is no way to monitor it, she sleeps in the same room with us.

AGO