

Supplemental Data

A Mutation in *CABP2*, Expressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment

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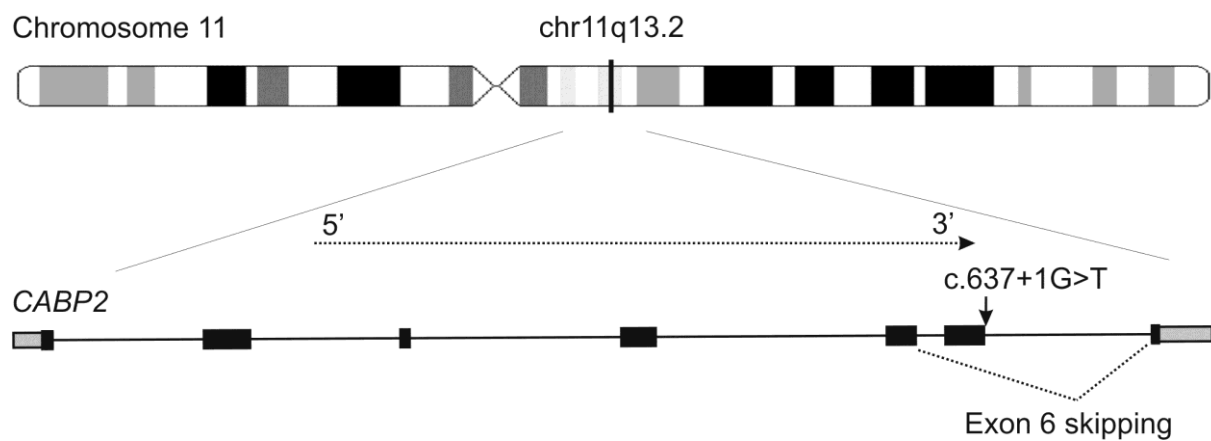


Figure S1. Gene Structure of *CABP2*

The *c.637+1G>T* mutation causes exon 6 skipping.

