

SUPPLEMENTARY FIGURES

Genome-wide linkage and haplotype sharing analysis implicates the MCDR3 locus as a candidate region for a developmental macular disorder in association with digit abnormalities

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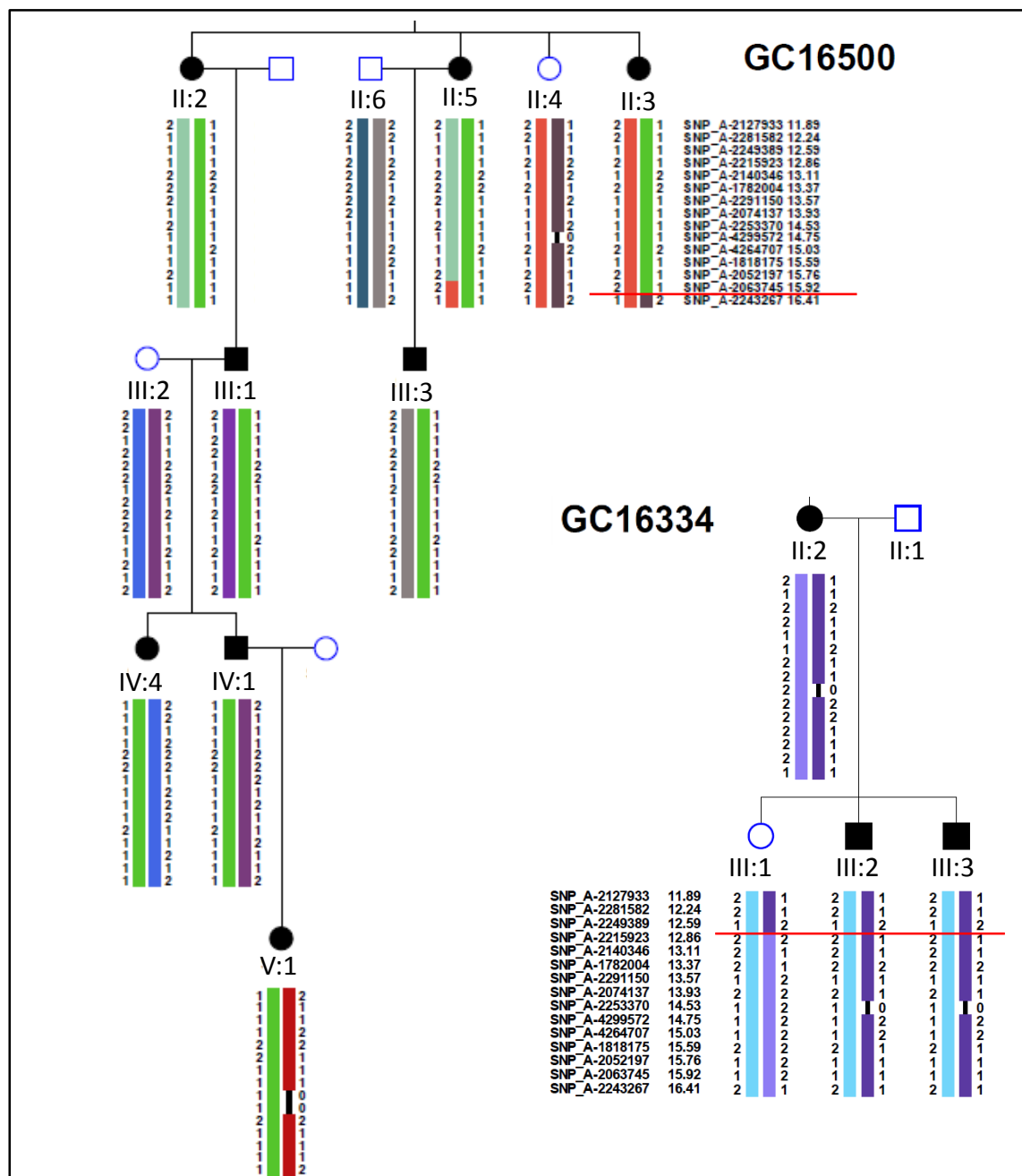
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Supplementary Figure 1

