# Visual acuities and refractions in patients with *PDE6C*-associated achromatopsia

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## Background

- PDE6C encodes a subunit of the photoreceptor phosphodiesterase integral to cone phototransduction.
- Biallelic pathogenic variants cause cone dysfunction/achromatopsia.[1,2]
- Variants in this gene are a rarer cause of achromatopsia in the UK, compared with *CNGA3* and *CNGB3*.[3,4]
- Here, we explored visual acuities and refractions in patients with molecularly confirmed disease.

## Methods

- The database of a large inherited retinal disease service was retrospectively reviewed to identify patients with *PDE6C*-associated achromatopsia.
- Best recorded visual acuities from each clinical visit were analysed.
- Snellen visual acuities were converted to logMAR equivalents.
- Where refractive data was available, spherical equivalents were calculated for each patient.

## Cohort Summary

**14** patients (10 female)

**13** families

**26.1** mean age at first visit (SD 14.4; range 1.5–44.7)

## Results

- Fourteen patients (10 females) from 13 families were identified.

**Acuities at first visit**

- Mean (SD) age at first visit was 26.1 (14.4) years (median 27.1; range 1.5 to 44.7).
- Mean (SD) acuity: 0.86 (0.27) for right eyes (median 0.95)
  0.86 (0.29) for left eyes (median 1.00)

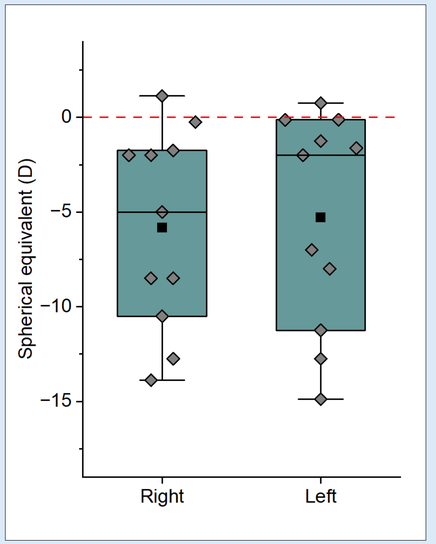
**Acuities over time**

- Follow up for more than 1 year was available for 13 patients
- Mean (SD) follow up, 10.1 (6.9) years; median 8.3 years.
- Mean (SD) ages at first and last visit: 25.3 (14.7) and 35.9 (18.9) years respectively.
  At 1st visit, mean (SD) acuities were 0.86 (0.28) right and 0.84 (0.30) left.
  At last visit, mean (SD) acuities were 0.90 (0.15) right and 0.91 (0.14) left.

**Refractions**

- Refractions were available for 11 patients (Figure 1).
- Mean (SD) age was 34.1 (20.1) years (median 37.8; range 2.6 to 58.8 years).
- Right eyes: mean (SD) refraction: –5.82 (5.24) D (median –5.00; range–13.88 to +1.13)
- Left eyes: mean (SD) refraction: –5.30 (5.69) D (median -2.00; range –14.88 to +0.75)
- Five patients (45%) had more than 6 dioptres of myopia in both eyes.

**Figure 1 (below). Spherical equivalent refractions for right and left eyes.**




## Conclusions

- High myopia was present **in nearly half of patients**, contrasting with other genetic causes of achromatopsia.
- Understanding which cone dysfunction syndromes predispose to significant refractive error may yield insight into mechanisms underlying myopia development.
- These data can help inform prognostic discussions and the design of future therapeutic trials.

**5 of 11 patients (45%) had > 6 dioptres of myopia in both eyes**

## References

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