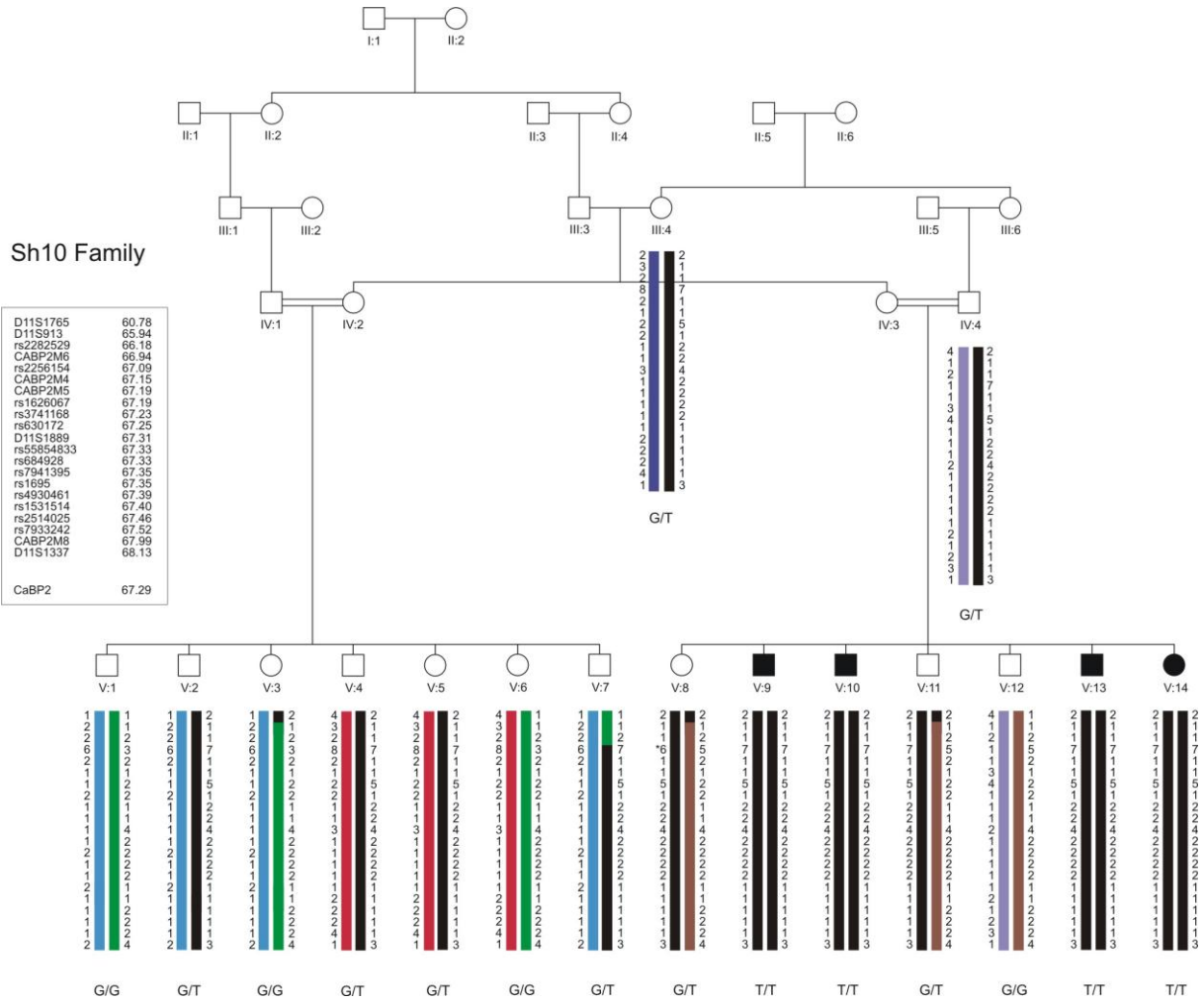


Figure S2. Fine Mapping of the Shared Haplotype Containing the c.637+1G>T Mutation in the Complete Sh10 Family

*Mutation of the microsatellite occurred.

Sh11 Family

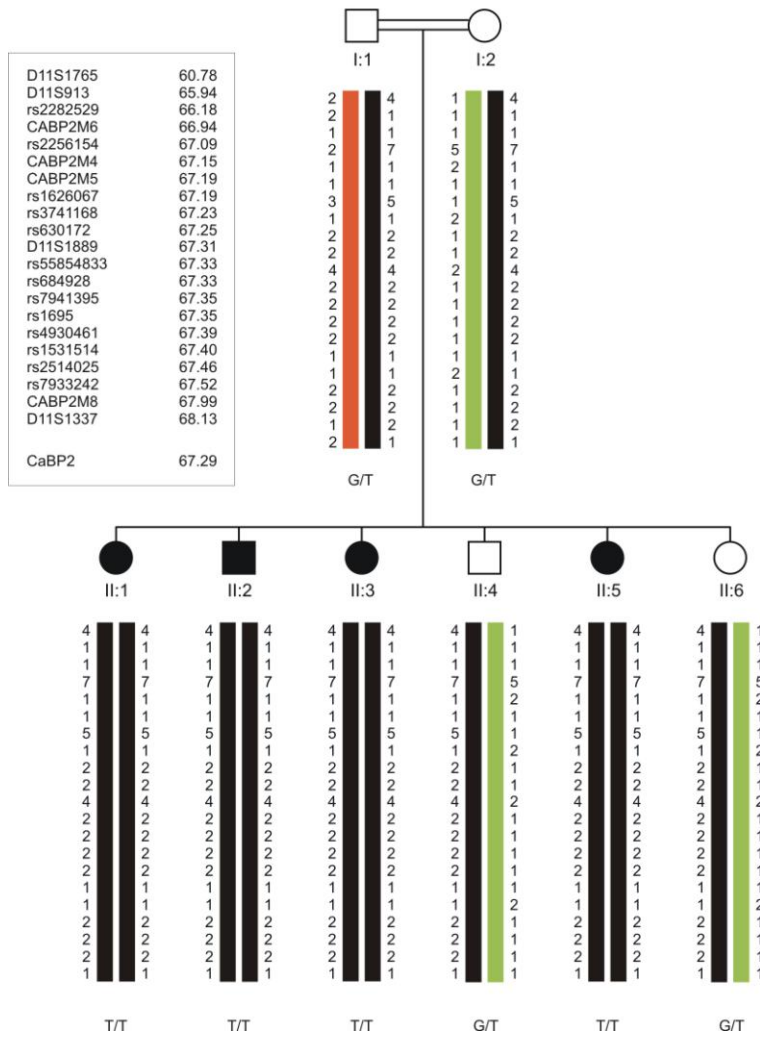


Figure S3. Fine Mapping of the Shared Haplotype Containing the c.637+1G>T Mutation in the Complete Sh11 Family