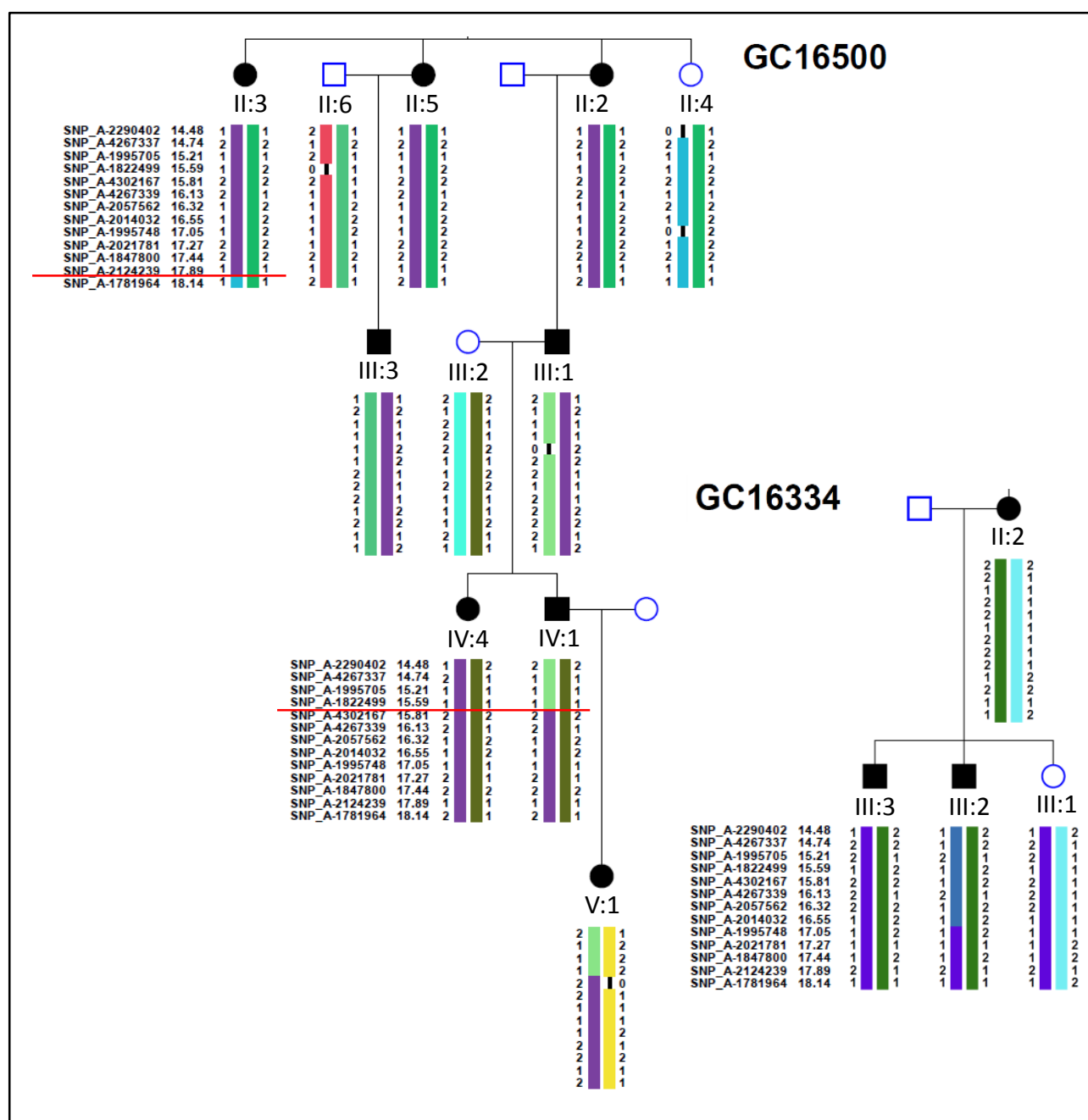
Supplementary Figure 2

Supplementary Figure 3

Supplementary Figure 4

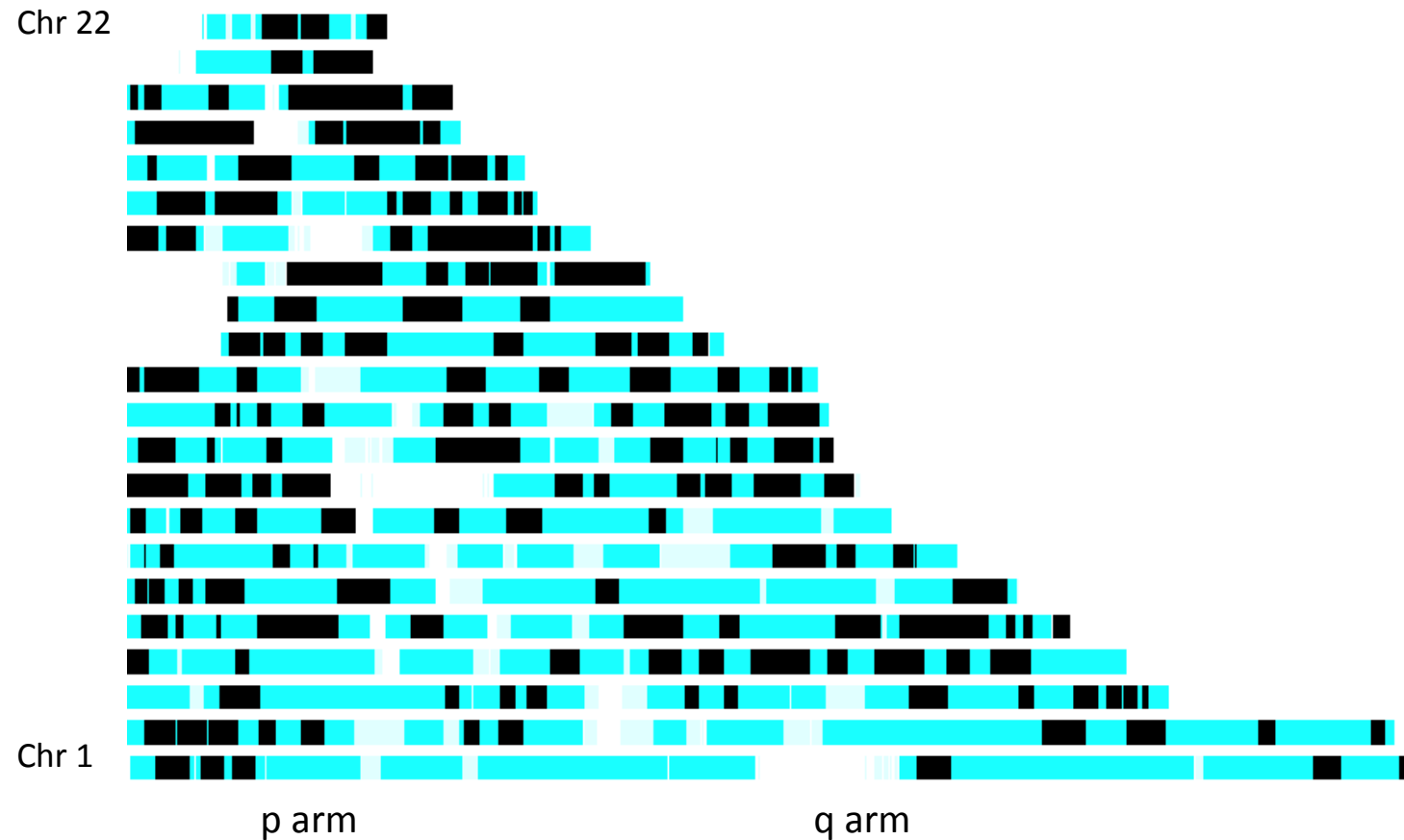


Supplementary Figure 1 Reconstructed haplotypes at chromosome 5p15.32 in family GC16500 and family GC16334. Light green and purple bars represent the haplotype segregating with the disease in family GC16500 and GC16334, respectively. Red lines indicate whether a recombination event was observed (individual II:3 in family GC16500 and individual III:1 in family GC16334).



