資料 8　レジストリ横断登録 2110 例のまとめ
（レジストリ症例から特発性全般てんかんと自然終息性てんかんを除いた）

Table 1
Demographics and clinical features of 2110 cases registered in Epilepsy Syndrome Registry.

Gender; n	Female 1021, male 1089
Ages (range, median); years	Age at registration, 0-85, 17; age at seizure onset, 0-83, 3
Main seizure	Focal impaired awareness seizure 723 (34.3%), spasm 323 (15.3%), bilateral tonic-clonic seizure 310 (14.7%), focal aware seizure 292 (13.8%), tonic 198 (9.4%), gelastic 76 (3.6%), myoclonus 55 (2.6%), clonic 35 (1.7%), absence 27 (1.3%), atonic 12 (0.6%), nonconvulsive status 19 (0.9%), convulsive status 14 (0.5%), others 16
Seizure type	Single 954 (45.2%), multiple 1156 (54.8%)
Frequency of main seizure	Daily 578 (27.4%), weekly 356 (16.9%), monthly 395 (18.7%), yearly 266 (12.6%), less than yearly or disappeared 515 (24.4%)
Neurological findings	No neurological findings 1391 (65.9%); hemiparesis 146 (6.9%), diplegia 21 (1.0%), quadriplegia 176 (8.3%), ataxia 121 (5.7%), involuntary movement 66 (3.1%), dysphagia 86 (4.1%), others 105 (overlapping) (5.0%), bedridden 205 (9.7%), no head control 84 (4.0%), artificial respiration 12 (0.6%), unknown 20
Intellectual impairment*	Not impaired 843 (40.0%); mild 334 (15.8%), moderate 248 (11.8%), severe 633 (30.0%), unknown 52
Neuropsychiatric findings	ASD 355 (16.8%), ADHD 39 (1.8%), memory disturbance 104 (4.9%), aphasia 17 (0.8%), executive dysfunction 46 (2.2%), other cognitive dysfunctions 28 (1.3%); delusion-hallucination 45 (2.1%), affective disorder 53 (2.5%), personality disorder 76 (3.6%), sleep disturbance 49 (2.3%), other psychiatric symptoms 38 (1.8%)
Laboratory testing	Gene mutation 228/301 (75.7%); SCN1A (71 cases), PCDH19 (9), CDKL5 (8), KCNT1 (6), STXBP1 (6) and others Cytogenetic abnormality 104/332 (31.3%): chromosomes 21 (23 cases), 15 (23), 20 (16), 1 (6) and others
EEG	Normal 159 (7.5%); suppression-burst 22 (1.0%), hypsarrhythmia 148 (7.0%), generalized spike-waves 292 (13.8%), CSWS 19 (0.9%), focal spikes 892 (42.3%), multifocal spikes 360 (17.1%), other paroxysms 88 (4.2%), rapid/fast rhythm 67 (3.2%), abnormal background activity 308 (14.6%); information not available 32

CT/MRI	Abnormal 1290 (61.1%) (bilateral 363); information not available 43
Etiology	Malformation of cortical development 264 (12.5%), tumor 176 (8.3%), neurocutaneous syndrome 117 (5.5%), infection 76 (3.6%), hypoxic-ischemic encephalopathy 69 (3.3%), cerebrovascular disorder 50 (2.4%), trauma 34 (1.6%), immune-mediated disorder 32 (1.5%), metabolic 25 (1.2%), degenerative disorder 18 (0.9%), others 486 (23.0%), unknown 767 (36.4%)
Therapy	Drug 2031 (96.3%), hormone (ACTH, steroid) 349 (16.5%), diet 65 (3.1%), surgery 547 (25.9%) (resection 327, hemisherotomy/-rectomy 37, callosotomy 102, stereotactic surgery 69, vagus nerve stimulation 68, other 12: multiple surgery 60); no previous treatment 39
Social status	Preschool 476 (22.6%), school 642 (30.4%) (school for disabled 420), employed 428 (20.3%) (employment for disabled 105), housekeeping 126 (6.0%), job training 44 (2.1%), job seeking 187 (8.9%), in need of life care 167 (7.9%) [150/956 (15.7%) of those aged ≥ 20 years]

ASD, autism spectrum disorder; ADHD, attention deficit hyperactivity disorder; CSWS, continuous spike waves during sleep; diet, ketogenic diet therapy

* Intellectual impairment: mild, IQ/DQ between 50-69; moderate, IQ/DQ between 35-49; severe, IQ/DQ less than 35. Assessed by the test results in 969 cases, otherwise from the information of caregivers.

