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Supplemental Data

Mutations in *FYCO1* Cause Autosomal-Recessive Congenital Cataracts

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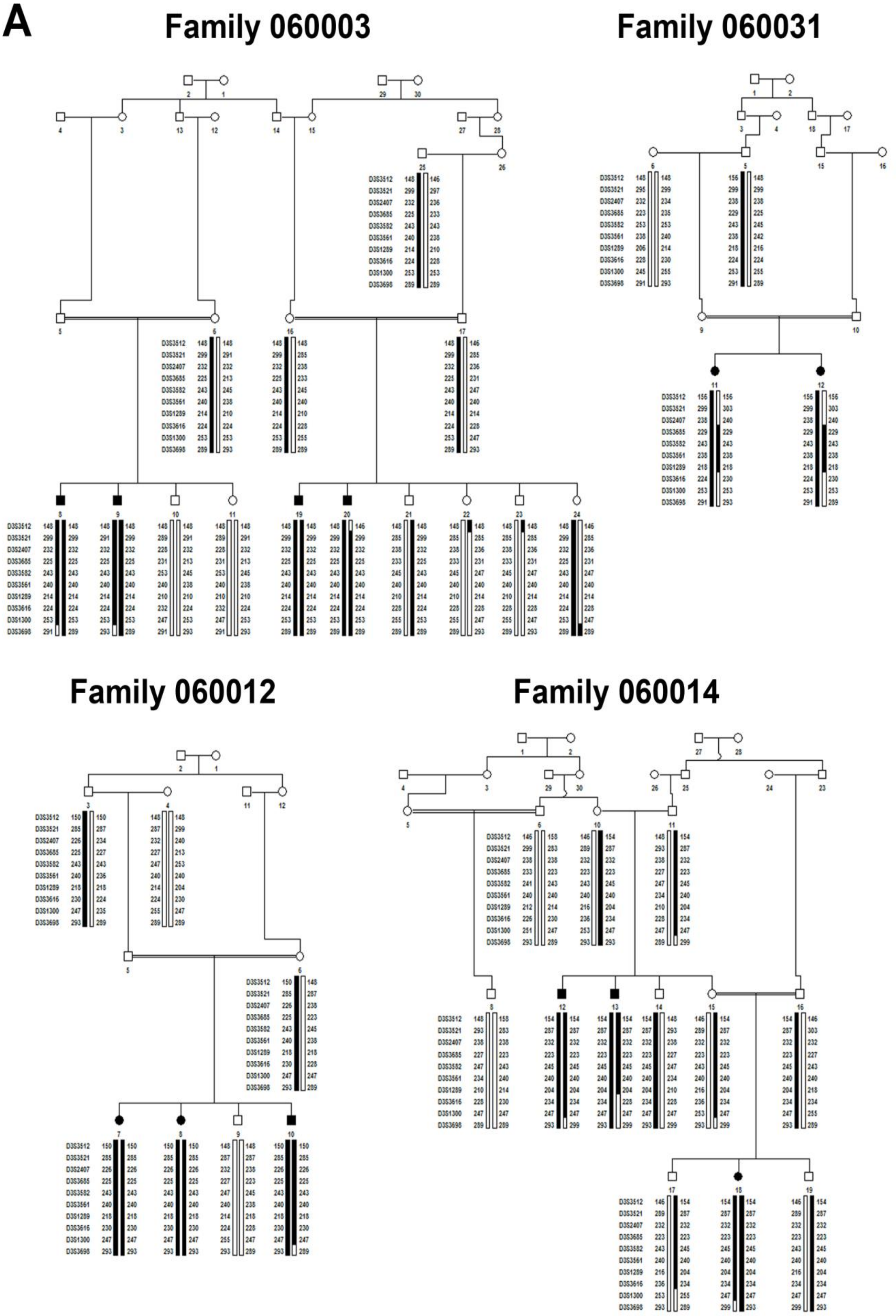
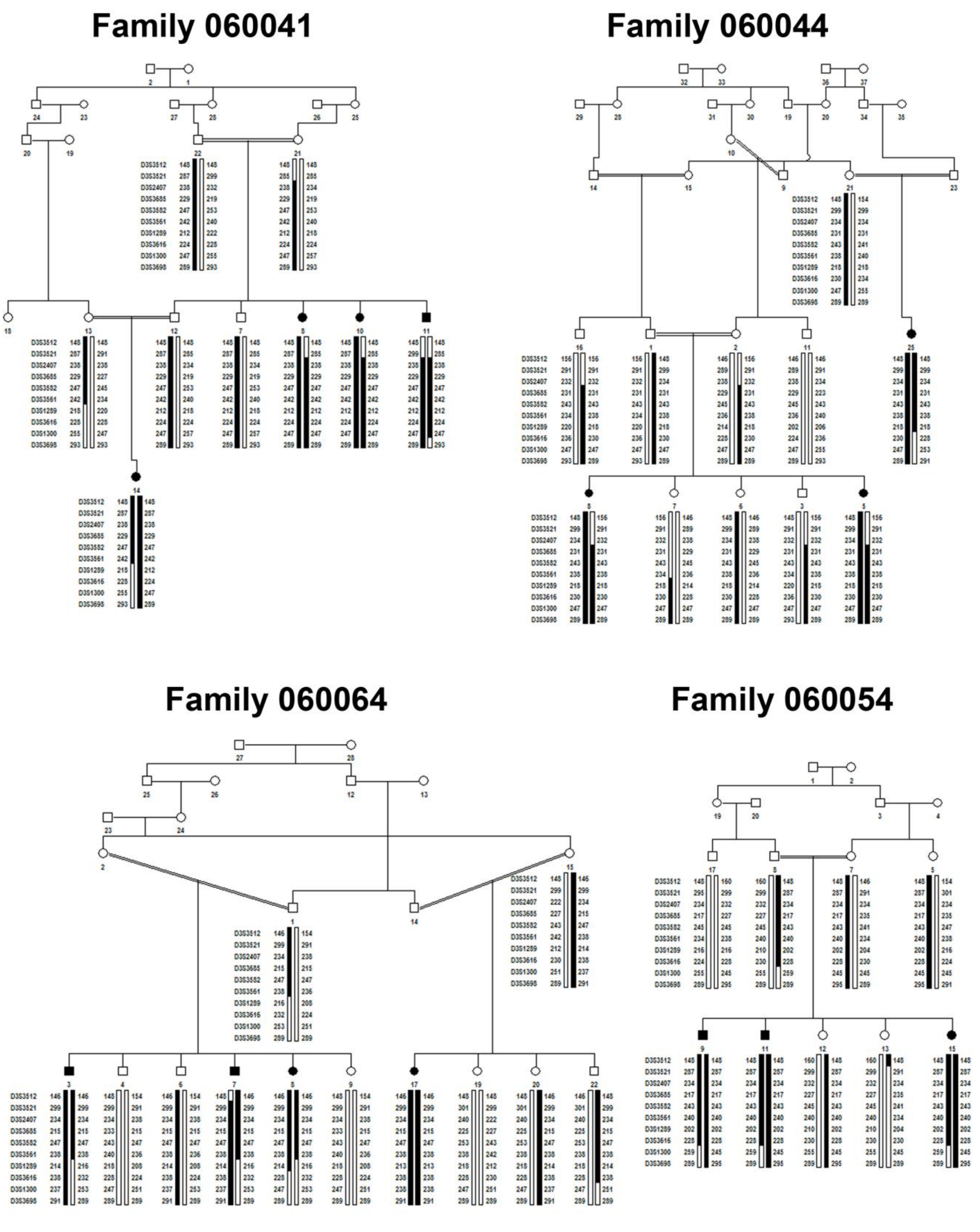

