

On-line Table 1: Clinical data of the sample*

Case	Sex	Onset (mo)	Gene	Microcephaly	Neurologic Presentation	Developmental Delay	Epilepsy	Other Clinical Signs
1	M	Birth	Unknown	+	ST	Severe		Pericarditis
2	M	Birth	AGS2	+	ST	Severe	+	
3	F	Birth	AGS1	+	ST	Severe		
4	F	Birth	AGS4	+	ST	Severe	+	Celiac disease
5	M	Birth	<i>hetAGS4</i>	+	ST	Severe	+	Gastroenterorrhagia
6	F	Birth	AGS2	+	ST	Severe	+	SL
7	M	Birth	Unknown	+	ST	Severe		Interstitial pneumonia
8	M	Birth	AGS2	+	ST	Severe	+	
9	M	Birth	AGS3	+	ST	Severe	+	Hepatosplenomegaly, SL
10	M	Birth	AGS1	+	ST	Severe	+	Glaucoma and gallbladder thickness
11	F	1	AGS2	+	ST	Severe		
12	F	1	AGS2	+	ST	Severe		
13	M	1	AGS1	+	ST	Severe		SL
14	M	1	AGS2		SD	Absent		SL
15	F	1	AGS2	+	ST	Severe	+	
16	M	1	AGS3	+	ST	Severe	+	
17	F	1	AGS2	+	ST	Severe		SL
18	M	1.5	AGS2	+	ST	Severe	+	SL
19	F	2	AGS2	+	ST	Severe	+	
20	F	2	Not done	+	ST	Severe		
21	M	2	AGS2	+	ST	Severe		
22	F	2.5	AGS2	+	ST	Severe	+	SL
23	M	2.5	AGS2	+	ST	Severe	+	SL
24	M	3	AGS2	+	ST	Severe	+	
25	F	3	AGS2	+	ST	Mild		
26	F	3	AGS2	+	ST	Severe		SL
27	F	4	AGS1	+	ST	Severe		
28	M	4	AGS2	+	ST	Severe		
29	F	5	AGS2	+	ST	Severe		
30	M	6	AGS2	+	ST	Severe		
31	F	8	Unknown	+	ST	Severe		Celiac disease, lupus
32	M	10	AGS2	+	ST	Severe		
33	F	11	AGS2	+	ST	Severe	+	SL
34	M	12	AGS2		SD	Absent		
35	F	12	Not done	+	ST	Severe		
36	F	15	AGS2		ST	Severe		Gastroenterorrhagia

Note:—ST indicates spastic-dystonic tetraplegia; SD, spastic diplegia; SL, skin lesions as typically observed in AGS; +, yes; *hetAGS4*, heterozygous mutation in AGS4.

* The patients are listed by age at onset of the disease.

On-line Table 2: Spatial distribution and symmetry of cerebral calcifications*

Case	Onset (mo)	Gene	LE	C	T	D	WM	O	Severity
1	Birth	Unknown	S			S	S		+
2	Birth	<i>AGS2</i>	S		S				+
3	Birth	<i>AGS1</i>					S		++
4	Birth	<i>AGS4</i>	S			S	A R > L		+
5	Birth	<i>hetAGS4</i>	S				S		++
6	Birth	<i>AGS2</i>	S						+
7	Birth	Unknown	A L > R						+
8	Birth	<i>AGS2</i>	S		S	S	S	Cerebral peduncle	++
9	Birth	<i>AGS3</i>	S			S	S		++
10	Birth	<i>AGS1</i>	S		S	S	S		++
11	1	<i>AGS2</i>	S						+
12	1	<i>AGS2</i>	S						+
13	1	<i>AGS1</i>	S				S		++
14	1	<i>AGS2</i>	S			S	S		+
15	1	<i>AGS2</i>	S			S			+
16	1	<i>AGS3</i>	S	S			S		++
17	1	<i>AGS2</i>	S		S		S		+
18	1.5	<i>AGS2</i>	S				S (CR)		+
19	2	<i>AGS2</i>	S		S	S	S (IC)	Brain stem	++
20	2	Not done	S						+
21	2	<i>AGS2</i>	S		S		S		+
22	2.5	<i>AGS2</i>	S			S			+
23	2.5	<i>AGS2</i>	S	S	S		S		+
24	3	<i>AGS2</i>	S				S (CR)		+
25	3	<i>AGS2</i>	S				S		+
26	3	<i>AGS2</i>	S						+
27	4	<i>AGS1</i>	S			S	S		++
28	4	<i>AGS2</i>	A R > L						+
29	5	<i>AGS2</i>	S						+
30	6	<i>AGS2</i>	S						+
31	8	Unknown	S						+
32	10	<i>AGS2</i>	S						+
33	11	<i>AGS2</i>	A R > L						+
34	12	<i>AGS2</i>	S				S		+
35	12	Not done	S			S	S		+
36	15	<i>AGS2</i>	S				S		+

Note:—LE indicates lentiform nucleus; C, caudate; T, thalamus; D, dentate; WM, white matter, O, other location, CR, corona radiata, IC, internal capsule, S, symmetric; A, asymmetric; +, mild-moderate; ++, severe; R, right; L, left.