Table 2. Seizure characteristics and associated symptoms of patients with various epileptic syndromes.

	n	Gender (female /male)	Age at registration (range, median)	Age at seizure onset (range, median)	Type of main seizure: n (%)	A: Frequency of main seizure: ≥ monthly n (%)	Multiple seizure type: n (%)	Therapy other than ASM: n (%)	B: Intellectual impairment*: n (%)	C: Prominent neurologic/ psychiatric symptoms: n (%)	Either A or B/C or both: n (%)
Dravet syndrome	89	47/42	0-38 (10)	0	GTC 51 (57.3), focal 18 (20.2)	67 (75.3)	71 (79.8)	Diet 8 (9.0)	75 (84.3)	Ataxia 43 (48.3), ASD 45 (50.6)	86 (96.6)
Epilepsy of infancy with migrating focal seizures	15	10/5	0-15 (3)	0-2 (0)	Focal 7 (46.7), tonic 6 (40)	13 (86.7)	13 (86.7)	Surgery 2 (13.3)	15 (100)	Bedridden 10 (66.7)	15 (100)
Epilepsy with myoclonic atonic seizures	11	3/8	3-15 (6)	0-5 (3)	Myoclonic atonic 11 (100)	7 (63.6)	9 (81.8)	Diet 4 (36.4)	6 (54.5)	ASD 4 (36.4)	9 (81.8)
Epileptic encephalopathy with CSWS	36	11/25	4-19 (9)	0-7 (3)	Focal 21 (58.3), absence 8 (22.2)	12 (33.3)	28 (77.8)	Surgery 6 (16.7)	26 (72.2)	ASD 16 (44.4), paresis 10 (27.8)	33 (91.7)
Lennox-Gastaut syndrome	85	50/35	5-50 (17)	0-11 (2)	Tonic 58 (68.2),	75 (88.2)	80 (94.1)	Hormone 29 (34.1), surgery 37 (43.5)	84 (98.8)	ASD 22 (25.6), paresis 17 (20.0)	85 (100)
Ohtahara syndrome	24	8/16	0-19 (4)	0	Tonic 11 (45.8), spasm 8 (33.3)	17 (70.8)	11 (45.8)	Surgery 10 (41.7)	22 (91.7)	Bedridden 14 (58.3)	24 (100)

Progressive myoclonus epilepsy	31	17/14	4-80 (33)	1-52 (12)	Myoclonus 16 (51.6), GTC 11 (35.5)	20 (64.5)	20 (64.5)	0thers 2 (6.5)	17 (54.8)	Involuntary movement 17 (54.8), ataxia 11 (35.5),	29 (93.5)
West syndrome including Aicardi syndrome	303	147/156	0-51 (3)	0-2 (0)	Spasm 263 (86.8),	226 (74.6)	137 (45.2)	Hormone 217 (71.6), surgery 48 (16.0)	259 (85.5)	Bedridden 108 (35.6), ASD 40 (13.2)	289 (95.4)

ASM, antiseizure medication; ASD, autism spectrum disorder; ADHD, attention deficit hyperactivity disorder; CSWS, continuous spike waves during sleep; Diet, ketogenic diet therapy; GTC, generalized tonic-clonic seizure; Hormone, ACTH or steroid therapy.
* Surgical therapy includes vagus nerve stimulation. ** Impairment: IQ/DQ less than 70.

Table 3. Seizure characteristics and associated symptoms of patients with epilepsy grouped according to etiology.

