

Table S1 Comprehensive overview of patients' clinical characteristics, iridocorneal angle morphology, and genetic defects

| Patient No. | Sex | Age at first visit | Presenting complaint | Onset age of glaucoma | Iridocorneal angle by number of quadrants (OD) | | | | | | Corneal clarity (OD) | Iridocorneal angle by number of quadrants (OS) | | | | | | Corneal clarity (OS) | Mutant gene | Nucleotide changes |
|-------------|-----|--------------------|-------------------------------------|-----------------------|--|--------|--------|--------|--------|--------|----------------------|--|--------|--------|--------|--------|--------|----------------------|-------------|--------------------------------------|
| | | | | | Type A | Type B | Type C | Type D | Type E | Type F | | Type A | Type B | Type C | Type D | Type E | Type F | | | |
| 1 | M | 1 m | Whitish appearance of the left eye | at birth | | 1 | 3 | | | | | 1 | 1 | 3 | | | | 2 | FOXC1 | c.388C>T |
| 2 | M | 1 m | Whitish appearance of both eyes | at birth | | 1.5 | | 2.5 | | | | 2 | - | - | - | - | - | 2 | FOXC1 | c.592_593delins C |
| 3 | F | 1 m | Whitish appearance of both eyes | at birth | | 4 | | | | | | 2 | 4 | | | | | 2 | FOXC1 | Gross deletion in 6: 1478641–2694750 |
| 4 | M | 1 m | Whitish appearance of both eyes | at birth | | 3.5 | | 0.5 | | | | 2 | 3.5 | | 0.5 | | | 2 | FOXC1 | c.503_504insGCGGCG |
| 5 | M | 2 m | Whitish appearance of both eyes | at birth | 1.5 | | 2.5 | | | | | 2 | 1.5 | | 2.5 | | | 2 | FOXC1 | c.392C>T |
| 6 | F | 9 y | Whitish appearance of both eyes | at birth | | 3 | 1 | | | | | 3 | 3 | 1 | | | | 3 | FOXC1 | c.430A>T |
| 7 | M | 1 m | Whitish appearance of both eyes | 1m | | 1.5 | | 2.5 | | | | 2 | 1.5 | | 2.5 | | | 2 | FOXC1 | c.380G>A |
| 8 | M | 4 m | Whitish appearance of the right eye | 4m | | | 4 | | | | 2.5 | - | - | - | - | - | - | 1 | FOXC1 | c.240_241insTAT |
| 9 | M | 5 m | Whitish appearance of both eyes | 5m | | 2.5 | 1.5 | | | | 2.5 | | 2.5 | 1.5 | | | | 2.5 | FOXC1 | Gross deletion in 6: 382441–2333070 |
| 10 | M | 10 y | Reduced vision in both eyes | 10y | 1 | 2.5 | | 0.5 | | | | 2 | 1.5 | 2.5 | | | | 2 | FOXC1 | c.275A>C |
| 11 | F | 14 y | Reduced vision in both eyes | 14y | | 1.5 | 2.5 | | | | | 1 | 1 | 3 | | | | 1 | FOXC1 | c.246C>G |
| 12 | M | 15 y | Reduced vision in both eyes | 15y | | | | 4 | | | | 1 | | | 4 | | | 1 | FOXC1 | c.482T>A |
| 13 | M | 15 y | Pain in both eyes | 15y | | 3 | 1 | | | | | 1 | 2.5 | 1.5 | | | | 1 | FOXC1 | c.205delC |
| 14 | F | 18 y | Reduced vision in both eyes | 18y | | 4 | | | | | | 1 | 3.5 | | | 0.5 | | 1 | FOXC1 | c.718_719del |
| 15 | F | 24 y | Reduced vision in both eyes | 24y | | 3 | 1 | | | | | 1 | 3 | 1 | | | | 1 | FOXC1 | c.718_719del |
| 16 | F | 5 m | Whitish appearance of both eyes | 5m | | | | 4 | | | 2.5 | | | | 4 | | | 2.5 | PITX2 | c.173T>G |
| 17 | M | 2 y | Reduced vision in both eyes | 2y | | 1.5 | | 2.5 | | | | 1 | 1.5 | | 2.5 | | | 1 | PITX2 | c.348delG |
| 18 | F | 4 y | Reduced vision in both eyes | 4y | | | | 3 | 1 | | | 1 | | | 3 | 1 | | 2 | PITX2 | c.103A>T |
