

Supplemental File 7. Cone densities along the temporal meridian in proband (IV-3), minimally symptomatic mother (III-12) and unaffected father (III-11) compared to the relevant reported normative values from Cooper et al. 2016.<sup>27</sup>

Eccentricity from foveal centre	0.5T	1T	2T	3T	4T	5T
Eccentricity bin from Cooper et al.	150 $\mu$ m	300 $\mu$ m	600 $\mu$ m	1000 $\mu$ m	1200 $\mu$ m	1600 $\mu$ m
IV-3	71.6 $\pm$ 5.8	41.8 $\pm$ 1.0	20.7 $\pm$ 1.1	14.5 $\pm$ 0.5	11.4 $\pm$ 1.6	10.7 $\pm$ 0.7
III-12	na	52.6 $\pm$ 2.2	28.8 $\pm$ 2.3	18.3 $\pm$ 0.7	14.8 $\pm$ 0.7	11.5 $\pm$ 0.8
III-11	78.5 $\pm$ 0.7	53.8 $\pm$ 1.1	32.2 $\pm$ 1.0	21.8 $\pm$ 1.1	16.6 $\pm$ 0.3	13.8 $\pm$ 0.5
Cooper et al.	90.7 $\pm$ 13.0	61.8 $\pm$ 8.4	39.0 $\pm$ 5.7	24.6 $\pm$ 5.0	18.6 $\pm$ 2.2	14.4 $\pm$ 1.7

Units are  $\times 10^3$  cones/mm<sup>2</sup>  $\pm$  standard deviation.

T – degrees temporal; na – not available.