Etiology		n	Gender (female /male)	Age at registration (range, median)	Age at seizure onset (range, median)	Type of main seizure: n (%)	A: Frequency of main seizure: ≥ monthly n (%)	Multiple seizure types: n (%)	Therapy other than ASM: n (%)	B: Intellectual impairment: n (%)	C: Prominent neurologic/ psychiatric symptoms: n (%)	Either A or B/C or both: n (%)
Structural												
	Focal cortical dysplasia	108	63/45	0-67 (17)	0-42 (3)	Focal 91 (84.3)	69 (63.9)	49 (45.4)	Surgery 53 (49.1)	54 (50.0)	ASD 21 (19.4), paresis 15(13.9)	96 (88.9)
	Neuronal migration disorders	37	21/16	0-48 (18)	0-28 (5)	Focal 29 (78.4)	26 (70.3)	22 (59.5)	Surgery 5 (13.5)	22 (59.5)	Paresis 6 (16.2)	36 (97.3))
	Hypothalamic hamartoma	72	33/39	2-53 (10)	0-10 (0)	Gelastic 72 (100), focal 24 (33.3)	20 (27.8)	48 (66.7)	Surgery 70 (97.2)	25 (34.7)	ADHD 7 (9.7)	42 (58.3)
	Cavernous hemangioma	25	14/11	2-76 (40)	1-73 (24)	Focal 23 (92.0)	15 (60.0)	15 (60.0)	Surgery 9 (36.0)	3 (12.0)	Memory disturbance 3 (12.0)	16 (64.0)
	Dysplastic tumor*	24	11/13	2-52 (20)	0-36 (4)	Focal 24 (100)	9 (37.5)	13 (54.2)	Surgery 19 (79.2)	10 (41.7)	ASD 6 (25.0), memory disturbance 3 (12.5)	17 (70.8)
	Trauma	32	5/27	5-77 (43)	0-73 (18)	Focal 28 (87.5)	12 (37.5)	19 (59.4)	Surgery 7 (21.9)	15 (46.9)	Paresis 12 (37.5) executive dysfunction 5 (15.6)	27 (84.4)

	Hypoxic-ischemic encephalopathy	29	9/20	1-53 (20)	0-20 (2)	Focal 22 (75.9)	22 (75.9)	16 (55.2)	Surgery 2 (6.9)	25 (86.2)	Paresis 14 (48.3), Bedridden 5 (17.2)	29 (100)
	Vascular disorder	41	11/30	6-69 (37)	0-66 (24)	Focal 35 (85.4)	17 (41.5)	18 (43.9)	Surgery 7 (17.1)	13 (31.7)	Paresis 14 (34.1), memory disturbance 7 (17.1)	32 (78.0)
	Mesial temporal lobe epilepsy with hippocampal sclerosis	197	109/88	4-77 (41)	0-75 (11)	Focal 197 (100)	122 (61.9)	116 (58.9)	Surgery 117 (59.4)	38 (19.3)	Memory disturbance 26 (13.2)	153 (77.7)
	Sturge-Weber syndrome	35	18/17	0-60 (5)	0-27 (0)	Focal 22 (62.9)	17 (48.6)	22 (62.9)	Surgery 25 (71.4)	21 (60.0)	Paresis 13 (37.1)	32 (91.4)
	Tuberous sclerosis complex	51	24/27	0-48 (11)	0-16 (0)	Focal 32 (62.7), tonic 9 (17.6)	37 (72.5)	31 (60.8)	Surgery 15 (29.4)	40 (78.4)	ASD 15 (29.4)	48 (94.1)
	Other structural	161	80/81	0-85 (28)	0-83 (9)	Focal 125 (77.6), tonic 18 (11.2)	113 (70.2)	80 (49.7)	Surgery 40 (24.8)	75 (46.6)	Paresis 30 (18.6), bedridden 14 (8.7)	105 (65.2)
Genetic												
	Angelman syndrome	26	11/15	1-41 (10)	0-31 (1)	Tonic 5 (19.2), myoclonus 5 (19.2)	16 (61.5)	12 (46.2)	0	26 (100)	Ataxia 8 (30.8), sleep disturbance 10 (38.5)	26 (100)

	Ring 20 epilepsy syndrome	16	11/5	6-65 (20)	1-14 (7)	Focal 10 (62.5) NCSE 4 (25.0)	16 (100)	11 (68.8)	Surgery 2 (12.5)	10 (62.5)	0	16 (100)
	Rett syndrome	37	37/0	5-44 (14)	0-17 (4)	Focal 12 (32.4), tonic 10 (27.0)	16 (43.2)	10 (27.0)	0	37 (100)	ASD 25 (67.6), sleep disturbance 14 (37.8), bedridden 10 (27.0)	37 (100)
	Other genetic	62	36/26	1-36 (9.5)	0-25 (0)	Focal 28 (45.2), GTC 11 (17.7)	34 (54.8)	33 (53.2)	Hormone 6 (9.7)	57 (91.9)	ASD 22 (35.5), bedridden 12 (19.4)	60 (96.8)
Metabolic												
	Metabolic disorder	17	5/12	1-47 (7)	0-42 (1)	Focal 8 (47.1), tonic 4 (23.5)	10 (58.8)	7 (41.2)	Diet 4 (23.5)	14 (82.3)	Paresis 7 (41.2), bedridden 5 (29.4)	15 (88.2)
Infectious												
	Infectious disease	60	24/36	1-55 (26)	0-38 (7)	Focal 54 (90.0)	45 (75.0)	38 (63.3)	Surgery 12 (20.0)	42 (70.0)	Paresis 13 (21.7)	57 (95.0)
Immune												
	Rasmussen encephalitis	20	9/11	10-48 (19)	0-25 (4)	Focal 20 (100)	19 (95.0)	2 (10.0)	Hormone 11 (55.0), surgery 9 (45.0)	14 (70.0)	Paresis 15 (75.0)	20 (100)

