The genetic basis of the disorder is mutations in the TSC1 or TSC2 gene, which leads to overactivation of the mammalian target of rapamycin (mTOR) protein complex and results in the development of benign tumors in different body systems such as the brain, skin, lungs, and kidney.

Autosomal dominant inheritance; however, spontaneous mutation accounts for the majority of cases.

Focal cortical dysplasia (FCD) is a localized cortical malformation and considerable morphological overlap exists between Focal cortical dysplasia type II B (FCD IIB) and neurological lesions associated with Tuberous sclerosis complex (TSC). Abnormal mTOR pathway secondary to somatic mTOR mutation and TSC gene mutation linked to PI3K/AKT/mTOR pathway have supported the hypothesis of common pathogenesis involved.

Two distinct tumor suppressor genes have been identified: the TSC1 gene (located on chromosome 9q34) codes for TSC1 (AKA hamartin), and the TSC2 gene (on chromosome 16p13.3) codes for TSC2 (tuberin). Only 1 gene needs to be affected to develop TSC. These proteins work together to inhibit the activation of rapamycin (mTOR). Genetic counseling for unaffected parents with one affected child: 1–2% chance of recurrence [1) 2)].

1)
European Chromosome 16 Tuberous Sclerosis Consortium. Identification and characterization of the tuberous sclerosis gene on chromosome 16. Cell. 1993;75:1305–1315.
2)
van Slegtenhorst M, deHoogt R, Hermans C, et al. Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. Science. 1997;277:805–808.

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