Supplementary Table 1. Developmental profiles of patients with *IQSEC2*-related encephalopathy

Patient	Gross motor	Fine motor/hand function	Language	Cognitive/adaptive function	Developmental/cognitive assessment	ASD features	Regression	Development before seizure onset
1	Independent walking achieved; motor dysfunction present	NA	NA	Severe intellectual disability	K-WISC at 6 years 2 months: FSIQ 23; SA 0.74 years, SQ 12.01	No ASD noted	NA	Delayed
2	Unable to walk independently (GMFCS level V)	Limited hand use; able to grasp objects within reach according to developmental assessment	Severely delayed; unclear infantile speech/babbling described	Moderate intellectual disability by FSIQ, with profound adaptive delay	K-WISC at 6 years 11 months: FSIQ 47; SA 0.26 years, SQ 3.75. Bayley-II showed markedly delayed developmental quotient performance	No ASD noted (CARS 9)	NA	Delayed
3	Independent walking achieved; motor developmental delay present	NA	NA	Moderate intellectual disability	K-WPPSI at 3 years 10 months: FSIQ 54; SA 1.25 years, SQ 34.46	ASD features present (CARS 38.5)	NA	Delayed
4	Independent walking achieved	NA	NA	Moderate intellectual disability/ developmental delay	Formal cognitive score unavailable; SA 2.73 years, SQ 48.75. External record described a developmental level of approximately 3-4 years	ASD features present (CARS 31.5)	NA	Delayed
5	Independent walking achieved, approximately 2-year developmental delay	NA	Two- to three-word sentences documented	Moderate intellectual disability/ developmental delay	Formal test score unavailable; medical records estimated IQ at approximately 30-40 and described cognitive functioning at approximately the 3–4-year level at 9 years 10 months	Unknown	NA	NA
6	Independent walking achieved; motor dysfunction present	NA	NA	Severe intellectual disability	Bayley-II at 2 years 5 months: developmental quotient <50; SA 1.21 years, SQ 50.00	ASD features present (CARS 38)	NA	Delayed
7	Independent walking achieved; GMFCS level improved from IV to II	NA	NA	Global developmental delay	Bayley-III at 1 year 6 months: developmental quotient 55; SA 0.56 years, SQ 37.33	Unknown	NA	Delayed
8	Independent walking achieved; motor developmental delay present	Fine motor/adaptive delay documented on Denver developmental assessment	Language delay documented on Denver developmental assessment	Moderate intellectual disability	K-WPPSI-IV at 5 years 11 months: FSIQ 41; SA 3.00 years, SQ 50.68. Denver Developmental Screening Test at 2 years 9 months showed delays in personal-social, fine motor-adaptive, language, and gross motor domains	No ASD noted (CARS 17)	NA	Delayed
9	Unable to walk independently; GMFCS level improved from V to IV	NA	NA	Global developmental delay	Bayley-III at 2 years 2 months: developmental quotient 55; SA 0.35 years, SQ 16.12	ASD present (CARS 27.5)	NA	Delayed
10	Independent walking possible with unsteady/dependent gait	NA	NA	Severe intellectual disability	No formal developmental score available; severe intellectual disability documented	Unknown	NA	Delayed

IQSEC2, IQ motif and Sec7 domain ArfGEF 2; ASD, autism spectrum disorder; NA, not available; K-WISC, Korean Wechsler Intelligence Scale for Children; FSIQ, full-scale intelligence quotient; SA, social age; SQ, social quotient; GMFCS, Gross Motor Function Classification System; CARS, Childhood Autism Rating Scale; K-WPPSI, Korean Wechsler Preschool and Primary Scale of Intelligence.

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