

Table 3. Clinical findings in three female heterozygous carriers of *CHRD1* variants.

	Family 3		Family 4			
Pedigree number	I:2		II:2		II:4	
Age at examination (y)	58		37		36	
Eye	RE	LE	RE	LE	RE	LE
BCVA	1.0	1.0	1.0	1.0	1.0	1.0
Refractive error	+0.5/-	-	-	-	-0.25/-3x4°	+0.5/-
Iris hypoplasia/atrophy	-	-	-	-	-	-
Arcus	-	-	-	.-	-	-
Shagreen	-	-	-	-	-	-
ACD (mm)	2.78	2.85	3.36	3.47	3.41	3.43
WTW (mm)	12.3	12.1	12.1	12.1	12.3	12.2
AL (mm)	23.44	23.42	22.94	23.11	24.24	24.10
CCT (µm)	521	527	471	472	443	441
ECD (cells/mm²)	2475	2392	NA	NA	2380	2344
Glaucoma	-	-	-	-	-	-
Other	-	-	Multiple iris nevi	Multiple iris nevi	KC (TKC 1-2)	-

ACD = anterior chamber depth (normal values under 97.5 percentiles ≤ 3.8 mm) (Gilani et al. 2013), AL = axial length, BCVA = best corrected visual acuity, CCT = central corneal thickness (normal values normal above 2.5 percentiles ≥ 481 µm) (Gilani et al. 2013), ECD = endothelial lens density, KC = keratoconus, LE = left eye, NA = not available data, RE = right eye, TP = thinnest pachymetry (normal values normal above 2.5 percentiles ≥ 479 µm), TKC = Topographic Keratoconus Classification derived from Pentacam, Y = years, WTW = horizontal white to white, - = not present, + = present