| Case          | This study                  | Alazami et al, El-Hattab et al | Shankia et al | Salehi Chaleshtori et al | Thomas et al | Total |
|--------------|-----------------------------|---------------------------------|---------------|--------------------------|--------------|-------|
| Detected variants ADAT3 (NM_138422.3) | c.587C>T; p.(Ala196Val) c.820C>T; p.(Glu274*) | c.430G>A; p.Val144Met | c.430G>A; p.Val144Met | c.99_106dup; p.Glu36Glyfs*44 | c.587C>T; p.(Ala196Val) c.586_587delinsTT; p.Ala196Leu |

**Age (years) / Gender**

| | 13 y / M | 9 y / F | 4 y / F | M/F | F/M |
|---|----------|---------|---------|-----|-----|

**Neurology**

| Intellectual disability | Severe | Severe | Severe | 39/39 | 2/2 | M/F | 2/2 | 47/47 (100%) |
|-------------------------|--------|--------|--------|-------|-----|-----|-----|--------------|
| Motor delay             | +      | +      | -      | +     | 1/1 | nd  | 1/2 | 19/21 (90%)  |
| Absent speech/limited speech | -      | -      | -      | +     | 1/3 | 1/1 | Speech delay | 2/2 | 22/43 (51%)  |

**Behavioural problems**

| Deep disturbances | Deep disturbances | Deep disturbances | Aggressive/ hyperactivity | nd | ADHD | - |
|-------------------|-------------------|-------------------|--------------------------|-----|------|---|

**feeding difficulties**

| - | + | + | - | + | + | - | + | + | - | + | + | - | - | - |

**Spasticity**

| Lower limbs | Lower limbs | 13/39 | - | - | 1/1 | 1/2 | 16/45 (36%) |

**Gait ataxia**

| 13/39 | 2/2 | nd | 1/2 | 22/43 (51%) |

**MRI findings**

| Increased signal intensity around hippocampus | White matter abnormalities, wide ventricles, arachnoidal cyst | - | 18/28 | 2/2 | nd | - | 21/34 (62%) |

**Growth parameters**

| Short stature | + | + | + | + | 11/15 | 2/2 | nd | 2/2 | 18/22 (82%) |

**Microcephaly**

| - | - | + | + | 22/39 | 2/2 | + | 1/2 | 27/47 (57%) |

**Facial features/dysmorphology**

| Forehead | High, Broad | - | - | 10/15 | Prominent/High | nd | High | 15/21 (71%) |
|----------|-------------|---|---|-------|-----------------|-----|-----|--------|
| Proptosis | + | + | - | 3/15 | 2/2 | nd | 1/2 | 9/22 (41%) |
| Epicantus | - | - | + | 6/15 | 2/2 | nd | 1/2 | 10/22 (45%) |
| Strabismus | Intermittent | Intermittent | Intermittent | Intermittent | 32/39 | 2/2 | - | 2/2 | 39/47 (83%) |
| Eyes, other | Deeply set eyes, anisocoria | Deeply set eyes, hyperopia | Deeply set eyes, narrow palpebral fissures | - | 7/15 | Slanted palpebral fissures | nd | |
| Ears | Squared superior portion of helix, short earlobe | Squared superior portion of helix, short earlobe | Squared superior portion of helix, short earlobe | - | - | - | - | - | - | - |
| Nose | High nasal bridge, notched ala nasea, low hanging columella | Notched ala nasea, low hanging columella | Notched ala nasea, low hanging columella | 8/15 | Prominent nose | Depressed nasal bridge | Bulbous nasal tip | |
| philtrum | Short, deep | Short, deep | Short, deep | Short, deep | nd | nd | nd | nd | 2/2 | (Long, smooth) |

**Prominent upper front teeth**

| + | + | - | Teeth abnormalities |

**Teething defects**

| TOP, VSD | VSD, PDA | |

**Other**

| IgA deficiency, no forceps grip | IgA deficiency, no forceps grip | Metopic craniosynostosis, constipation, high palate, vomiting, IgA deficiency | Plagiocephaly, synophrysis, wide mouth, full lips, high arched palate, elongated face | Torticollis, myopia, clinodactyly, flat feet, gastroesophageal reflux disease |

### Abbreviations: nd=not determined, PDA=patent ductus arteriosus, SD= standard deviation, VSD=tetralogy of fallot, VSD=ventricular septal defect, y=years

+ present, - absent

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| Case | Age (years) | Gender | Intellectual disability | Motor delay | Absent speech/limited speech | Hypotonia | Feeding difficulties | Spasticity | Social delays | MRI findings | Growth parameters | Facial features/dysmorphology | Other |
|------|-------------|--------|-------------------------|-------------|-----------------------------|-----------|---------------------|------------|-----------------|-------------|-----------------------|---------------------------|-------|
| II:1 | 13 y        | M      | Severe                  | +           | +                           | +         | +                   | +          | nd              | Increased signal intensity around hippocampus | +                | +                | IgA deficiency, no forceps grip |
| II:2 | 9 y         | F      | Severe                  | +           | +                           | +         | +                   | -          | nd              | White matter abnormalities, wide ventricles, arachnoidal cyst | -                | -                | IgA deficiency, no forceps grip |
| II:3 | 4 y         | F      | Severe                  | +           | +                           | +         | +                   | +          | nd              | -           | +                    | +                | +                | IgA deficiency, no forceps grip |

### References:

- Alazami et al.
- El-Hattab et al.
- Shankia et al.
- Salehi Chaleshtori et al.
- Thomas et al.
- Alazami et al, El-Hattab et al.