Summary of findings in more than 4200 individuals with TSC and their families in whom disease-causing mutations have been identified

- Jones et al [1999] identified exonic and whole-gene deletions in \textit{TSC1} and \textit{TSC2} and small mutations in 120 of 150 (80\%) individuals with TSC, of whom 130 represented simplex cases (i.e., individuals who have no family history of TSC) and 20 were familial cases.

- In a study of 38 familial cases, 183 simplex cases, and three of unknown status, Dabora et al [2001] identified small mutations in either \textit{TSC1} or \textit{TSC2} in 166 (74\%) probands.

- Using mutation scanning and direct sequencing, Southern blotting, and FISH analysis in 490 families with TSC, Sancak et al [2005] identified small mutations in either \textit{TSC1} or \textit{TSC2} in 342 (70\%).

- Using mutation scanning and direct sequencing to screen for mutations in 325 families who met diagnostic criteria for TSC, Au et al [2007] identified 243 (75\%) who had small mutations in either \textit{TSC1} or \textit{TSC2}.

References


