	Case no	Age, sex	GA at birth	Gene	Variant	Perinatal complications	Epilepsy	Developmental delay	Hemiparesis	Notable brain imaging findings	Antithrombotic therapies
	1	3, F	35w6d	COL4A1	c.3655G>C, p.(Gly1219Arg)	In-utero ventriculomegaly	Yes, focal	Global, severe	No	Unilateral schizencephaly, bilateral ventriculomegaly, thin CC	None
	2	4, M	39w	COL4A1	c.903+1G>A, (Splice donor)	None	No	Global, mild	No	White matter gliosis	None
	3	4, M	39w1d	COL4A1	c.3307G>A, p.(Gly1103Arg)	In-utero IVH	Yes, focal	Global, mild	No	Bilateral IVH, PVL, bilateral ventriculomegaly, thin CC	None
	4	13, M	40w	COL4A1	c.2317 G>A, p.(Gly773Arg)	In-utero ventriculomegaly	Yes, LGS	Global, severe	No	Bilateral ventriculomegaly	None
	5	8, M	34w	COL4A1	c.3208G>A, p.(Gly1070Arg)	Placental infarct	Yes, focal	Global, severe	No	Unilateral porencephaly, white matter gliosis	None
Family 1	6	15, F	39w	COL4A1	c.3307G>A, p.(Gly1103Arg)	In-utero stroke	Yes, focal	Global, mild	Yes	Intrauterine stroke, ventriculomegaly	None
	7	13, M	40w	COL4A1	c.3307G>A, p.(Gly1103Arg)	In-utero IVH	Yes, focal	Global, mild	Yes	Unilateral porencephaly, PVL, thin body of CC	None
	8	12, F	40w	COL4A1	c.3307G>A, p.(Gly1103Arg)	In-utero stroke	No	No	No	Unilateral porencephaly, white matter gliosis	None
Family 2	9	11, F	39w	COL4A1	13q33.3-4 deletion	None	No	Global, mild	No	White matter gliosis	None
	10	10, M	28w	COL4A1	13q33.3-4 deletion	Respiratory distress	No	Global, mild	No	White matter gliosis	None
	11	8, F	43w	COL4A1	13q33.3-4 deletion	None	No	Global, mild	No	Focal polymicrogyria, pachygyria, white matter gliosis	None
	12	31, F	40w	COL4A1	13q33.3-4 deletion	None	Yes	No	Yes	Acute ischemic infarct	Aspirin
	13	15wk, M	34w	COL4A1	c.3095G>A, p.(Gly1032Asp)	In-utero IVH	Yes	No	No	Unilateral schizencephaly	None
	14	11, M	39w	COL4A1	c.2788G>C, p.(Gly930Arg)	Jaundice, IVH	Yes	Global, severe	No	Bilateral IVH, unilateral ventriculomegaly, white matter gliosis, thin CC	None
	15	9, F	40w	COL4A1	c.2263G>A, p.(Gly755Arg)	In utero stroke, neonatal seizures	Yes, focal	Motor only, mild	No	Intrauterine stroke, white matter gliosis, unilateral porencephaly, thin body of CC	None
	16	16mo, M	39w	COL4A2	c.3155G>T p.(Gly1052Val)	In-utero ventriculomegaly	No	Motor only, mild	Yes	Unilateral ventriculomegaly, IVH	None

Findings	Recommendations
Structural brain abnormalities, abnormal vasculature, cerebral aneurysms	Obtain MR imaging of brain and head and neck vessels, consider serial imaging to screen for aneurysms
Cataracts, colobomas, eye anterior segment dysgeneses, retinopathy	Refer for Ophthalmology evaluation
Kidney cysts, nephropathy with hematuria, proteinuria	Screen with renal function testing, renal ultrasound, and urinalysis
Hemolytic anemia	Screen with complete blood count with differential
Ischemic and/or hemorrhagic strokes with undefined risk of recurrence	Consider antithrombotic agents judiciously in select patients with natural history of ischemic strokes only and/or comorbidities which escalate risk of ischemic stroke significantly
Seizures, spasticity, dystonia	Refer for Neurology clinic follow-up with anti-seizure, muscle relaxant agents as indicated
Developmental delay	Establish physical, occupational and speech therapies for development early
Autosomal dominant inheritance	Obtain genetic counseling and discuss screening of family members