

SUPPLEMENTARY MATERIAL

## Supplementary material

**Table i.** List of *WISP3* primers

| Primer            | Sequence                   | Length of the amplicon (bp) | Annealing temperature (°C) |
|-------------------|----------------------------|-----------------------------|----------------------------|
| Forward primer 1F | 5'TCACTGCGAAGGCAGGTTATT 3' | 508                         | 57.6                       |
| Reverse primer 1R | 5'TGCCATTACCTGAAAGGGAGA 3' |                             |                            |
| Forward primer 2F | 5' CCTGTTGGGGGAAATCTTCT3'  | 531                         | 57.6                       |
| Reverse primer 2R | 5'TACAATGGAGCCAGTCCCACT 3' |                             |                            |
| Forward primer 3F | 5'TCCTGTGAAGGAGTTCCAAA 3'  | 553                         | 57.9                       |
| Reverse primer 3R | 5'TCCCTGTCTGAGGCAAAGATT 3' |                             |                            |
| Forward primer 4F | 5'AGGCAAAGCAGAAAAATGCAA 3' | 559                         | 62.6                       |
| Reverse primer 4R | 5'ATCCCAACCTCCAAAACACAC 3' |                             |                            |
| Forward primer 5F | 5'AAGGGTAAAGAGAGTGCTGGA 3' | 518                         | 56.3                       |
| Reverse primer 5R | 5'AAACAAGTAGATTTGCCACCA 3' |                             |                            |

bp- Base Pairs

**Table ii.** Previously described *WISP3* mutations throughout the world

| Exon/intron         | Nucleotide change                 | Amino acid change                 | Patient origin                                  | Reference         |
|---------------------|-----------------------------------|-----------------------------------|---|-------------------|
| exon 1              | c.43delGC                         | p.A15fs*                          | United States                                   | 1                 |
| intron 1            | g.insT+2IVS1                      | Alters intron 1 splice donor site | Jordan  | 1                 |
| intron 1            | c.49-1G>A                         |                                   | India   | 2                 |
| exon 2              | c.136C>T                          | p.G46*                            | China   | 3                 |
| exon 2              | c.156C>A                          | p.C52*                            | Turkey, Lebanon, Syria, Italy, France and India | 1, 4-6, our study |
| exon 2              | c.182G>T                          | p.C61F                            | Poland  | 7                 |
| exon 2              | c.185delC                         | p.P62Lfs*21                       | Turkey  | 7                 |
| exon 2              | c.197G>A                          | p.S66N                            | United States, Italy                            | 7                 |
| exon 2              | c.232T>C                          | p.C78R                            | France  | 4                 |
| exon 2              | c.233G > A                        | p.C78Y                            | India   | 5, our study      |
| exon 2              | c.236-237 CC>AA                   | p.A79E                            | Italy   | 7                 |
| exon 2              | c.246delA                         | p.P82fs*21                        | Saudi Arabia, Jordan                            | 4                 |
| exon 2              | c.248G > A                        | p.G83E                            | Lebanon, Syria, Turkey and India                | 4,5,6, our study  |
| exon 2              | c.296A>T                          | p.Y99F                            | India   | 2                 |
| exon 2              | c.298T>A                          | p.C100S                           | India   | 2                 |
| exon 2              | c.327C>A                          | p.Y109*                           | Turkey  | 7                 |
| exon 2              | c.340T > C                        | p.C114R                           | India   | 5                 |
| exon 2              | c.341 G>A                         | p.C114Y                           | China   | 3                 |
| exon 2              | c.342T>G                          | p.C114W                           | China   | 8                 |
| exon 2              | c.342_343delTG                    | p.A115Lfs*16                      | Turkey  | 7                 |
| intron 2            | c.347-2A>G                        |                                   | India   | 2                 |
| intron 2            | c.347-1_347-3delCAG               |                                   | India   | 2                 |
| exon 2/3            | c.348C>A                          | p.Y116*                           | India   | 5, our study      |
| exon 3              | c.433T > C                        | p.C145R                           | India   | 5                 |
| exon 3              | c.434G>A                          | p.C145Y                           | Italy   | 4                 |
| exon 3              | c.530C>A                          | p.S177*                           | India   | 2                 |
| exon 3              | c.535_536delITG or c.536_537delGT | p.C179fs*                         | Syria   | 4                 |
| exon 3              | c.589G>C                          | p.A197Lfs*35                      | Syria   | 4                 |
| exon 3              | c.589G>A                          | p.A197Lfs*35                      | Italy   | 7                 |
| exon 4              | c.593_597delATAGA                 | p.Y198*                           | India   | our study         |
| exon 4              | c.621_622delAAinsT                | p.K207Nfs*25                      | United States                                   | 7                 |
| exon 4              | c.624_625insA                     | p.C209Mfs*21                      | China   | 9                 |
| exon 4              | c.667T>G                          | p.C223G                           | China   | 10                |
| exon 4              | c.683_684insT                     | p.N229*                           | India   | 2                 |
| exon 4              | c.685_686insATCTA                 | p.R230Lfs*4                       | India   | 2                 |
| exon 4              | c.670G>A                          | p.G224R                           | Italy   | 7                 |
| exon 4              | c.677G>T                          | p.G226V                           | United Kingdom                                  | 7, our study      |
| exon 4              | c.682T > C                        | p.S228P                           | India   | 5                 |
| exon 4              | c.708dupC                         | p.N237Qfs*3                       | Turkey  | 7                 |
| exon 4              | c.716_722delAAATGAG               | p.E239fs*16                       | China   | 9                 |
| exon 4              | c.725-726delAA                    | p.K242Rfs*36                      | Italy   | 7                 |
| exon 4              | c.727_731delGAGAA                 | p.E243Kfs*34                      | Turkey  | 7                 |
| exon 4              | c.729_735delGAGAAAA               | p.M243fs*15                       | China   | 9                 |
| exon 4              | c.739_740delITG                   | p.C247Lfs*31                      | Germany, India                                  | 5, 11             |
| exon 4              | c.740_741delGT                    | p.C247Lfs*31                      | India   | 2                 |
| exon 4              | c.756C>A                          | p.C252X                           | China   | 7                 |
| exon 4 and intron 4 | c.779_783+1d eITAAAGG             |                                   | India   | 2                 |

(continued)

**Table ii.** (continued)

| Exon/intron | Nucleotide change | Amino acid change | Patient origin | Reference    |
|-------------|-------------------|-------------------|----------------|--------------|
| exon 5      | c.802T > G        | p.C268G           | India          | 5            |
| exon 5      | c.804delC         | p.Q269Nfs*44      | India          | 2            |
| exon 5      | c.850G>T          | p.G284*           | Turkey         | 7            |
| exon 5      | c.857C>G          | p.S286*           | Turkey         | 7            |
| exon 5      | c.863insAC        | p.Q289Lfs*25      | United States  | 4            |
| exon 5      | c.866_867insA     | p.S290Efs*13      | China          | 8,9          |
| exon 5      | c.866_867delAG    | p.S290Lfs*12      | Iran           | 4            |
| exon 5      | c.947_951delAATTT | p.Q316Rfs*5       | India          | 5            |
| exon 5      | c.993G>A          | p.W331*           | Italy          | 4            |
| exon 5      | c.1000T>C         | p.S334P           | China          | 8            |
| exon 5      | c.1004G>A         | p.C335Y           | Italy          | 7            |
| exon 5      | c.1010G > A       | p.C337Y           | India          | 5, our study |
| exon 5      | c.1013A>T         | p.Q338L           | Japan          | 12           |

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