The American Journal of Human Genetics, Volume *94*

Supplemental Data

Mutations in *CSPP1* Lead to Classical Joubert Syndrome

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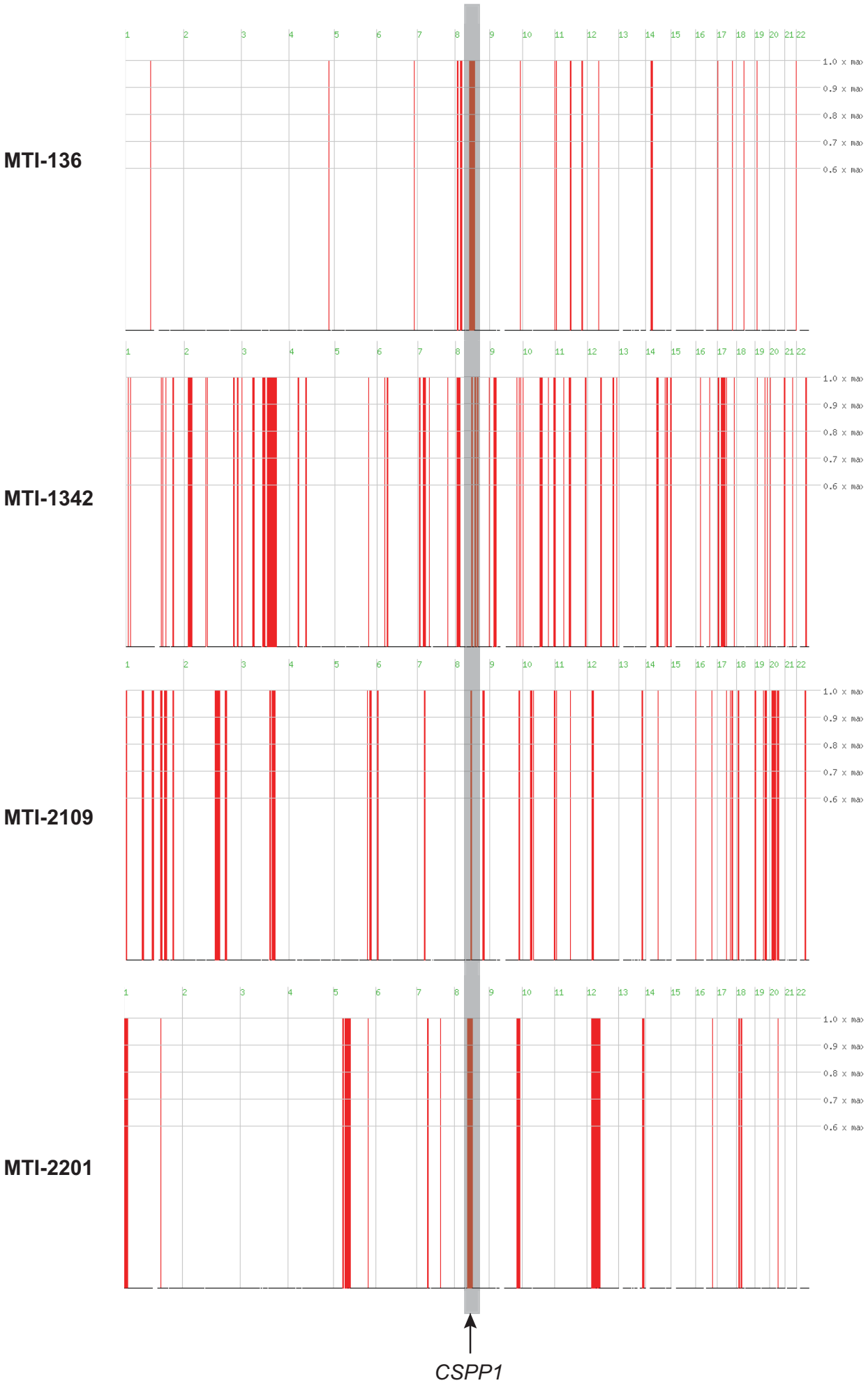
MTI-136
MTI-1342
MTI-2109
MTI-2201
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22
1.0 x ma
0.9 x ma
0.8 x ma
0.7 x ma
0.6 x ma
CSPP1

Figure S1. Homozygosity Map of Affected Individuals

Homozygosity plots showing homozygous blocks (red) in affected individuals from families MTI-136, MTI-1342, MTI-2109, and MTI-2201, all with homozygous *CSPP1* mutations. Gray: homozygous block comprising *CSPP1*, overlaps in all affecteds. Arrow: location of *CSPP1*.

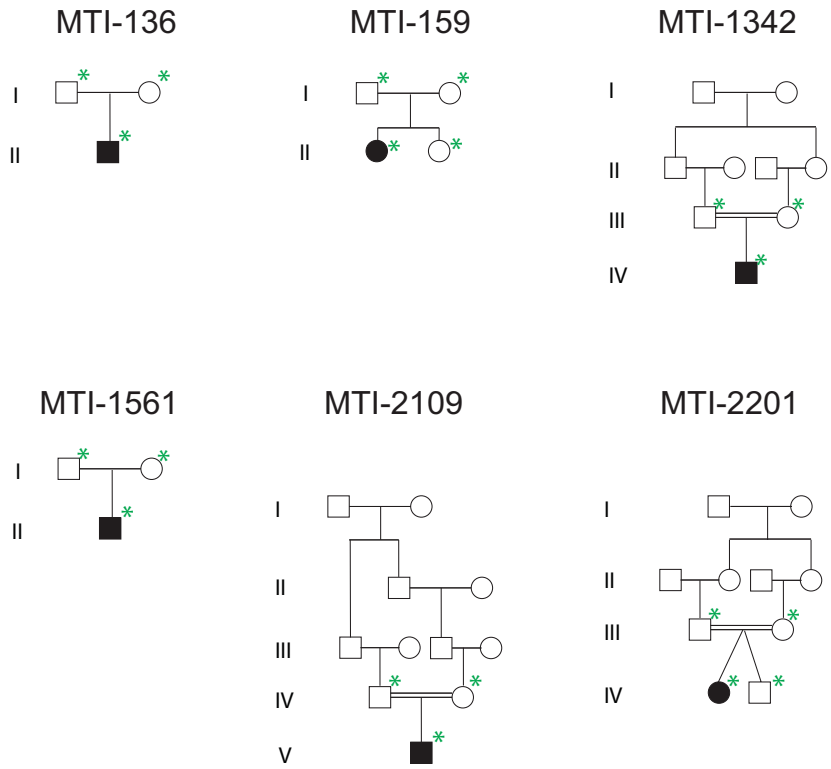

Figure S2. Pedigrees with *CSPP1* Mutated Children with Joubert Syndrome

MTI-1342, MTI-2109, and MTI-2201 are consanguineous families, and MTI-136, MTI-159, and MTI-1561 are nonconsanguineous. Green asterisk indicates individuals Sanger sequenced to test segregation.