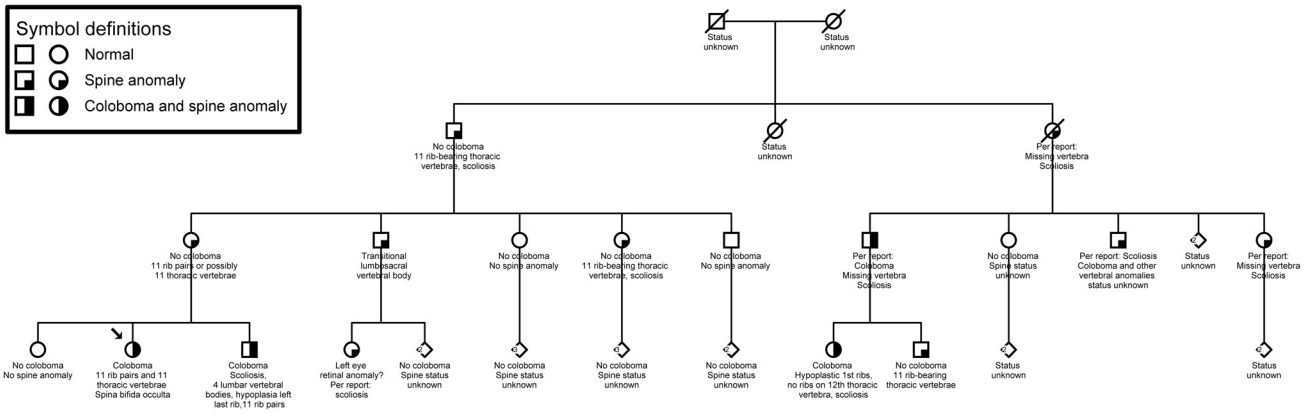
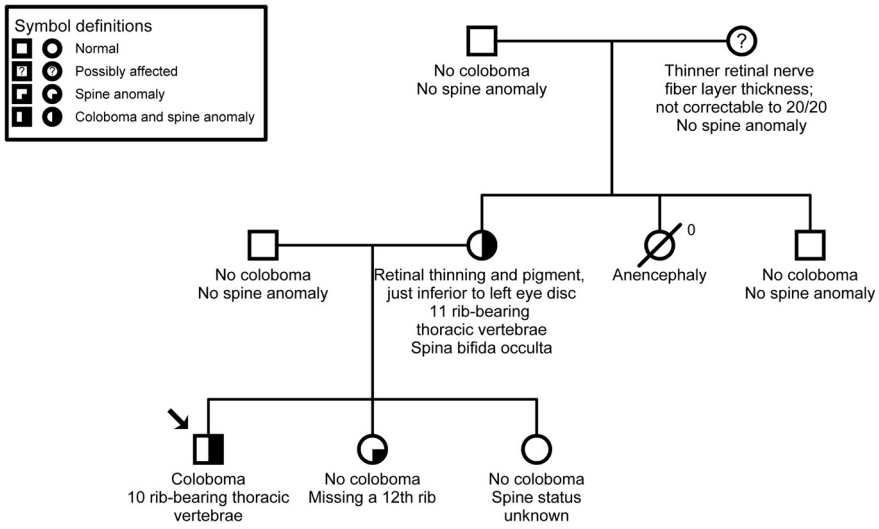


SUPPLEMENTAL FIGURE 1. Uveal coloboma and vertebral anomalies in Family 1. In this pedigree of Family 1, the proband (arrow) and her brother have coloboma and vertebral anomalies. The proband's second cousin and her mother's first cousin also have coloboma and vertebral anomalies. Eight other relatives have spine anomalies but no coloboma on anterior and posterior segment examination, suggesting the coloboma phenotype is incompletely penetrant.



SUPPLEMENTAL FIGURE 2. Uveal coloboma and vertebral anomalies in Family 2. In Family 2 (unrelated to Family 1), the proband (arrow) and his mother have both coloboma and vertebral anomalies. The proband's sister has a unilateral absent 12th rib, but no coloboma on examination.

SUPPLEMENTAL TABLE 1. Uveal Coloboma Patients Excluded From Analysis (n = 25)

	No. of Participants
13q deletion, mosaic	1
2q37 deletion	1
47 XXX	1
Branchio-oculo-facial (BOF) syndrome	3
CHARGE syndrome	5
Chorioretinal dystrophy	1
Anomaly of the optic nerve	1
Kabuki syndrome	1
Macular coloboma	2
Morning glory malformation	3
Optic nerve pit	1
Papillo-renal syndrome	4
45 X (Turner syndrome)	1

SUPPLEMENTAL TABLE 2. Summary of Recent Population-Based Studies on Uveal Coloboma

Study	Design	Study Participants	Main Ocular Findings	Main Systemic Findings
Nakamura 2011[5]	Retrospective review of pediatric patients in Olmsted County, Minnesota	33 pediatric patients (<19 years) with ocular coloboma, including patients with CHARGE syndrome	*Location of coloboma:* 36% anterior segment 39% posterior segment 29% anterior & posterior segment *Concomitant ocular anomalies:* 33% amblyopia 30% strabismus 15% microphthalmia/anophthalmia 55% with bilateral AMC 73% with coloboma	36% developmental delay/mental retardation 21% cardiac anomalies 21% ear anomalies 18% skeletal anomalies 15% urogenital anomalies
Shah 2011[7] and 2012[6]	Prospective study of patients in the United Kingdom, recruited through an established surveillance system	135 children with newly diagnosed AMC	*Location of coloboma:* 14% anterior segment 43% posterior segment 43% anterior & posterior segment *Concomitant ocular anomalies (in addition to AMC):* 25% cataract 16% nystagmus 6% optic nerve abnormalities 4% detached retina	60% with extraocular abnormalities 32% craniofacial 21% cardiovascular 15% gastrointestinal 14% CNS 12% hearing impaired 12% growth retardation 10% genitourinary 10% musculoskeletal
Current study	Retrospective review of patients seen at the National Eye Institute in Bethesda, Maryland	99 patients (children and adults) with nonsyndromic coloboma	*Location of coloboma:* 8% anterior segment 23% posterior segment 69% anterior & posterior segment *Concomitant ocular anomalies:* 29% microphthalmia 19% strabismus 16% cataract 15% microcornea 14% nystagmus 10% amblyopia	53% cardiac anomalies 17% CNS anomalies 16% developmental delay 15% audiologic abnormalities 13% skeletal anomalies 7% urogenital anomalies

AMC = anophthalmia, microphthalmia, and typical coloboma; CNS = central nervous system.