A
Family 060003
Family 060031
Family 060012
Family 060014
D3S3512
D3S3521
D3S2407
D3S3685
D3S3582
D3S3561
D3S1289
D3S3616
D3S1300
D3S3698



B

Family 060041

Family 060044

Family 060064

Family 060054

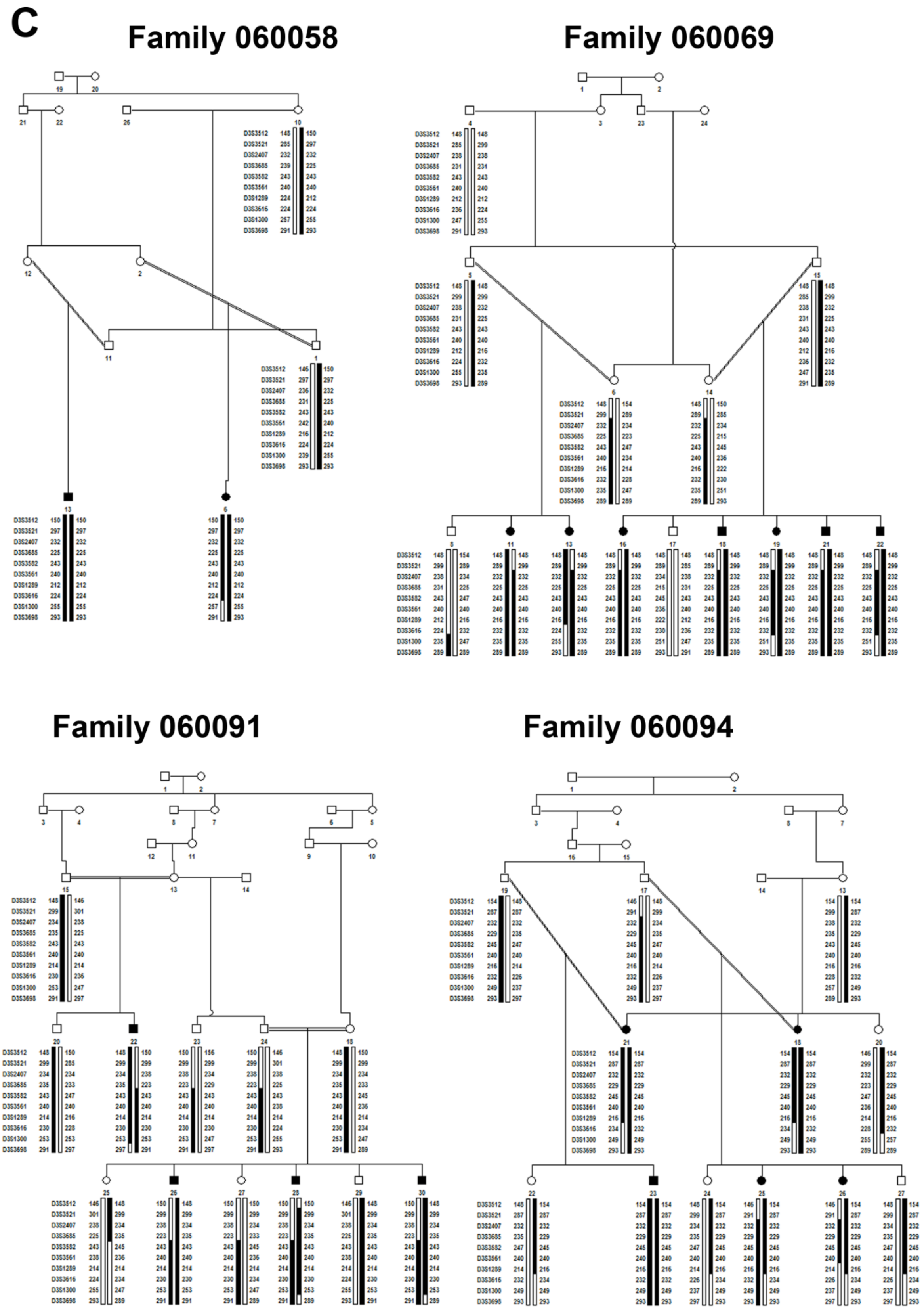

C
Family 060058
Family 060069
Family 060091
Family 060094

Figure S1. Pedigrees with haplotypes for ten consecutive chromosome 3p microsatellite markers. The blackened bars correspond to affected haplotypes with alleles that cosegregate with the disease and that are homozygous in affected individuals.