* The subjects are listed by age at onset of the disease.

On-line Table 3: Spatial distribution and severity of white matter abnormalities*

Case	Onset (mo)	Gene	F	P	T	O	CC	IC/EC	OR	CWM
1	Birth	Unknown	+++	++	+++	++	++	++		
2	Birth	AGS2	+++	++	+++†	++	++	+		+
3	Birth	AGS1	+++†	++	+++†	++				
4	Birth	AGS4	+++	++	+++†	++	++	++		
5	Birth	hetAGS4	++	++	+	+				
6	Birth	AGS2	+++	++	+++	++			+	
7	Birth	Unknown	++	++	++	++	++			
8	Birth	AGS2	+++†	++	+++†	++				
9	Birth	AGS3	+++	++	+++	++				
10	Birth	AGS1	+++†	++	+++†	++	++	+		
11	1	AGS2	+++†	++	+++	++	++	+		
12	1	AGS2	+++†	++	+++	++				
13	1	AGS1	+++	+++	+++†	+++	+++	+++	++	++
14	1	AGS2	++	++	++	++	++	+++		
15	1	AGS2	+++	++	+++	++				
16	1	AGS3	+++†	++	+++†	++				
17	1	AGS2	+++	++	+++†	++				
18	1.5	AGS2	+++	++	+++	++	+	+		+
19	2	AGS2	+++†	+++	+++†	+++	+++	+++	+	
20	2	Not done	++	++	++	++	+	+	+	
21	2	AGS2	++	++	++	++				
22	2.5	AGS2	+++	+	++	+				
23‡	2.5	AGS2	+++†	++	+++†	++	++			
24	3	AGS2	++	++	++	++				
25	3	AGS2	+		+					
26	3	AGS2	+++	++	+++	++				
27	4	AGS1	+++	++	+++	++				
28	4	AGS2	+++	++	+++	++				
29	5	AGS2	+++	++	+++	++				
30	6	AGS2	++	++	++	++				
31	8	Unknown	++	++	++	++			+	
32	10	AGS2	+++	++	+++	++				
33	11	AGS2	+	+	+	+				
34	12	AGS2	++	+	++	+				
35	12	Not done	++	+	+	+			++	
36	15	AGS2	++	++	++	++				

Note:—F indicates frontal; P, parietal; T, temporal; O, occipital; CC, corpus callosum; IC/EC, internal capsule, external and extreme capsule; OR, optic radiation; CWM, cerebellar white matter; +, mild signal intensity alteration; ++, moderate signal intensity alteration; +++, severe signal intensity alteration.

* The subjects are listed by age at onset of the disease.

† Presence of cystic areas.

‡ The white matter abnormalities were asymmetric in this patient only.

On-line Table 4: Neuroradiologic follow-up of 19 subjects with AGS*

Case	Age at First Exam	Exams	0–1 Years			1–2 Years			2–4 Years			4–10 Years		
			C	L	A	C	L	A	C	L	A	C	L	A
1	13 mo	2 MRI, 2 CT			S	S	S							
8	5 yr	1 MRI, 4 CT										S	S	S
9	0.5 mo	1 MRI, 2 CT			S	S	S							
10	2 mo	2 MRI, 2 CT			S	W	W							
13	2 mo	2 MRI, 1 CT	S	W	W									
14	3 mo	1 MRI, 2 CT				S	S	S						
15	6 mo	3 MRI, 1 CT			S	S	S							
17	3 mo	3 MRI, 1 CT	W	S	S							S	S	S
18	8 mo	3 MRI, 2 CT				S	S	S				S	S	I
19	3 mo	2 MRI, 1 CT	S	W	W									
21	7 yr	1 MRI, 4 CT										S	S	W
22	5 mo	2 MRI, 2 CT										S	S	S
23	4 mo	3 MRI, 1 CT	W	S	S							S	S	W
25	12 mo	2 MRI, 1 CT				S	S	S						
27	6 mo	1 MRI, 2 CT	W	S	S							S	S	S
28	7 mo	5 MRI, 3 CT	S	S	W							S	S	I
30	6 mo	1 MRI, 1 CT	S	S	S									
32	9 mo	2 MRI, 1 CT	S	S	S									
36	17 mo	2 MRI, 2 CT				Wa	W	W						

Note:—MRI indicates MR imaging; C, calcifications; L, leukoencephalopathy; A, cerebral atrophy; W, worsened; Wa, first appearance; S, stable; I, improved.

* The subjects are listed by age at onset of the disease.