## Genomic Variants and Variations in Malformations of Cortical Development



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## **KEYWORDS**

- Malformations of cortical development Genomic variants Somatic mutation
- Microcephaly Megalencephaly Cortical dysplasia Lissencephaly
- Polymicrogyria

## **KEY POINTS**

- Development of the cerebral cortex is a tightly regulated process, and disruption in any part of this process can lead to malformations of cortical development (MCDs).
- MCD can primarily be classified into abnormalities of neurogenesis, abnormalities of neuronal migration, and abnormalities of postmigrational development.
- Recent advances in genomic technology have allowed for an unprecedented expansion in the knowledge of these disorders and have elucidated molecular pathways that can serve as targets for therapeutic interventions.

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