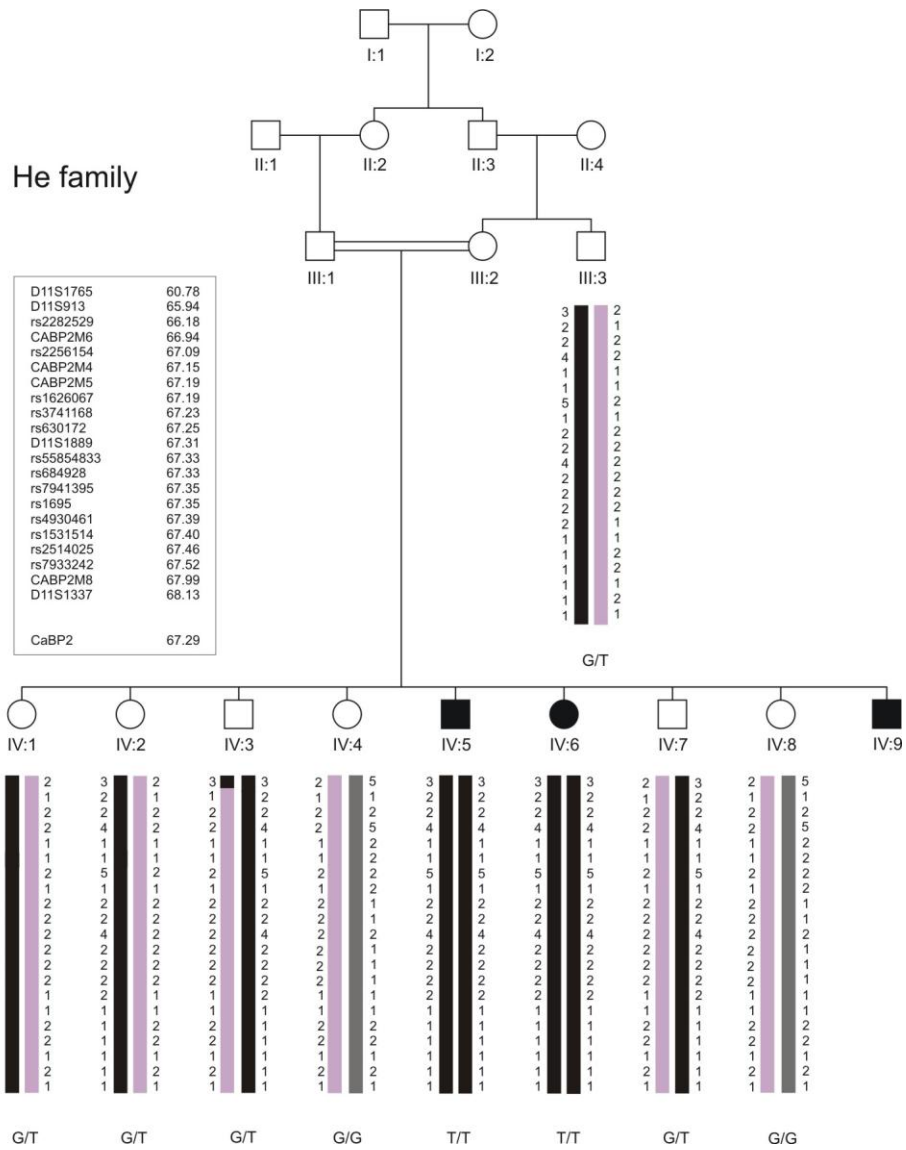


Figure S4. Fine Mapping of the Shared Haplotype Containing the c.637+1G>T Mutation in the Complete He Family

