

Supplemental Table S1. Summary of the clinical information of the patients with *PURA*-related neurodevelopmental disorders

	Shimojima et al. 2011	Hosoki et al. 2012	Brown et al. 2013	Bonaglia et al. 2015	Present case	Total in deletions	Lalani et al. 2014	Hunt et al. 2014	Tanaka et al. 2015	Okamoto et al. 2017	Total in nucleotide alterations
Deletion	2/2	3/3	2/2	1/1	1/1	9/9					
Nucleotide alteration											
Truncating							4/11	2/4	2/6	0/1	8/22
Nontruncating							7/11	2/4	4/6	1/1	14/22
De novo inheritance	2/2	3/3	2/2	1/1	1/1	9/9	10/10	4/4	6/6	1/1	21/21
Hypotonia	2/2	3/3	2/2	NA	1/1	8/8	11/11	3/4	NA	1/1	15/16
Feeding difficulties	2/2	3/3	1/2	1/1	1/1	8/9	11/11	3/4	NA	1/1	15/16
Respiratory difficulties	1/2	3/3	2/2	1/1	1/1	8/9	10/10	3/4	NA	1/1	14/15
Seizures	1/2	1/3	0/2	1/1	1/1	4/9	9/11	1/4	1/5	0/1	11/21
Seizure onset	12m	4y	NA	3y	12m		3w-4y	14m	NA		
Seizure type	LGS	tonic	NA	drop attack/ hemiclonic/ tonic	infantile spasms		myoclonie/ tonic clonic/ LGS	epileptic spasms	infantile spasms/ myoclonie	NA	
Abnormal EEG	2/2	2/3	1/2	1/1	1/1	7/9	7/8	2/4	1/5	1/1	11/18
Psychomotor delay	2/2	3/3	2/2	1/1	1/1	9/9	11/11	4/4	6/6	1/1	22/22
Nonverbal	2/2	3/3	2/2	1/1	1/1	9/9	8/8	1/4	4/6	1/1	14/19
Ambulatory	0/2	0/3	2/2	0/1	0/1	2/9	1/8	2/4	4/6	0/1	7/19
Distinctive facial features	2/2	3/3	2/2	1/1	1/1	9/9	9/11	4/4	NA	1/1	14/16
Delayed myelination	2/2	2/3	2/2	NA	1/1	7/8	NA	2/4	1/6	0/1	3/11

NA, not available; w, weeks; y, years; w, weeks; LGS, Lennox-Gastaut syndrome