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Translation defects

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Analysis of patients with an inherited brain disease has provided the first link between translation initiation factors and human disease. Leukoencephalopathy with vanishing white matter (VWM) occurs during childhood and is a chronic-progressive disorder (involving rapid additional deterioration following febrile infection or minor head trauma). In the Advanced Online Publication of Nature Genetics, Leegwater *et al.* report mutations in VWM patients within two subunits of the eIF2B translation initiation factor (DOI: 10.1038/ng764). They identified a pedigree of nine families with a common haplotype (EN) to narrow down the hunt to a region of chromosome 3q27 containing around 25 genes. Genomic sequence analysis identified mutations in EIF2B5, encoding the ε subunit of eIF2B, a eukaryotic translation initiation factor. Analysis of genomic loci encoding other eIF2B subunits led the researchers to discover additional mutations in VWM families without linkage to chromosome 3q27, and these were due to mutations in the *EIF2B2* gene on chromosome 14q24, encoding the eIF2B β-subunit. The activity of eIF2B is regulated by stress, and the authors suggest that the function of eIF2B in the heat-shock response might account for the neurological deterioration of VWM patients following fever conditions.

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