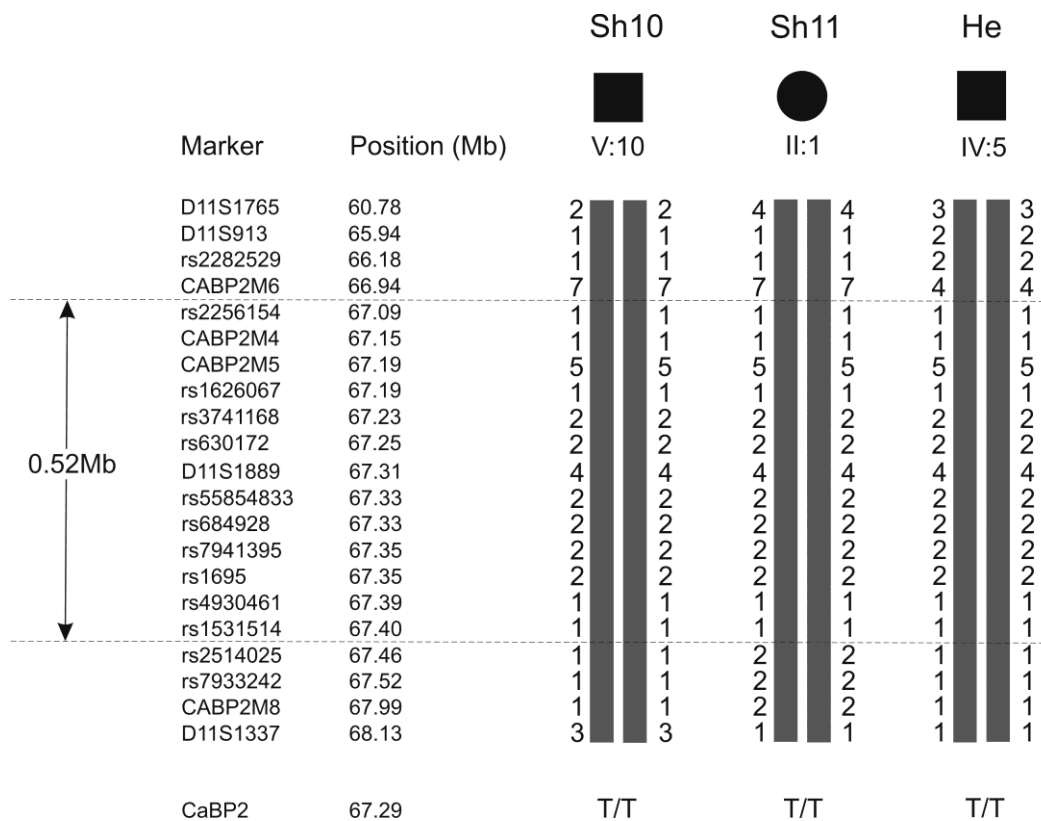


Figure S5. Fine Mapping of the Shared Haplotype Containing the c.637+1G>T Mutations in Sh10, Sh11, and He Families

One affected individual from each family is shown. Recombinations in markers CABP2M6 and rs2514025 reveals a shared ancestral haplotype of 0.52 Mb. Full pedigrees are shown in Figures S1–S3.

