Somatic mosaicism and variable penetrance in doublecortin-associated migration disorders


X-linked isolated lissencephaly sequence (XLIS) and subcortical band heterotopia (SBH) are allelic disorders caused by mutations in the doublecortin (DCX) gene. This genetic analysis of seven families revealed four novel mutations in the DCX gene. The authors detected a high rate of somatic mosaicism in male and female patients with variable penetrance of bilateral SBH including nonpenetrance in a heterozygous woman. In addition, the authors implemented prenatal diagnosis in a family with SBH/XLIS.

**Type:** journal paper/review (English)

**Date of Publishing:** 28-1-2003

**Journal Title:** Neurology (60/2)

**ISSN Electronic:** 1526-632X

**Pages:** 329-32