

Table e-1.																																																												
Family ID	CCA-184				CCA-385				CCA-386	CCA-388		CCA-389		CCA-405		CCA-436	CCA-516	CCA-523		CCA-531	CCA-532	CCA-542			CCA-543	CCA-557	CCA-572	CCA-590	CCA-611	CCA-635	CCA-636																													
Family Data																																																												
Age at MRI	18 yrs	14 yrs	11 mo	2 mo	1yr	5 yrs	20 mo	1 mo	14 mo	7 yrs	7 yrs	2 yrs	4 yrs	9 yrs	5 yrs	3 yrs	2 yrs	4 yrs	16 yrs	14 yrs	2 yrs	3 yrs	11 yrs	10 yrs	8 mo	unk	5 yrs	1 mo	2 yrs	2 yrs																														
Age at onset of seizures	NA	NA	NA	NA	NA	NA	NA	?	NA	NA	NA	NA	?	18mos	?	NA	NA	NA	?	?	?	?	?	?	?	?	?	?	?	?																														
Age at onset of symptoms	birth	birth	birth	birth	3days	birth	NA	birth	2years	8mos	8mos	?	?	18mos	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?																														
Consanguineous	+				+				+				+				+				+				+																																			
Gender	M	M	M	M	M	M	M	F	F	F	M	M	F	M	M	M	F	F	F	F	M	F	F	M	F	M	M	F	M	M																														
Multiplex	+				+				+				+				+				+				+																																			
Number of affecteds	6				2				3				2				1				3				2				7				3				1				2				1				2				1				2			
Number Pts w/ MRIs	4				2				1				1				2				2				1				3				1				2				1				1				1				2							
Pt Pedigree ID	IV-I-2	IV-I-4	IV-I-9	IV-I-7	IV-I-5	IV-II-7	IV-8	II-III-2	III-4	III-1	III-2	III-2	III-1	IV-5	IV-6	IV-I-1	IV-II-2	III-4	III-3	III-5	III-9	V-I-3	III-1	III-2	III-1	III-2	III-1	III-2	III-1	III-1	III-1	III-1	III-1	III-1	III-1	III-1	III-1	III-1																						
MRI Findings																																																												
Anterior commissure	+																																																											
CCA Type	HwoD	HwoD	CAG	APC	HwoD	HwoD	KIN	STR	CAG	STR	STR	APC	STR	KIN	KIN	KIN	CAG	KIN	APC	HwoD	CAG	ANR	APC	CAG	ANR	APC	HwoD	KIN	STR	STR																														
Cerebellar affection	-	-	-	-	+ CVH	+ CVH	-	+	+PanCA	-	-	+	-	-	-	-	+ CVH	+ CVH	-	+ CVH	+ CVH	+ CVH	-	+ PCH	+ PCH	+ CVH	-	-	-	+PCH	+ PCH																													
Colpocephaly	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Cysts (CC or Cb)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
GMH	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Ventriculomegaly	+	+	+	+	+	-	-	-	-	-	-	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Lipomas	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Microcephaly	-	-	-	-	-	-	-	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
ON Abnormalities	-	-	-	-	+Col	+Col	-	-	+ ONA	+ ONA	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Pituitary Hypoplasia	-	-	-	-	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Probst Bundle (PB)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
PMG	-	-	-	-	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Septum Pellucidum Abnl	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
WMD	-	-	-	-	-	-	-	-	-	+	+	+	+	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Clinical Findings																																																												
CV disease	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Dysmorphic Facies	+	+	+	+	-	-	-	+	-	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Endocrine disease	-	-	-	-	+GH Def	+GH Def	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Hypotonia/Flacidty	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
MR/DD	+MLD	+MLD	+MLD	+MLD	-	-	+	-	-	+	+	-	-	+	+	-	-	-	+SEV	+SEV	+SEV	+	-	-	-	-	-	-	-	-	-																													
Respiratory abnormalities	-	-	-	-	+	-	-	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Seizures	-	-	-	-	-	-	-	+	-	-	-	-	+	+	+	-	-	-	+ABS	+ NCtrl	+ABS	-	-	-	-	-	-	-	-	-	-																													
Skeletal anomalies	+MED	+MED	+MED	+MED	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
SOD	-	-	-	-	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Spasticity	-	-	-	-	-	-	+	+SEV	-	+MOD	-	+	+	+	+	+	+	+	-	-	-	-	-	-	-	-	-	-	-	-	-																													
Visual abnormalities	-	-	-	-	-	+	-	+ CBLND	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+VEP Abnl	+VEP Abnl	-	-	-	-	-	-	-																													

Legend: CCA types: HwoD=Hypoplasia without dysplasia, APC=Apple Core CC, ANR=Anterior Remnant CC, STR=Stripe CC, KIN=Kinked CC, CAG=Complete absence of CC. Rest of table: ? unknown, + Present, -Absent, +D (decreased size: referring to anterior commissure) +E (enlarged size: referring to anterior commissure), Abnl = Abnormal, ABS = Seizures (Absence type), ASD = Atrial Septal Defect , BPC = Blake's Pouch Cyst, Cb = Cerebellar , CBLND = Cortical Blindness, CC = Corpus Callosum, CCA = Corpus Callosum Abnormality, Col = Optic Colobomas, CSP = Cavum Septum Pellucidum, Ctrl = Controlled (referring to seizures), CV = Cardiovascular, CVH = Cerebellar Vermis Hypoplasia, Def = Deficiency, DD = Developmental Delay, F = Female, GH = Growth Hormone, GMH = Gray Matter Heterotopias, mo = months, M = Male, MED = Multiple Epiphyseal Dysplasia, MLD=Mild, Mos=Months,MR = Mental Retardation, NA= Not applicable, NCtrl = Not Controlled (referring to seizures), ON = Optic Nerve, ONA = Optic Nerve Atrophy, OPCA = Olivo Ponto Cerebellar Atrophy, PanCA= Pan Cerebellar Atrophy (referring to cerebellar atrophy), PB= Probst Bundles, PCH= Ponto-Cerebellar Hypoplasia, PLD=Polydactyly, PMG=Polymicrogyria, PS = Perisylvian, PST= Pulmonary Stenosis (referring to CV problems), Pt=Patient, SEV=Severe, SOD=Septo Optic Dysplasia, VEP= Visual Evoked Potentials , WMD=White Matter Disease, unk=unknown, yrs=years