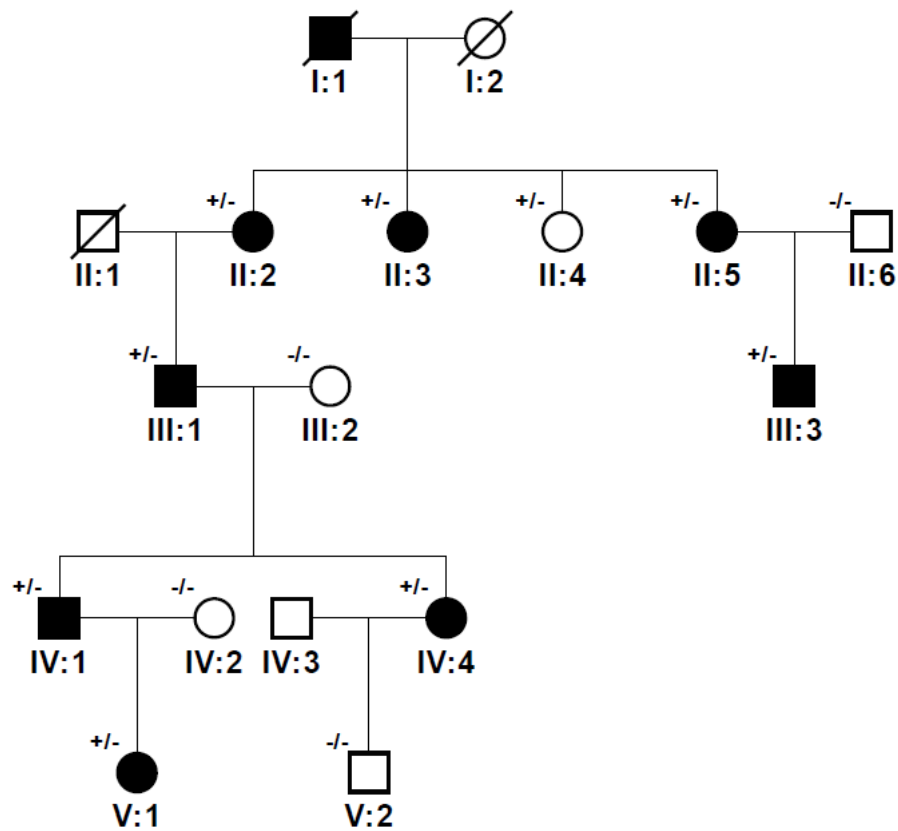
Supplementary Figure 2 Reconstructed haplotypes at chromosome 9p24.1 in family GC16500 and family GC16334. Purple and dark green bars represent the haplotype segregating with the disease in family GC16500 and GC16334, respectively. Red lines indicate whether a recombination event was observed (individual II:3 and IV:1 in family GC16500).

Regions with a Conserved Homozygote Haplotype (RCHH)



Supplementary Figure 3 Identification of the candidate regions for family GC16334 using the Homozygosity Haplotype (HH) approach.¹ Three affected family members were included in the analysis. A densitogram of the genomic Regions with a Conserved Homozygosity Haplotype (RCHHs) is depicted. The darker the colour, the more individuals share a HH in the region. Black regions indicate RCHHs that are shared by all 3 affected family members included in the analysis.

GC16500



Supplementary Figure 4 Segregation analysis of the *ZNF774* frameshift deletion (c.517del:p.His173ThrfsTer79) in 8 affected, 2 unaffected and 3 unaffected married-in individuals from family GC16500. The symbols $+/-$ and $-/-$ indicate presence and absence of the mutation respectively. Individual II:4 was examined at age 69 years. No fundus photographs were available. She had minimal macular drusen in the right eye only and no digit abnormalities. Given her age and the unilateral mild retinal phenotype commonly seen in the older population, it is highly unlikely that individual II:4 is affected and so should not be considered a genetic carrier of the condition.

REFERENCES

1. Miyazawa H, Kato M, Awata T, et al. Homozygosity haplotype allows a genomewide search for the autosomal segments shared among patients. *Am J Hum Genet.* 2007;80(6):1090-1102. doi:10.1086/518176.