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Malformations of cortical development (MCD) are a group of developmental disorders frequently causing epilepsy. Although next generation sequencing can help identify substantial genetic variants, a correct gene diagnosis of MCD relies on the correlations with neuropathology. I will report the genetic landscape of MCD with refractory epilepsy (RE) whose diagnosis were rmly established in a multidisciplinary epilepsy team at Taipe Veterans General Hospital in Taiwan. Sixty-six patients were recruited. eir MCD types include: FCD (51, 77.3%), heterotopia (4, 6.06%), polymicrogyri, Dandy-Walker malformation and lissencephaly. Tuberous sclerosis comple was not included. ese patients were rst screened by targeted sequencing (TS) of 66 genes causative for MC and epilepsy encephalopathy. For those with a potential candidate variant identi ed, they were submitted to who exome sequencing to con rm the variant is the best pathogenic candidate. Reported pathogenic variant or nov but potentially disease-causative variants were identi ed in 28 patients (42%). Among them, nine were familia cases (32%). In the 38 genetic not-assigned individuals, only two had a positive family history (5.3%). Nine varia (32/1%) occurred in the GATOR1 complex genes (DEPDC5/NPRL2/NPRL3). e hit rate was the highest, reaching 78% (7/9), in severe and di use MCD, like Dandy-Walker malformation and lissencephaly. For FCD, the hit rate was 55% (28/51). Our results supported that rapid screening by tTS of known disease-causative genes is e cien enhance genetic diagnosis of MCD, particularly in severe and di use MCD and FCD. Brain MRI and neuropatholog are essential to determine the pathogenicity of identi ed variants.

% L R J U D S K \

Yo-Tsen Liu earned her MD at National Taiwan University, Taiwan and completed her neurological residency training and became a neurology consultant at Taipei Veterans General Hospital (TVGH). After winning “Studying Abroad Scholarship” supported by Taiwan’s Ministry of Education, she studied her PhD at Institute of Neurology, University College London, London, UK in 2010~2014. She is now a neurology consultant at Division of Epilepsy, Neurological Institute, TVGH and Assistant professor at Faculty of Medicine and Institute of Brain Science, National Yang-Ming University, Taiwan. Her research interests are the applications of next-generation sequencing in neurological diseases, focusing on epilepsy and movement disorders.

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