

Comprehensive multi-omic profiling of somatic mutations in malformations of cortical development

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Supplementary information

Supplementary Tables

Supplementary Table 1. The cohort list and corresponding sequencing methods. The 293 cases are listed in each row and corresponding sequencing methods used for a given sample were described.

Supplementary Table 2. AmpliSeq primer pool designs. (a) Ampliseq primer pool design used in phase 1. (b) Highlighting table for the known genes where somatic or germline variants are previously detected in FCD/HMEs and the candidate ones known as PI3K-AKT3-mTOR interactors (c) Ampliseq primer pool design used in phase 3.

Supplementary Table 3. Summary of SNV calls across three phases of genetic discovery. (a) 1181 raw sSNV calls derived from the combination of variant callers described in Extended Data Fig. 1b. (b) Validated 108 brain sSNVs from (a). (c) Annotation table of the genes listed in (b) based on GO terms.

Supplementary Table 4. Summary of phenotype and genotype information for the ‘genetically solved’ cases.

Supplementary Table 5. Summary table for snRNAseq DEG and WGCNA analysis. (a) Cell markers are generated by the FindAllMarkers function in Seurat v4 package. (b) 11 MEs and their gene members are generated by WGCNA. (c) DEG analysis results include all cell types or in a cell-type restricted manner. (d) All enriched terms from DEGs.

Supplementary Table 6. Summary table for false discovery estimation.

Supplementary Table 7. Estimation of probability of observing recurrency in mutated genes in MCD.

Supplementary Table 8. Primer sequences used for orthogonal validation and amplifying mutation-carrying DNA fragments.

Supplementary Table 9. Detailed statistical information on Fig.1d, Fig. 3, Extended Data Fig. 5 and 6.