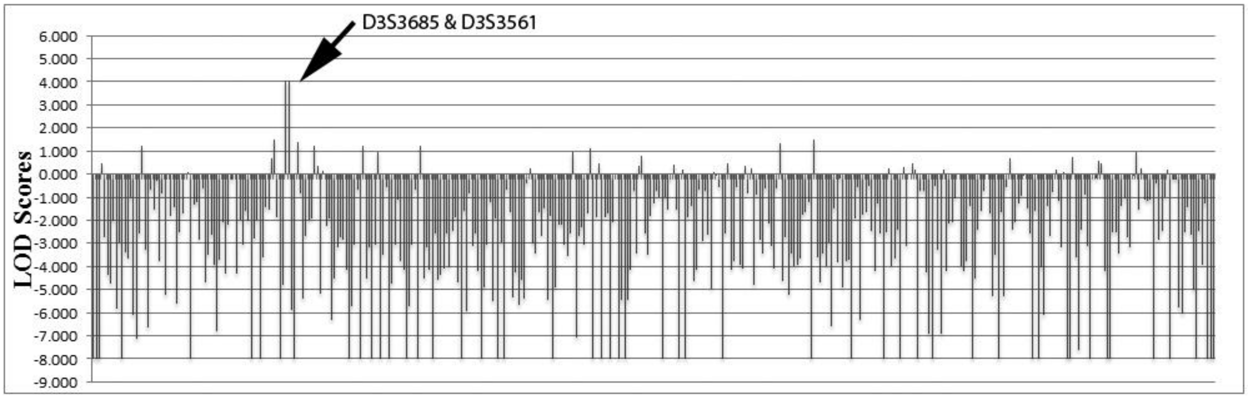


Figure S2. Graphical Illustration of the Two-Point LOD Scores Obtained during the genome-wide scan at θ = 0 for family 60041 (PKCC041). Similar genome wide analyses using STR markers from the md-10 linkage panels (ABI, Foster City, CA) were carried out on all families except for approximately10 that underwent exclusion analysis with closely linked STR markers. Note: A two point LOD scores of -8 represents - ∞.

Table S1: Primer sequences for genotyping of the genetic markers on 3p22-p21. Genotyping primers for 3p22-p21 incorporating a M13 tag in italics.

Marker	Forward (5’>3)	Reverse (5’>3’)	Amplicon Length(bp)
D3S3512	*CACGACGTTGTAAAACGAC*AGATCCCACACCTGCCTCC	ACCAAGTTTTTAAGGCCAATGC	132-162
D3S3521	*CACGACGTTGTAAAACGAC*CAGTAAGTCCATGCCAGGG	GCAGCAGTTACCTTGGAGTG	246-306
D3S2407	*CACGACGTTGTAAAACGAC*TGTACCCTATAGAAGTAAGACCAGG	GGAGTTCAAGGTTACAGTCAGC	223-241
D3S3685	*CACGACGTTGTAAAACGAC*AACAACCCAGCTCTTTGTG	AATACAGGTAACCCTGCCC	196-240
D3S3582	*CACGACGTTGTAAAACGAC*CGATGTGGCTCTGAACTC	AGGGCCTGTTTCCCTAAG	239-255
D3S3561	*CACGACGTTGTAAAACGAC*TCCTGGGGACTGTGATG	GGTGACTGGAGGTTCAAG	204-248
D3S1289	AAAGCAACTTGTAAGAGAGCA	CTCCTAGATATAATCACTGGCA	197-215
D3S3616	*CACGACGTTGTAAAACGAC*TTCAACTCAGAGACACTCAAGTGAC	CCCCTGGGAAGTTACATCAC	223-243
D3S1300	AGCTCACATTCTAGTCAGCCT	GCCAATTCCCCAGATG	217-241
D3S3698	*CACGACGTTGTAAAACGAC*GGTTACCAGGGTCTGGG	GAAGAGATTACCATAATGAAGCC	289-297

Table S2. Primers used for amplification and sequence analysis of human *FYCO1*. All primers were amplified using a touchdown protocol beginning at 64°, decreasing by 2° each two cycles, until finishing at a final annealing temperature of 57°.

