

On-line Table 1: Clinical and demographic data																			
Patient	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
Sex	M	F	M	F	M	F	M	M	F	M	M	M	M	F	M	M	M	F	F
Age at last examination	15	28	53	2	1	6	7	8	9	13	16	4 m	1	2	3	5	3	2	2
Age at diagnosis	10	26	57	4	5	3	0	6	2	4	6	2	1	1	3	4	2	1	1
Onset symptom	SPR Ataxia		SPR	Microc PMR	Cataract	Microc.	Microc. PMR	PMR Hypotony	PMR Hypotony	Microc. PMR	Microc. Cataract PMR	Microc. Cataract PMR	Microc. Cataract PMR	Microc. Cataract PMR	Microc. Cataract PMR	Microc. IUGR	Arthrog.	Microc. Arthrog. PMR	
Age at onset	2	4	childhood	0		14 m	0	3 m	9 m	2 m	6 m	0	0	0	0	0	0	1 m	0
Age at death			57	5	10							4				7	3	2	
CS group	III	III	III	I	I	I	I	I	I	I	I	II	II	II	II	II	II	COFS	COFS
Genetic mutation	CSB	CSB	CSB	CSB	CSB	CSB	CSA	CSA	CSA	CSA	CSA	CSB	CSB	CSB	CSB	CSB	CSB	CSB	CSB

Note:—M indicates male; F, female; SPR, staturo-ponderal retardation; Microc, microcephaly; PMR, psychomotor retardation; Microph, microphthalmia; Arthrog, arthrogryposis; CSA and CSB, genes mutated in Cockayne Syndrome; age: years except m, months; IUGR, intrauterine growth retardation.

On-line Table 2: CT and MR results

Patient	1		2		3		4		5		6		7		8		9		10		11		12		13		14		15		16		17		18		19	
Cockayne subtype	CSIII		CSIII		CSIII		CSI		CSI		CSI		CSI		CSI		CSI		CSI		CSI		CSII		CSII		CSII		CSII		CSII		CSII		COFS		COFS	
Age at examination	6	15	28	44	53	3 m	2	1	6	7	8	5	9	4	13	16	4 m	1	2	3	5	6 m	3	3 m	2	9 m	2											
Type of exam	M	C/M	C/M	C	M	M	C	C/M	C/M	C/M	C/M	M	C	M	C/M	C/M	C/M	C/M	C/M	C/M	C/M	M	M	C/M	M	C/M	C/M											
Calcifications																																						
Putamen		++		+++		+	++	+++	+	+++	+	+++		++	++	+++		++	++	++		+++	+++		+++		+++		++		++		++		++			
Pallidum			+						+							+		+++	++				+++				+++							++				
Cortex										++																												
WM																																						
Dentate nuclei			+	+++																																		
Caudate				++							+																											
Thalamus																																				+		
Leptomeningeal vessels																																						
Atrophy/hypoplasia																																						
Brain stem	+	++	+++		++	-	-	-	+	+++	++	+++	++	++	+++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
Cerebellar hemispheres	+	++	++		+	-	-	-	+	+++	++	+++	++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
Vermis	+	++	++		+	-	-	-	+	+++	++	+++	++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
WM																																						
Cortex	+	++	+++		++	+	+	+	+	+++	++	+++	++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
Corpus callosum	+	++	+++		++	-	-	-	+	+++	++	+++	++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
Ventricle enlargement	+	++	+++		++	+	+	+	+	+++	++	+++	++	++	+++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++	++		
WM signal-intensity																																						
Hemispheric WM																																						
T1WI	m	m	u		NA	u	NA	NA	m	m	m	m	NA	NA	m	m	u	m	NA	m	NA	u	u	u	u	u	u	u	u	u	u	u	m	m	m			
T2WI	u	u	u		u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u			
Tigroid aspect on T2WI	++	++	-		-	+	+	+	+	+	+	+	NA	-	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-			
Corpus Callosum																																						
T1WI	m	m	m/gen u		m	u	m	m	m	m	m	m	m	m	m	m	u	m	m	m	m	u	u	u	u	u	u	u	u	u	u	m/gen u	m	m	m			
T2WI	m	m	m/gen u		m	u	m	m	m	m	m	m	m	m	m	m	u	m	m	m	m	u	u	u	u	u	u	u	u	u	m/gen u	m	m	m				
Internal Capsule																																						
Posterior limb																																						
T1WI	m	m	m		NA	m	NA	NA	m	m	m	m	NA	NA	m	m	m	u	NA	m	NA	m	u	u	u	m	m	m	m	m	m	m	m	m	m			
T2WI	m	m	m		m	m	m	m	m	m	m	m	NA	NA	m	m	m	u	u	u	u	m	u	u	u	m	m	m	m	m	m	m	m	m				
Anterior limb																																						
T1WI	m	m	m		NA	m	NA	NA	m	m	m	m	NA	NA	m	u	u	u	NA	m	NA	u	u	u	u	u	m	m	m	m	m	m	m	m	m			
T2WI	m	m	u		u	u	u	u	u	u	u	u	NA	NA	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u	u			
1H-MR Spectroscopy																																						
WM																																						
NAA/Cr	↓	↓	↓			↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓			
Cho/Cr	↑	↑	↑			↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑			
Lactate	+	+	+			+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+			
Gray Matter																																						
NAA/Cr	↓	↓	↓			↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓	↓			
Cho/Cr	↑	↑	↑			↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑	↑			
Lactate	+	+	+			+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+			

Note—Age indicates year, except m, months; C, CT scan; M, MR imaging; m, myelinated; u, unmyelinated; NA, not available or not analyzable; m/gen u, corpus callosum myelinated except the unmyelinated genu; -, absence; +/++/+/+++, grading scale for calcifications, atrophy, and signal-intensity abnormalities (refer to text); WM, white matter; ¹H-MR spectroscopy results: =, equal; ↓, reduced; +, lactate presence; T1WI, T1-weighted imaging; T2WI, T2-weighted imaging; NAA/Cr and Cho/Cr were calculated relative to healthy control subjects.