| 19 | F | 5 y | Reduced vision in both eyes | 5y | | | | 4 | | | 2 | | | | 4 | | | 1 | PITX2 | c.269G>C |
| 20 | F | 13 y | Pain in the left eye | 13y | - | - | - | - | - | - | | 1 | | | 4 | | | 1 | PITX2 | c.253-11A>G |
| 21 | M | 17 y | Reduced vision in both eyes | 17y | | | | | | | 4 | 1 | | | | | 4 | 1 | PITX2 | c.190C>G |
| 22 | M | 18 y | Reduced vision in both eyes | 18y | 2.5 | | | 1.5 | | | | 1 | 3 | | 1 | | | 1 | PITX2 | c.158_173GGTAGCT |
| 23 | M | 24 y | Reduced vision in the left eye | 24y | | | | 3 | 1 | | | 1 | 1 | | 3 | | | 1 | PITX2 | c.253-1G>A |
| 24 | M | 25 y | Reduced vision in both eyes | 25y | 3 | | | 1 | | | | 1 | 3 | | 1 | | | 1 | PITX2 | c.377delC |
| 25 | M | 30 y | Reduced vision in the left eye | 30y | 3 | | | | | 1 | | 1 | 3 | | | | 1 | 1 | PITX2 | c.253-11A>G |
| 26 | M | 35 y | Reduced vision in both eyes | 35y | 0.5 | | | 3.5 | | | | 1 | 0.5 | 1 | | 2.5 | | 1 | PITX2 | c.539_551del |
| 27 | F | 41 y | Pain in both eyes | 41y | | 0.5 | | 0.5 | 3 | | | 2 | | 1.5 | | 2.5 | | 2 | PITX2 | c.173T>C |
| 28 | F | 46 y | Pain in both eyes | 46y | 0.5 | | 0.5 | | 3 | | | 2 | | | 2.5 | 1.5 | | 2 | PITX2 | c.253-11A>G |
| 29 | M | 61 y | Reduced vision in both eyes | 61y | 2.5 | 0.5 | 0.5 | | | 0.5 | | 1 | 3.5 | | 0.5 | | | 1 | PITX2 | c.282G > A |
| 30 | F | 10 y | Reduced vision in both eyes | 10y | - | - | - | - | - | - | | 2 | 0.5 | 0.5 | | 3 | | 1 | / | NA |
| 31 | M | 14 y | Reduced vision in both eyes | 14y | 0.5 | | | 3.5 | | | | 1 | - | - | - | - | - | 1 | / | NA |
| 32 | M | 14 y | Reduced vision in both eyes | 14y | | | 4 | | | | | 1 | | 4 | | | | 1 | / | NA |
| 33 | M | 15 y | Reduced vision in both eyes | 15y | | 4 | | | | | | 1 | 4 | | | | | 1 | / | NA |
| 34 | M | 16 y | Pain in both eyes | 16y | | | | | | | 4 | 1 | | | | | 4 | 1 | / | NA |
| 35 | M | 22 y | Reduced vision in both eyes | 22y | | | | | | | 4 | 2 | | | | | 4 | 1 | / | NA |
| 36 | F | 22 y | Pain in both eyes | 22y | | 3 | 0.5 | 0.5 | | | | 1 | 3 | 0.5 | | 0.5 | | 1 | / | NA |
| 37 | M | 32 y | Pain in both eyes | 32y | 3 | | 1 | | | | | 2 | 3 | | 1 | | | 2 | / | NA |
| 38 | M | 34 y | Reduced vision in both eyes | 34y | 3 | 1 | | | | | | 1 | 2.5 | 1.5 | | | | 1 | / | NA |
| 39 | F | 36 y | Reduced vision in both eyes | 36y | 1 | | | 0.5 | 2.5 | | | 1 | 1 | | | 0.5 | 2.5 | 1 | / | NA |
| 40 | F | 36 y | Pain in both eyes | 36y | 1 | | | 3 | | | | 1 | - | - | - | - | - | 1 | / | NA |
| 41 | F | 42 y | Reduced vision in the right eye | 42y | | 3 | 0.5 | 0.5 | | | | 1 | | 3 | 1 | | | 1 | / | NA |
| 42 | M | 70 y | Reduced vision in both eyes | 70y | 3 | 1 | | | | | | 2 | 3 | 1 | | | | 1 | / | NA |

Type A: pure high iris insertion; Type B: posterior embryotoxon; Type C: iris process; Type D: trabecular-iris synechia; Type E: peripheral iridocorneal adhesion; Type F: goniodysgenesis. No. 14 and No. 15 are sisters; No. 25 is the father of No. 20. OD, right eye; OS, left eye; M, male; F, female; -, excluded in this study; /, negative for the variant; NA, not applicable.