

Appendix 1. The clinical features of aniridia patients in the two Chinese families.

Patient	Age (years) / Gender	Eye	Visual acuity	Best corrected visual acuity	Aniridia	Lens
FAMILY-1:II2	46/F	OD	FC	15/200	Partial	Aphakia
		OS	FC	FC	Partial	Posterior subcapsular cataract
FAMILY-1:II3	44/F	OD	20/200	NA	Complete	Cortical cataract
		OS	20/200	NA	Partial	Posterior subcapsular cataract
FAMILY-1:III1	23/F	OD	10/200	10/200	Complete	Cortical cataract
		OS	FC	20/200	Complete	IOL
FAMILY-1: III4	19/F	OD	NA	NA	Complete	Cortical cataract
		OS	NA	NA	Complete	Cortical cataract
FAMILY-1:III5	7/M	OD	30/200	40/200	Complete	No
		OS	30/200	40/200	Complete	No
FAMILY-2:II2	26/F	OD	FC	FC	Complete	Posterior subcapsular cataract
		OS	FC	FC	Complete	Posterior subcapsular cataract
FAMILY-2:III1	3/F	OD	30/200	30/200	Complete	No
		OS	40/200	40/200	Complete	No

M: male; F: female; NA: not available; IOL: intraocular Lens;  
FC: finger counting

Nystagmus	Iop (mmHg)	Glaucoma	Alial Length (mm)	Corneal curvature (D)	Endothelial cells density (/mm)
Yes	31	Yes	29.16	Min:42.0 MAX:42.10	2574
Yes	26	Yes	28.29	Min:42.0 MAX:42.54	2660
Yes	17	No	26.63	Min:44.42 MAX:44.81	3198
Yes	19	No	25.94	Min:42.88 MAX:47.63	3110
Yes	18	No	NA	NA	NA
Yes	20	No	23.9	Min:41.0 MAX:41.7	3236
Yes	NA	NA	NA	NA	NA
Yes	NA	NA	NA	NA	NA
Yes	10	No	20.09	Min:42.10 MAX:45.75	NA
Yes	12	No	20.12	Min:41.45 MAX:44.40	NA
Yes	16	No	NA	NA	NA
Yes	18	No	NA	NA	NA
Yes	13	No	NA	NA	NA
Yes	15	No	NA	NA	NA

B-scanning	Surgery and trauma history
Vitreous bodies opaque, posterior scleral staphyloma	phaco + trabeculectomy + anterior vitreous body incision
Vitreous bodies opaque, posterior scleral staphyloma	No
Vitreous bodies opaque, posterior scleral staphyloma	No
Vitreous bodies opaque, posterior scleral staphyloma	No
Vitreous bodies opaque	No
Vitreous bodies opaque	phacoemulsification + IOP implantation
NA	No
NA	No
NA	No
NA	No
Vitreous bodies opaque	No
Vitreous bodies opaque	No
Vitreous bodies opaque	correction of ptosis
Vitreous bodies opaque	No