

Supplemental Information

From University of Auckland

RHO: c.133T>C, p.Phe45Leu

Primer sequences as follows:

RHO_c.133F: 5'-TTCTCCAATGCGACGGGTGTGG-3' (Own design- PrimerBLAST on NCBI)

RHO_c.133R: 5'-TGGACGGTGACGTAGAGCGTGA-3' (Own design- PrimerBLAST on NCBI)

Used FastStart Taq enzyme kit from Roche, with 3.5mM final concentration of MgCl₂.

PCR cycle as follows:

| | degC | time |
|-------------------------------|------|--------|
| initial denaturation | 96 | 5min |
| 30x - denaturation | 94 | 30secs |
| -annealing | 63 | 1min |
| -extension | 72 | 1min |
| final extension | 72 | 10min |

CNGA3: c.1580T>G, p.Leu527Arg

Primer sequences as follows:

CNGA3_1580F: 5'-GAGCCTCCCAGACAAGCTGAAG-3' (Own design- PrimerBLAST on NCBI)

CNGA3_1580R: 5'-AGTAGCCAATGCTGCGGATGTT-3' (Own design- PrimerBLAST on NCBI)

Used FastStart Taq enzyme kit from Roche, with 3.5mM final concentration of MgCl₂.

PCR cycle as follows:

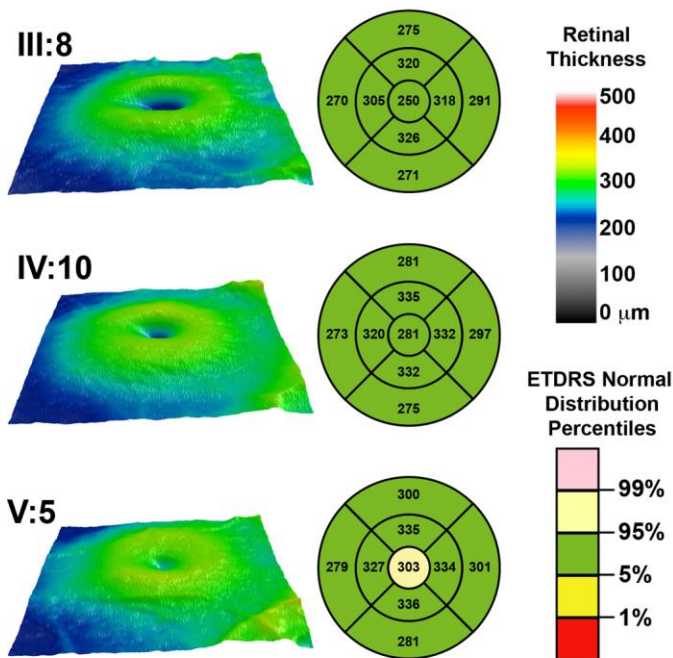
| | degC | time |
|-------------------------------|------|--------|
| initial denaturation | 96 | 5min |
| 30x - denaturation | 94 | 30secs |
| -annealing | 57 | 1min |
| -extension | 72 | 1min |
| final extension | 72 | 10min |

Following column purification with HighPure PCR purification kit (Roche Diagnostic, Mannheim, Germany), the product was sequenced directly according to protocols accompanying the ABI *BigDye*

terminator kit v3.1 (Applied Biosystems Inc, Foster City, Ca, USA). Bidirectional sequencing of amplicons was undertaken on an ABI 3700 prism genetic analyzer (Applied Biosystems Inc, Foster City, Ca, USA).

From University of Tuebingen

PCR fragments were subjected to a clean-up protocol (ExoSAP-IT; GE Healthcare, Freiburg, Germany) and then to direct DNA sequencing with dye terminator chemistry (BigDye Terminator ver. 1.1; Applied Biosystems [ABI], Darmstadt, Germany). All sequences were run on a capillary sequencer (ABI 3100; ABI) and analyzed with proprietary sequence trace analysis (Sequence Analysis, ver. 5.1; ABI) or sequence trace alignment software (SeqMan; DNASTAR, Madison, WI).



Supplemental Figure Caption

Supplemental Figure 1 – Topographical retinal thickness profiles, along with Early Treatment Diabetic Retinopathy (ETDRS) thickness maps for 3 of the subjects in the current study. Retinal thickness was normal in these 3 subjects, all of whom were positive for the RHO c. 133T>C; p.F45L mutation.