The epidemiology of CuZn-SOD mutations in Germany: a study of 217 families

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We screened 217 patients from Germany (n = 213), Austria (n = 2) and Switzerland (n = 2) with a positive family history for amyotrophic lateral sclerosis (ALS) for mutations in the copper/zinc superoxide dismutase (SOD1) gene. We found that 13% of the families tested carried mutations. By analyzing inheritance, we detected a clear-cut co-segregation in 5 of the 28 families; however, in two families with an established mutation, co-segregation was absent. In Germany, the R115G mutation is comparatively frequent and exhibits a specific aggressive phenotype. The L144F mutation, which is the most prevalent mutation in the Balkan countries, and the D90A mutation which is the most frequent SOD1 mutation globally, seem to be the second most common disease-causing mutations in Germany.