

Case	This study			Alazami et al, El-Hattab AY. et al	Sharkia et al	Salehi Chaleshtori et al	Thomas et al	Total
	II:1	II:2	II:3					
Detcted variants ADAT3 [NM_138422.3]	c.587C>T; p.(Ala196Val) c.820C>T; p.(Gln274*)			c.430G>A; p.Val144Met Ψ, §	c.430G>A; p.Val144Met Ψ	c.99_106dup; p.Glu36Glyfs*44 Ψ	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	F/M	
<b>Neurology</b>								
Intellectual disability	Severe	Severe	Severe	39/39	2/2	Mild	2/2	47/47 (100%)
Motor delay	+	+	+	13/15	1/1	nd	2/2	19/21 (90%)
Absent speech/limited speech	+	+	+	13/15	1/1	Speech delay	2/2	19/21 (90%)
Hypotonia	+	+	nd	16/36	2/2	nd	2/2	22/43 (51%)
Behavioural problems	Sleep disturbances	Sleep disturbances	Sleep disturbances	Aggressive/ hyperactivity	nd	ADHD	-	
Feeding difficulties	+	-	+		+	nd	2/2	
Spasticity	-	Lower limbs	Lower limbs	13/39	-	nd	1/2	16/45 (36%)
Gait ataxia	+	+	nd		nd			
Gait difficulties	+	+	nd		2/2		nd	
Seizures/Epilepsy (age of onset)	Absense (2-3 y)	Initially primary focal secondary generalized (2.5y), later absense (3y)	Absense (3 y)	6/39	-	nd	-	9/44 (20%)
MRI findings	Increased signal intensity around hippocampus	White matter abnormalities, wide ventricles, arachnoidal cyst	-	18/28	2/2	nd	-	21/34 (62%)
<b>Growth parameters</b>								
Short stature	+	+	+	11/15	2/2	nd	2/2	18/22 (82%)
Microcephaly	-	-	+	22/39	2/2	+	1/2	27/47 (57%)
<b>Facial features/dysmorphology</b>								
Forehead	--	High, Broad	-	10/15	Prominent/High	nd	High	15/21 (71%)
Hypertelorism	+	+	-	3/15	2/2	nd	2/2	9/22 (41%)
Epicanthus	-	-	+	6/15	2/2	nd	1/2	10/22 (45%)
Strabismus	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	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 alae nasea, low hanging columella	Notched alae nasea, low hanging columella	High nasal bridge, notched alae nasea, low hanging columella	8/15	Prominent nose	Depressed nasal bridge	Bulbous nasal tip	
Philtrum	Short, deep	Short, deep	Short, deep	nd	nd	nd	2/2 (Long, smooth)	
Prominent upper front teeth	+	+	+		Teeth abnormalities			
<b>Other</b>								
Tapering fingers	+	+	+					
Cardiac defects	TOF, VSD	-	VSD, PDA					
Other	IgA deficiency, no forceps grip	IgA deficiency	Metopic craniosynostosis, constipation, high palate, vomiting, IgA deficiency		Plagiocephaly, synophrysis, wide mouth, full lips, high arched palate, elongated face		Torticollis, myopia, clinodactyli, flat feet, gastroesophageal reflux disease	

Ψ homozygous variant

§ c.382G>A; p.Val128Met in NM\_138422.1

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

+ present, - absent