	Other autoimmune disorder	28	18/10	6-75 (41)	1-74 (22)	Focal 26 (92.9)	23(82.1)	16 (57.1)	Immune 9 (32.1), surgery 4 (14.3)	5 (17.9)	Memory disturbance 8 (28.6)	26 (92.9)
Unknown												
	Other focal epilepsy without known etiology	342	155/187	0-85 (27)	0-83 (10)	Focal 333 (97.4)	198 (57.9)	157 (45.9)	Surgery 35 (10.2), hormone 12 (3.5)	128 (37.4)	ASD 68 (19.9), memory disturbance 24 (7.0), paresis 16 (4.7)	267 (78.1)
	Other generalized epilepsy without known etiology	58	26/32	1-63 (20.5)	0-42 (6.5)	GTC 26 (44.8), absence 10 (17.2)	30 (51.7)	28 (48.3)	Hormone 3 (5.5)	27 (46.6)	ASD 10 (17.2)	45 (77.6)
	Unclassified epilepsy without known etiology	27	9/18	0-63 (25)	0-49 (9)	GTC 13 (48.1), focal 6 (22.2)	10 (37.0)	13 (48.1)	Others 2 (7.4)	13 (48.1)	Ataxia 3 (11.1), involuntary movement 3 (11.1)	19 (70.4)

NCSE, nonconvulsive status epilepticus.
*Dysplastic tumor includes dysembryoplastic neuroepithelial tumor and ganglioglioma.

Table 4
Number of patients with seizures occurring less than monthly and without comorbidities

	n	Seizure frequency: yearly n (%)	Seizure frequency: < yearly or none n (%)	Total n(%)
Angelman syndrome	26	0	0	0
Dravet syndrome	89	3	0	3 (3.4)
Epilepsy due to autoimmune disease	28	1	1	2 (7.1)
Epilepsy due to cavernous hemangioma	25	3	6	9 (36.0)
Epilepsy due to dysplastic tumor	24	1	6	7 (29.2)
Epilepsy due to focal cortical dysplasia	108	7	6	13 (12.0)
Epilepsy due to hypothalamic hamartoma	72	0	28	28 (38.9)
Epilepsy due to hypoxic-ischemic encephalopathy	29	0	0	0
Epilepsy due to infectious disease	60	2	1	3 (5.0)
Epilepsy due to metabolic disorders	17	0	2	2 (11.8)
Epilepsy due to neuronal migration disorders	37	1	0	1 (2.7)
Epilepsy due to other genetic disorders	62	2	0	2 (3.2)
Epilepsy due to other structural disorders	161	9	10	19 (11.8)
Epilepsy due to trauma	32	1	4	5 (15.6)
Epilepsy due to vascular disorder	41	3	5	8 (19.5)
Epilepsy of infancy with migrating focal seizures	15	0	0	0
Epilepsy with myoclonic atonic seizures	11	0	2	2 (18.2)
Epileptic encephalopathy with CSWS	36	1	2	3 (8.3)
Lennox-Gastaut syndrome	85	0	0	0
Mesial temporal lobe epilepsy	197	12	27	39 (19.8)
Ohtahara syndrome	24	0	0	0
Other epileptic syndromes	11	1	0	1 (9.1)
Other focal epilepsy without known etiology	342	39	31	70 (20.5)
Other generalized epilepsy without known etiology	58	3	8	11 (19.0)
Progressive myoclonus epilepsy	31	0	2	2 (6.5)

Rasmussen encephalitis	20	0	0	0
Rett syndrome	37	0	0	0
Ring 20-epilepsy syndrome	16	0	0	0
Sturge-Weber syndrome	35	1	2	3 (8.6)
Tuberous sclerosis complex	51	0	3	3 (5.9)
Unclassified epilepsy without known etiology	27	3	5	8 (29.6)
West syndrome incl Aicardi syndrome	303	1	10	11 (3.6)
Total	2110	94	161	255 (12.1)