Primers for	Forward (5’>3)	Reverse (5’>3’)	Amplicon Length (bp)
FYCO1_EX2	CAGATAGGGTGACCTTTGTGC	GATGTGACCTGAGGACTCCAA	400
FYCO1_EX3	CACTGACCACTTGCTCCTCAT	GGATCACACTTGGAGGATTGA	380
FYCO1_EX4	GTGCTGGCACCTAAGAAGCTA	ACCACCCATAAACTCCCACTT	342
FYCO1_EX5	GGGTAAAATTGGGTGACATTTT	CATTTCTGCACGGGATGA	276
FYCO1_EX6	GAAATTGGTTTGTGTGCCTGT	GACTGGTGGCAATCTTCAAAA	352
FYCO1_EX7	TGGAATCTGTTGGCTTTGACT	GGAACAGCAGCCATAACTGAG	377
FYCO1_EX8-1	GGGCAGGTAATGCCTGTATTT	AGTCCTCCTCCTTCTCTGCTG	479
FYCO1_EX8-2	AGAGAGAACCAGGAGCTGAGG	CTGTCTGTTGACCTCCTCGAC	498
FYCO1_EX8-3	AAACCAGCATTTAGCCAGCTT	CAGAGACCTGACCTCCTGCT	451
FYCO1_EX8-4	GAGTTGTCTGGGAAGGGACA	TCCTTCTCCAGCTCCCTGT	500
FYCO1_EX8-5	GGCAGGTGAGAAGAATGAGG	GAGCCCTAAGCTCTCTGTGC	500
FYCO1_EX8-6	ACTTGGGTGACCAGATGGAG	CTGCTGCACAAGGGTTTTGT	473
FYCO1_EX8-7	CCCACCCACTGACAATGAA	CTCCACTCGCTCCTTTTCC	499
FYCO1_EX8-8	AGCTGGGCATCCAGGTTT	CCTGCACCCACAGTACATTCT	461
FYCO1_EX9	AAAGCTCATTGAGGAGCCAGT	GGGTCAGGGTACAAACTAGGG	380
FYCO1_EX10	GGTTCATGGCCTCTTGAACTT	GGCTTTTCTTGCAAAAGGACT	385
FYCO1_EX11	GCAGTAGCACCCCTCATTAGA	CCTGAGACAGCAGCAAGAAAG	387
FYCO1_EX12	TGGATTCTCTCTCCTTTTCTTCC	GGTGCCAACACCTATCACAGA	289
FYCO1_EX13	TCATGGCCACATCCTAACTTG	GGAAGTGGCACCTAGAGAACA	400
FYCO1_EX14	TAAATTGTGCCTCAGCAGCTT	AATCCTGCTTCCAGTGTCTCA	370
FYCO1_EX15	AAAATCTGCCAGGGTCTCTGT	AGAATAAACGAAGCCCCACTG	379
FYCO1_EX16	GGGCTTGTCCTCTCAAGAGTC	AGAAAGGCCTGCCCAAGTA	397
FYCO1_EX17	GCAACACACTCAGCCTCCTAC	AGAACACTGGAACACCACAGG	370
FYCO1_EX18	CCTCAGGAGAGTCCACAGTGA	CAGGGGCTGAGTTGATGATT	400

Table S3. Allele frequencies in the Pakistani population for chromosome 1 markers used in fine mapping.

Marker	Alleles frequency in the Pakistani population
D3S3512	136(.01), 138(.01), 144(.02), 146(.24), 148(.41), 150(.05), 152(.03), 154(.1), 156(.04), 158(.08), 160(.01)
D3S3521	285(.11), 287(.11), 289(.11), 291(.12), 293(.07), 295(.03), 297(.01), 299(.33), 301(.07), 303(.03), 307(.01)
D3S2407	222(.04), 226(.02), 228(.03), 230(.05), 232(.39), 234(.02), 236(.17), 238(.21), 242(.06), 248(.01)
D3S3685	207(.01), 215(.03), 217(.04), 219(.02), 221(.01), 223(.02), 225(.14), 227(.07), 229(.16),231(.11), 233(.08), 235(.17), 237(.1), 239(.01)
D3S3582	239(.02), 241(.03), 243(.27), 245(.24), 247(.26), 251(.03), 253(.14), 255(.01)
D3S3561	234(.29), 236(.1), 238(.14), 240(.32), 242(.12), 244(.03)
D3S1289	204(.09), 206(.01), 208(.05), 210(.01), 212(.18), 214(.11), 216(.26), 218(.22), 220(.04), 222(.03)
D3S3616	214(.01), 216(.01), 222(.01), 224(.17), 226(.12), 228(.21), 230(.15), 232(.13), 234(.09), 236(.07), 238(.02), 240(.01)
D3S1300	235(.2), 237(.01), 245(.02), 247(.27), 249(.01), 251(.06), 253(.21), 255(.16), 257(.03), 259(.02), 261(.01)
D3S3698	289(.51), 291(.14), 293(.28), 295(.01), 297(.03), 299(.03)