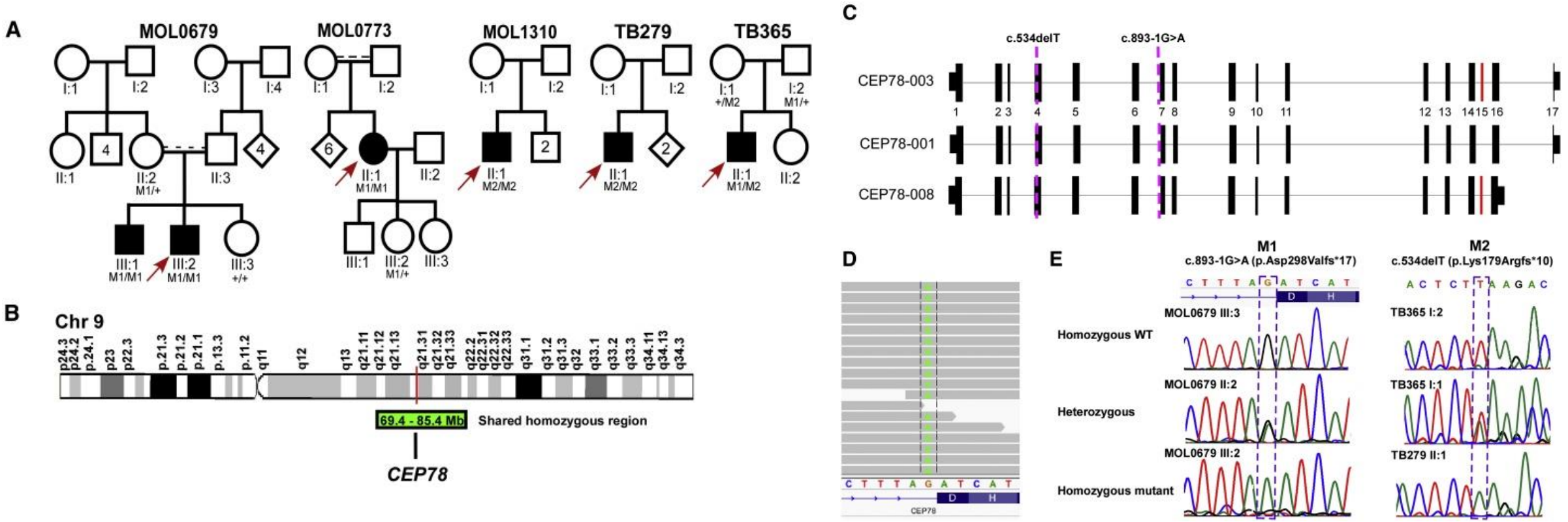

***CEP78* Genomic Arrangement and Mutation Identification**

(A) Pedigree of families with *CEP78* mutation. Filled symbols designate affected individuals. Red arrows represent index case. Family number is depicted above each family tree and individual number below each symbol. The *CEP78* genotype of recruited individuals is depicted below the individual number: M1, c.893-1G>A; M2, c.534delT; +, wild-type allele. Genomic DNA was isolated using Maxwell® 16 Blood DNA Purification Kit (Promega).

(B) The chromosomal region harboring *CEP78* at chromosome 9.

(C) Schematic representation of three different transcripts of *CEP78* and the location of the identified mutations (pink dashed line). Black rectangles represent constitutive exons and red represent an alternatively-spliced exon.

(D) Exome sequence alignment of the c.893-1G>A mutation region in MOL0679 III:1. WES analysis was performed using Nextera Rapid Capture Expanded Exome kit on a HiSeq2500 platform (Illumina). Mapping to the human reference sequence (hg19, GRCH37) was performed using BWA, variants were called using GATK pipeline, and annotation of variants was done with ANNOVAR. Only a representative set of reads is depicted.

(E) Chromatograms of a homozygous wild-type (top), a heterozygous unaffected family member (middle), and a homozygous affected individual (bottom).