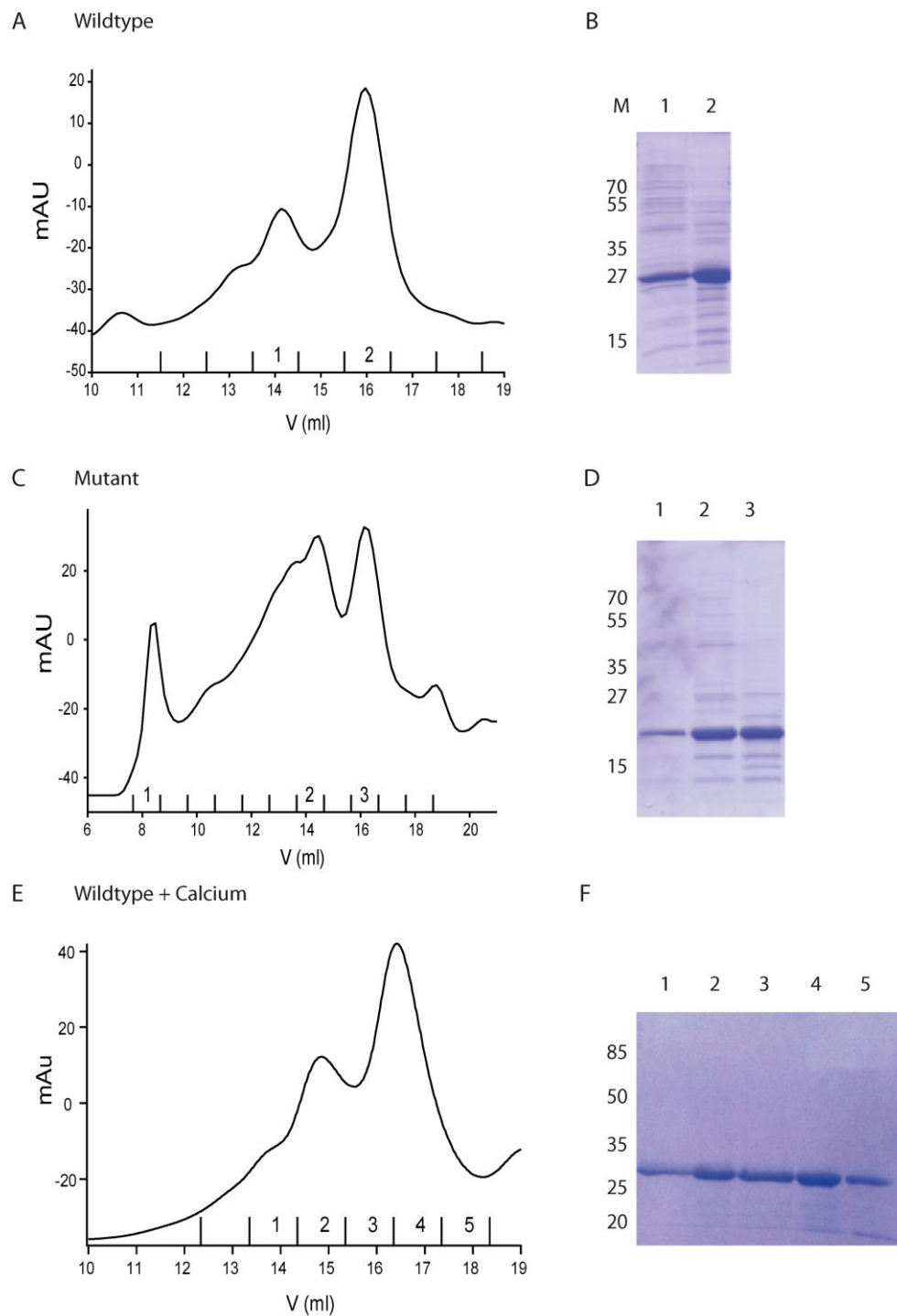


Figure S6. Gel filtration of WT and MT CaBP2

(A) Gel filtration run of wild-type protein. Monomeric protein with the molecular mass of 26 kDa was expected at 16.1 ml elution volume.

(B) Photo of 12.5 % SDS-gel of marked fractions of A.

(C) Gel filtration run of mutant protein. Monomeric protein with the molecular mass of 21 kDa was expected at 16.6 ml elution volume.

(D) Photo of 12.5 % SDS-gel of marked fractions of C.

(E) Gel filtration run of wild-type protein in presence of 1 mM Ca²⁺.

(F) Photo of 12.5 % SDS-gel of marked fractions of E.

Table S1. Primer Sequences

Primer Name	Primer Sequence^a
SD6	5' – TCTGAGTCACCTGGACAACC – 3'
SA2	5' – ATCTCAGTGGTATTTGTGAGC - 3'
dUSD2	5' – CUACUACUACUAGTGAAGTGCCTGTGACAAGCTGC - 3'
dUSA4	5' – CUACUACUACUACACCTGAGGAGTGAATTGGTCG - 3'
CABP201	5' – GAGAGGATCCGGGAAGTGTGCCAAGCGG – 3'
CABP202	5' – TAATACGACTCACTATAGG – 3'
fwd1	5' – GAGAGGATCCGGGAAGTGTGCCAAGCGG – 3'
rev1	5' – CTCCCGGTCGAGT TGGGTGGCGGCAAT - 3'
fwd2	5' – ACCCAACTCGACCGGGAGCTGCGGCC - 3'
rev2	5' - TAATACGACTCACTATAGG – 3'
rev3	5' - ATTGGTGTCCCTACTCCCGGAAGGCGTCCC - 3'
fwd3	5' – TCCGGGAGTA <i>GGACACCAATGGGGACGGC</i> - 3'

^aLetters in italic indicate mutated sites